

**This Report
reflects the law
as at 14 March 2003**

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The Hon Daryl Williams AM QC MP
Attorney-General

Senator the Hon Kay Patterson
Minister for Health and Ageing

28 March 2003

Dear Attorney-General and Minister,

The Protection of Human Genetic Information

On 5 February 2001, the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee of the National Health and Medical Research Council (AHEC) received a reference, pursuant to the *Australian Law Reform Commission Act 1996* and the *National Health and Medical Research Act 1992*, respectively, to undertake an Inquiry into matters relating to the protection of human genetic information.

Those terms of reference were amended by your agreement, as stated in a letter from the Attorney-General to the ALRC dated 25 January 2002, to change the reporting date for the Joint Inquiry to 31 March 2003.

On behalf of the ALRC Commissioners and AHEC Members involved in this reference, we are pleased to present the final report of the Joint Inquiry: *ALRC 96 Essentially Yours: The Protection of Human Genetic Information in Australia*. Owing to the breadth of the subject matter, and the consequent length, this report is presented in two volumes.

Yours sincerely,

Professor David Weisbrot
President, ALRC

Dr Kerry Breen
Chair, AHEC

Associate Professor Brian Opeskin
Commissioner, ALRC

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Terms of Reference

COMMONWEALTH OF AUSTRALIA

Australian Law Reform Commission Act 1996

National Health and Medical Research Act 1992

PROTECTION OF HUMAN GENETIC INFORMATION

1. We, DARYL WILLIAMS, Attorney-General of Australia, and MICHAEL WOOLDRIDGE, Minister for Health and Aged Care, having regard to—

- the rapid advances in human genetic technology; and
- the scientific and medical applications of human genetic information which are, or could be, of benefit to the Australian community; and
- ethical concerns in relation to the collection, storage and use of human genetic samples and information; and
- the potential for inappropriate use or application of human genetic samples and information; and
- evidence of, and the potential for, use of human genetic information by a number of sectors including employment; health, including medical research, pharmaceuticals and health administration; insurance and superannuation; intellectual property; and law enforcement; and
- emerging issues about the control of, ownership of, and intellectual property rights in relation to human genetic samples and information;

refer to the Australian Law Reform Commission and the Australian Health Ethics Committee of the National Health and Medical Research Council for inquiry and report pursuant to subsection 20 (1) of the *Australian Law Reform Commission Act 1996* and paragraph 35 (3) (c) of the *National Health and Medical Research Act 1992* respectively, matters relating to —

- (a) whether, and to what extent, a regulatory framework is required—
 - (i) to protect the privacy of human genetic samples and information; and

- (ii) to provide protection from inappropriate discriminatory use of human genetic samples and information; and
 - (iii) to reflect the balance of ethical considerations relevant to the collection and uses of human genetic samples and information in Australia; and
 - (b) any related matter.
2. In performing their functions in relation to this reference, the Commission and the Australian Health Ethics Committee shall—
- (a) conduct this inquiry jointly; and
 - (b) identify and consult with relevant stakeholders, including the Privacy Commissioner and the Human Rights and Equal Opportunity Commission, and ensure widespread public consultation; and
 - (c) have regard to the following matters —
 - (i) the rapid advances in human genetic technology including progress of research towards the mapping of the human genome; and
 - (ii) the scientific and medical applications of human genetic information which are, or could be, of benefit to the Australian community; and
 - (iii) evidence of, and the potential for, the inappropriate use or application of human genetic information; and
 - (iv) the range of Australian ethical opinion as to which, if any, uses and applications of human genetic information are ethically acceptable; and
 - (v) the global dimensions of issues relating to research, regulation and the protection of interests; and
 - (vi) any relevant existing or proposed international law and obligations; and
 - (vii) any relevant constitutional issues; and
 - (viii) any relevant existing or proposed Commonwealth legislation; and
 - (ix) the implications of the recent decision by Australian health ministers to develop a national health information network; and

-
- (x) developments in other jurisdictions, including legislative and other regulatory action; and
 - (xi) relevant research and discussion of human genetic information privacy and discrimination issues.

3. The Commission and the Australian Health Ethics Committee are to report to the Attorney-General and the Minister for Health and Aged Care by 30 June 2002.*

Dated 5 February 2001

Daryl Williams
ATTORNEY-GENERAL

Michael Wooldridge
MINISTER FOR HEALTH AND AGED CARE

* In his letter of 25 January 2002, the Attorney-General stated that he and the Minister for Health and Ageing, the Hon Kay Patterson agreed to extend the deadline for the final report under this reference to 31 March 2003.

Participants

Australian Law Reform Commission

The Division of the ALRC constituted under the *Australian Law Reform Commission Act 1996* (Cth) for the purposes of this reference comprises the following:

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Professor David Weisbrot

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Associate Professor Brian Opeskin (Commissioner)

Mr Ian Davis (Commissioner)

Professor Anne Finlay (Commissioner)

Justice Ian Coleman (part-time Commissioner) (to December 2002)

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Professor Don Chalmers, Dean of Law, University of Tasmania, and immediate past
Chair of AHEC

Mr Malcolm Crompton, Federal Privacy Commissioner

The Hon Justice Arthur Emmett, Federal Court of Australia
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Dr Trevor Mudge, Vice President, Australian Medical Association
Professor Marcia Neave, Chair, Victorian Law Reform Commission
Mr Chris Puplick, President, Anti-Discrimination Board of NSW & NSW Privacy Commissioner
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Ms Jane-Ann Jones (June to August 2002)

Ms Kaye Sperling (February 2001 to April 2001)

Ms Helen Willimott (April 2001 to February 2002)

List of Submissions

Name	<i>Submission no</i>
Aboriginal and Torres Strait Islander Social Justice Commissioner— Human Rights and Equal Opportunity Commission	G160
Ms Deonna Ackermann and others	G220
ACROD Limited	G239
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Confidential	G074CON
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Confidential	G100CON
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Abbreviations

AAA	American Anthropological Association
ABA	Australian Broadcasting Authority
ABI	Association of British Insurers
ACCC	Australian Consumer and Competition Commission
ACCI	Australian Chamber of Commerce and Industry
ACGT	Advisory Committee on Genetic Testing (UK)
ACPO	Association of Chief Police Officers (UK)
ACT	Australian Capital Territory
ACTU	Australian Council of Trade Unions
ADF	Australian Defence Force
AFL	Australian Football League
AFP	Australian Federal Police
AFS	Australian Financial Services
AGSA	Association of Genetic Support of Australasia
AHEC	Australian Health Ethics Committee
AHMAC	Australian Health Ministers' Advisory Council
AIATSIS	Australian Institute of Aboriginal and Torres Strait Islander Studies
AIS	Australian Institute of Sport
AIS Support Group	Androgen Insensitivity Syndrome Support Group
ALRC	Australian Law Reform Commission
AMA	Australian Medical Association
AMC	Australian Medical Council
AMWU	Australian Manufacturing Workers' Union
ANTA	Australian National Training Authority
APMC	Australasian Police Ministers' Council
APRA	Australian Prudential Regulation Authority
ARC	Australian Research Council
ARTG	Australian Register of Therapeutic Goods
ASC	Australian Sports Commission
ASHG	American Society of Human Genetics
ASIC	Australian Securities and Investments Commission
ATSIC	Aboriginal and Torres Strait Islander Commission
ATSIC Act	<i>Aboriginal and Torres Strait Islander Commission Act 1989</i> (Cth)
AUD	Australian dollars
CASA	Civil Aviation Safety Authority Australia
CCRC	Criminal Cases Review Commission (UK)
CDAMS	Committee of Deans of Australian Medical Schools
CD-ROM	Compact disc—read only memory
CHOICE	Australian Consumers' Association
CF	Cystic Fibrosis

CJD	Creutzfeldt-Jakob disease
CMT	Charcot-Marie Tooth disease
COAG	Council of Australian Governments
CODIS	Combined DNA Index System (US)
CPMC	Committee of Presidents of Medical Colleges
CRC	Cooperative Research Centre
<i>Crimes Act</i>	<i>Crimes Act 1914 (Cth)</i>
CSAA	<i>Child Support (Assessment) Act 1989 (Cth)</i>
CSIRO	Commonwealth Scientific and Industrial Research Organisation
CTE	Chronic traumatic encephalopathy
Cth	Commonwealth
DDA	<i>Disability Discrimination Act 1992 (Cth)</i>
DDLS	Disability Discrimination Legal Service
DIMIA	Department of Immigration and Multicultural and Indigenous Affairs
DMD	Duchenne Muscular Dystrophy
DNA	Deoxyribonucleic acid
DP 66	Discussion Paper 66, <i>Protection of Human Genetic Information</i>
DVI Database	Disaster Victim Identification Database
EDR	External dispute resolution
EEOC	Equal Employment Opportunity Commission (US)
EINSHAC	Einstein Institute for Science, Health and the Courts (US)
ELSI	Ethical, legal and social implications
ESHG	European Society for Human Genetics
<i>Evidence Act</i>	<i>Evidence Act 1995 (Cth)</i>
FAP	Familial Adenomatous Polyposis
FBI	Federal Bureau Investigation (US)
FICS	Financial Industry Complaints Service
FL Regulations	<i>Family Law Regulations 1984 (Cth)</i>
FLA	<i>Family Law Act 1975 (Cth)</i>
FLC	Family Law Council
FSRA	<i>Financial Services Reform Act 2001 (Cth)</i>
FSS	Forensic Science Service (UK)
GAIC	Genetics and Insurance Committee (UK)
GPA	Genetic Privacy Act (US)
GTRAP	Gene and Related Therapies Research Advisory Panel
HCM	Hypertrophic cardiomyopathy
HD	Huntington's disease
HGC	Human Genetics Commission (UK)
HGCA	Human Genetics Commission of Australia
HGSA	Human Genetics Society of Australasia
HIV	Human Immunodeficiency Virus
HREC	Human Research Ethics Committee
HREOC	Human Rights and Equal Opportunity Commission

HREOC Act	<i>Human Rights and Equal Opportunity Commission Act 1984 (Cth)</i>
HREOC Regulations	<i>Human Rights and Equal Opportunity Commission Regulations 1989 (Cth)</i>
HUGO	Human Genome Organisation
HWSCA	Heads of Workplace Safety and Compensation Authorities
IARC	Immigration Advice and Rights Centre
ICA	Insurance Council of Australia
IDR	Internal dispute resolution
IEC	Insurance Enquiries and Complaints Ltd
IFSA	Investment and Financial Services Association
IP 26	Issues Paper 26, <i>Protection of Human Genetic Information</i>
IPP	Information Privacy Principle
IVD	In-vitro diagnostic device
LR	likelihood ratio
MBS	Medical Benefits Scheme
MCCOC	Model Criminal Code Officers Committee
<i>Migration Act</i>	<i>Migration Act 1958 (Cth)</i>
<i>Migration Regulations</i>	<i>Migration Regulations 1994 (Cth)</i>
MOC	Medical Officer of the Commonwealth
MOU	Memorandum of Understanding
MSAC	Medical Services Advisory Committee
mtDNA	Mitochondrial DNA
NATA	National Association of Testing Authorities, Australia
National Statement	NHMRC's <i>National Statement on Ethical Conduct in Research Involving Humans</i>
NBAC	National Bioethics Advisory Commission (US)
NCEHR	National Council on Ethics in Human Research (Canada)
NCIDD system	National Criminal Investigation DNA Database system
NFITAB	National Finance Industry Training Advisory Body
NHMRC	National Health and Medical Research Council
NHMRC Act	<i>National Health and Medical Research Council Act 1992 (Cth)</i>
NHS	National Health Service (UK)
NIFS	National Institute of Forensic Science
NOHSC	National Occupational Health and Safety Commission
NPAAC	National Pathology Accreditation Advisory Council
NPAP	National Priority Action Plan
NPP	National Privacy Principle
NRCOHSR	National Research Centre for Occupational Health and Safety Regulation
NSC	National Screening Committee (UK)
NSW	New South Wales
NSWCCL	New South Wales Council for Civil Liberties
NT	Northern Territory

OECD	Organization for Economic Cooperation and Development
OFPC	Office of the Federal Privacy Commissioner
OGTR	Office of the Gene Technology Regulator
PCR	Polymerase Chain Reaction
PFA	Police Federation of Australia
PID	Public Interest Determination
PIN	Personal Identification Number
PKU	Phenylketonuria
PMCI	Peter McCallum Cancer Institute
<i>Privacy Act</i>	<i>Privacy Act 1988 (Cth)</i>
QIMR	Queensland Institute of Medical Research
Qld	Queensland
RAAF	Royal Australian Air Force
RACGP	Royal Australian College of General Practitioners
RACP	Royal Australian College of Physicians
RCPA	Royal College of Pathologists of Australasia
RDA	<i>Racial Discrimination Act 1975 (Cth)</i>
REC	Research Ethics Committee
RNA	Ribonucleic Acid
SA	South Australia
SACGT	Secretary's Advisory Committee on Genetic Testing (US)
SDA	<i>Sex Discrimination Act 1984 (Cth)</i>
SIDS	Sudden Infant Death Syndrome
SIFS	State Institute of Forensic Sciences (NSW)
SMA	Sports Medicine Australia
SNPs	Single nucleotide polymorphisms
STRs	Short tandem repeats
Tas	Tasmania
TB	Tuberculosis
TGA	Therapeutic Goods Administration
TSD	Tay-Sachs disease
UAG	CrimTrac User Advisory Group
UNESCO	United Nations Educational Scientific and Cultural Organisation
Vic	Victoria
WA	Western Australia
WHO	World Health Organisation
WRA	<i>Workplace Relations Act 1996 (Cth)</i>
XL	X-linked

Executive Summary

Essentially Yours

Essentially Yours: The Protection of Human Genetic Information in Australia (ALRC 96, 2003) represents the culmination of a major, two-year inquiry by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) of the National Health and Medical Research Council (NHMRC). The Report, which contains 144 recommendations for reform, is the product of an extensive research and community consultation effort—the most comprehensive consideration of the ethical, legal and social implications of the ‘New Genetics’ ever undertaken.

The Terms of Reference directed the ALRC and AHEC to consider, with respect to human genetic information and the samples from which such information is derived, how best to:

- protect privacy;
- protect against unfair discrimination; and
- ensure the highest ethical standards in research and practice.

From the beginning, the Inquiry recognised the need for public engagement and widespread consultation, involving the general community as well as experts and interest groups. To this end, the Inquiry released an Issues Paper (IP 26, October 2001) and a Discussion Paper (DP 66, August 2002) to promote public education and debate, conducted 15 open forums around Australia, initiated over 200 meetings with interested parties in Australia and overseas, and received over 300 written submissions.

The ‘New Genetics’ is remarkable for the speed of scientific and technological development. In 50 years, we have moved from the first published description of the DNA double helix by Watson and Crick, to the effective mapping of the entire human genome by the Human Genome Project consortium.

The experience of the Inquiry, mirrored overseas, is that the rapid pace of change has produced two powerful, but conflicting, social reactions. On the one hand, there is very strong public support for breakthroughs promising better medical diagnosis and treatments, and for assisting with law enforcement (including identification of missing or deceased persons); on the other, there are anxieties about increased loss of privacy and the potential for genetic discrimination, as well as about the capacity to regulate genetic science in the public interest.

The major challenge for the Inquiry was to find a sensible path that meets twin goals: to foster innovations in genetic research and practice that serve humanitarian ends, and to provide sufficient reassurance to the community that such innovations will be subject to proper ethical scrutiny and legal (and other) controls.

The current methods of regulation and conflict resolution involve a patchwork of federal, state and territory laws; official guidelines; personal and professional ethics; institutional restraints; peer review and pressure; oversight by public funding authorities and professional associations; supervision by public regulatory and complaints-handling authorities; private interest; and market pressures.

The Inquiry's brief was to scrutinise the existing regimes, and then tailor them—if necessary and to the extent possible—to the particular needs and demands of genetic testing and information. In some instances, the Inquiry has recommended new forms of regulation to address existing gaps. Successfully fulfilling this brief has involved not only providing adequate protections against the *unlawful* use of genetic information, but also putting into place measures and strategies aimed at ensuring that where such information may be used lawfully, it will be used properly, fairly and intelligently.

Achieving justice in this complex area is not susceptible to a simple vindication of individual rights. Careful consideration of the legal and policy issues thrown up by the use of genetic samples and information requires a wide range of interests to be balanced. Although relatively easy to articulate in the abstract, achieving the proper balance is difficult in practice, since various interests will compete and clash across the spectrum of activity.

For example, as discussed throughout this Report, human genetic information has a strong familial dimension—an individual's genetic information will usually reveal information about, and have implications for, his or her parents, grandparents, siblings, children, and generations to come. Thus, there may be circumstances in which an individual's presumptive right to privacy, and to the confidentiality of the doctor-patient relationship, may be called into question by the competing needs of genetic relatives.

Similarly, a balance must be struck in a number of other areas in such a way as to recognise and accommodate broad social interests rather than individual ones—such as in the compulsory acquisition of DNA samples by law enforcement authorities; the ability of researchers to gain a waiver of individual consent requirements; the imposition of restrictions on employers from requiring genetic testing and information from their employees; or in limiting the ability of a person to initiate parentage testing without the knowledge and consent of the child and the other parent.

In an earlier era, the typical centrepiece of any significant law reform effort was the recommendation of a major new piece of legislation. However, in a more complex environment in which authority is much more diffused, modern law reform efforts are likely to involve a mix of strategies and approaches, including legislation and regulations; official standards and codes of practice (such as those promulgated by the NHMRC and the federal Privacy Commissioner); industry codes and best practice standards; education and training programs; better coordination of governmental and intergovernmental programs; and so on. The recommendations contained in this Report are addressed to a wide range of parties and not merely to the Commonwealth Government. For this reason, the Report contains an 'Implementation Schedule',

making clear the lines of responsibility for implementation of the various recommendations.

The Inquiry's key recommendations include the following:

- A standing Human Genetics Commission of Australia (HGCA) should be established to provide high-level, technical and strategic advice to Australian governments, industry and the community about current and emerging issues in human genetics, as well as providing a consultative mechanism for the development of policy statements and national guidelines in this area.
- Discrimination laws should be amended to clearly prohibit unlawful discrimination based on a person's real or perceived genetic status.
- Privacy laws should be harmonised and tailored to address the particular challenges of human genetic information. Among other things, this will require extending privacy protection to genetic samples as well as genetic information. However, the familial dimension of genetic information also requires acknowledgment—for example, doctors should be authorised to disclose personal genetic information to a genetic relative in circumstances where disclosure is necessary to lessen or prevent a serious threat to an individual's life, health, or safety.
- A new criminal offence should be created to prohibit an individual or a corporation from submitting another person's sample for genetic testing, or conducting such testing, knowing (or recklessly indifferent to the fact) that this is done without the consent of the person concerned or other lawful authority.
- Ethical oversight of genetic research should be strengthened by: ensuring that all genetic research complies with NHMRC standards; better supporting Human Research Ethics Committees (HRECs); providing more guidance to researchers and research participants about best practice; developing new rules to govern the operation of human genetic research databases; and tightening reporting requirements.
- An ethical dimension should be added to the accreditation standards of the National Association of Testing Authorities, Australia (NATA), and only accredited laboratories should be permitted to conduct genetic testing for health and medical purposes. The Therapeutic Goods Administration (TGA) should be empowered to regulate genetic testing devices that may be provided directly to the public.
- As a matter of priority, Australian governments should develop strategies designed to assess and respond to the need for increased and adequately resourced genetic counselling services.
- Employers should not gather and use genetic information except in rare circumstances, for example, where this is necessary to protect the health and

safety of workers or third parties, and the action complies with stringent standards developed by the HGCA and the National Occupational Health and Safety Commission (NOHSC).

- A range of safeguards and improved policies and practices should be applied to the insurance industry's use of genetic information (including family history) for underwriting purposes. This will be aimed at ensuring that: genetic information must be used in a scientifically reliable and actuarially sound manner, in accordance with HGCA recommendations; reasons must be provided for unfavourable underwriting decisions; industry complaints handling processes are improved and extended to cover review of underwriting decisions based on genetic information; and industry education and training in this area will be significantly enhanced.
- DNA parentage testing should be conducted only with the consent of each person sampled, or pursuant to a court order. In the case of a child who is unable to make an informed decision, testing should go ahead only with the consent of both parents, or pursuant to a court order. In those cases in which agreement cannot be reached—for example, because a mature child or a person with parental responsibility withholds consent or is unavailable—a court may authorise testing, after taking the child's interests into account. In order to ensure high ethical standards and technical competence, DNA parentage testing should be conducted only by NATA-accredited laboratories, operating in accordance with the specific accreditation standards in this area. Information about the availability of genetic counselling should be provided to the parties.
- In order to facilitate an effective national approach to sharing DNA information for law enforcement purposes, Australian governments should develop national minimum standards with respect to the collection, use, storage, destruction and index matching of forensic material (and the DNA profiles created from such material). No inter-jurisdictional sharing of information should be permitted except in accordance with these national minimum standards.

Part A: Introduction

Chapter 1 provides an introduction and background to the Report, and includes the details of the Inquiry's research and public consultation efforts. Chapter 2 is intended to serve as a basic 'primer' on genetics and human health, to provide the scientific background for this Report. Among other things, the chapter describes the emerging understanding of genetic science in the wake of the Human Genome Project, including: the nature of DNA, RNA, genes and chromosomes; patterns of inheritance of genetic traits; the interplay among genes and between genetics and the environment; and the implications of the 'New Genetics' for human health.

Chapter 3 examines the nature of genetic information and some of the facts (and myths) about the implications that may be drawn from information about a person's genetic status. One of the central issues for the Inquiry was whether to accept

arguments in favour of ‘genetic exceptionalism’—that is, the idea that genetic information is so fundamentally different from, and more powerful than, all other forms of personal health information that it requires different or higher levels of legal protection. On the other hand, genetic ‘inclusivists’ argue that genetic information is neither distinctive nor unique in its ability to predict an individual’s health, but indicates only a rough range of probabilities.

The Inquiry has concluded that an exceptionalist approach would divorce genetic information from the principles, processes and institutions that have been developed over time to provide ethical oversight of research, ensure best practice in clinical medicine, protect personal privacy, and prohibit unlawful discrimination. However, the Inquiry does accept that there are some special features and issues attaching to genetic information which make it necessary to engage in a thorough inspection of existing principles, practices and safeguards, and of the legal, ethical and regulatory landscape, to ensure that all of these are adequate to the task.

Chapter 3 also considers the dangers of ‘genetic essentialism’—a reductionist view of human beings as essentially consisting of their genes, with human worth describable in the language of genetics. The challenge for society is to maintain its moral and ethical compass, supporting those aspects of genetic science that reduce pain and suffering and increase quality of life, while firmly resisting the use of this knowledge to diminish personal freedom and personal responsibility, or create new opportunities for unfair discrimination.

It may be the case that scientific and technological advances will be so rapid that some of the bases for policy-making may be dated in a relatively short span of years. Chapter 4 seeks to address the ways in which the Inquiry’s recommendations are intended to have longevity in the context of rapid scientific change, such as through the establishment of institutions charged with the responsibility of providing expert advice about emerging issues in human genetics, as they arise.

Accordingly, in Chapter 5, the Inquiry recommends the establishment of an independent, standing advisory body—the HGCA—following the lead of the United Kingdom and Canada in this regard. The principal role of the HGCA would be to provide on-going, high-level technical advice to Australian governments about existing and emerging issues in human genetics, and the ethical, legal and social implications arising from these developments. The HGCA should have balanced and broad-based membership, involving community representation as well as persons with the appropriate technical expertise.

The HGCA also would play a leadership role at the national level in promoting harmonisation of laws and practices; promoting public engagement and community and professional education; and developing policy statements and national guidelines in this area, in association with other governmental agencies or the relevant industries and organisations. For example, the Inquiry recommends that the HGCA be assigned specific responsibility for assessing and making recommendations on whether

particular genetic tests should be used for insurance underwriting or in employment for compelling occupational health and safety purposes.

Part B: Regulatory Framework

Part B sets the scene for the bulk of the Report's recommendations by examining the existing regulatory framework in respect of ethics, privacy and discrimination that applies to the collection, use and protection of human genetic samples and information. The discussion in Chapter 6 deals with the way ethical considerations influence the various contexts in which human genetic samples and information are used. In contrast, the discussions of privacy in Chapters 7 and 8 and of discrimination in Chapter 9 are based largely around formal legal regulation.

Chapter 6 describes the nature of ethical considerations, the features of genetic information that raise these issues and the ways that decisions involving ethical considerations can be justified. It describes the many philosophical and theological concepts and theories that have contributed to the contemporary understanding of ethics—including, for example, 'principlist', 'consequentialist' and 'critical' ethical theory. The Inquiry believes that any successful scheme for regulating genetic information in the public interest must incorporate and internalise a strong ethical dimension, and keep in mind the important role that ethics should play in areas that come under formal, legal regulation.

Chapter 7 summarises the existing complex, fragmented and overlapping frameworks for the protection of information and health privacy based on the *Privacy Act 1988* (Cth) (*Privacy Act*) and similar state and territory legislation. The chapter recommends that the Commonwealth, States and Territories, as a matter of high priority, should pursue the harmonisation of information and health privacy legislation as it relates to human genetic information.

A key issue identified by the Inquiry, and examined separately in Chapter 8, is whether information and health privacy legislation should be extended to cover genetic samples. Although genetic samples hold a great deal of personal information that may be revealed by DNA testing and analysis, the samples themselves do not currently receive protection under the *Privacy Act*. The Inquiry recommends that the coverage of the *Privacy Act* be extended to provide minimum, legally enforceable, privacy standards for the handling of identifiable genetic samples.

Chapter 9 describes the legal framework for preventing discrimination in Australia at the federal, state and territory levels, and includes a discussion of current developments in the international arena. The Inquiry considered the desirability of 'stand alone' legislation to address discrimination on the ground of genetic status, but concludes that working within the existing legal framework is more likely to promote certainty and consistency, with the efficiency of building upon existing understandings and practices. The Inquiry recommends amendment of the *Disability Discrimination Act 1992* (Cth) (DDA) to clarify that the legislation applies to unlawful discrimination based on genetic status.

Part C: Genetic Testing

Chapter 10 observes that genetic testing can be divided into three broad categories based on the purpose of the testing, namely: medical testing, identification testing and kinship testing. Medical testing includes diagnostic testing; predictive or presymptomatic testing; genetic carrier testing; screening testing; pre-implantation and prenatal testing and testing for medical or scientific research. Identification testing or forensic testing is mainly performed on the non-coding portion of the human genome, to construct a ‘DNA profile’. This identification process is used in criminal investigations to exclude or identify a suspect, in searches for missing persons, and in the identification of deceased persons. These categories are not fixed—genetic testing sought for one purpose can sometimes reveal unintended information, such as where a medical test reveals incidental information about parentage (or, more sensitively, about misattributed parentage).

Chapter 11 considers a range of issues concerning access to genetic testing. To date, medical practitioners have been the primary ‘gatekeepers’ of access to genetic testing and the genetic information derived from it, at least for clinical purposes. Genetic testing for medical purposes usually requires a referral from a medical practitioner. However, genetic testing products and services also may be provided directly by laboratories, and testing kits increasingly are being marketed directly to the public.

Chapter 11 describes the laboratory accreditation system and makes recommendations aimed at requiring laboratories to be accredited and to enhance accreditation standards to ensure high ethical standards in genetic testing. The chapter then focuses on issues concerning the availability and use of genetic testing services provided directly to the public. The Inquiry recommends reforms to enable the TGA to regulate the products used in genetic testing provided directly to the public more effectively, including DNA identification test kits, used in parentage and other kinship testing.

Chapter 12 examines the serious ethical and privacy concerns that arise from non-consensual testing. A great deal of sensitive, personal information can be derived from genetic testing of a bodily sample, with important health, legal, and social implications for individuals, families and others. Access to genetic testing is now readily and increasingly available. Existing law provides only limited protection against non-consensual testing.

The Inquiry concludes that there should be additional legal protection against the unauthorised testing of genetic samples and, to this end, recommends a new criminal offence for enactment into Commonwealth, state and territory law. The Inquiry recommends that criminal liability should attach to any individual or corporation that, without lawful authority, submits a sample for genetic testing, or conducts genetic testing on a sample, knowing (or recklessly indifferent to the fact) that the person from whom the sample was taken did not consent to such testing.

Part D: Human Genetic Research

Part D examines ethical, privacy and related issues concerning the use of genetic samples and information in the conduct of human genetic research. Chapter 13 summarises the present regulatory framework for the ethical conduct of research centred on the NHMRC's *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) and on review of research proposals by HRECs.

Human genetic research generates knowledge with the potential to improve dramatically the diagnosis, prevention and treatment of serious health and medical problems. Research also can reveal information about an individual's susceptibility to disease and hence about the individual's future health. The recommendations in this Part are intended to balance the interests of researchers—who need access to human genetic samples and information from many sources—and the needs of individuals and their relatives—whose autonomy must be respected and whose privacy must be protected.

The requirements for institutions or HRECs to be registered with the NHMRC and to follow the processes set down in the National Statement are incomplete. For example, there is currently no obligation on private research bodies to adhere to the provisions of the National Statement. Chapter 14 examines these matters and, in light of the increasing commercialisation of human genetic research, recommends strengthening of the mechanisms by which compliance with the National Statement is enforced.

The concept of consent is fundamental to the legal and ethical regulation of human research. Other than in limited and defined circumstances, the National Statement generally requires consent to the use of human tissue samples, genetic material and genetic information in medical research. Chapter 15 highlights concerns about various aspects of consent and, in particular, whether the current provisions under which HRECs may *waive* consent requirements are adequate to the tasks of protecting privacy and autonomy. A related issue concerns the extent to which researchers are able to obtain consent from potential research participants for the use of their genetic samples or information for unspecified future research. The Inquiry recommends the imposition of new reporting obligations for HRECs about proposals for which waiver of consent has been granted under the National Statement; and augmenting training and other support for HRECs in relation to decision making on waiver of consent. The Inquiry also recommends amending the National Statement to provide clear guidance about obtaining consent to unspecified future human genetic research.

The Inquiry suggests that it would be useful for the NHMRC to develop information and advice, including examples and practical guidance, for the preparation of human genetic research protocols and consent forms for human genetic research. The intention of these documents would be to give further direction to researchers and HRECs about what the NHMRC considers to be best practice in the conduct of human genetic research. The content of these documents is discussed in Chapter 16. Particular concerns relate to the coding and de-identification of genetic samples and information,

and to the disclosure to research participants of the potential commercialisation of research outcomes.

Chapter 17 discusses a range of options aimed at strengthening the current system of ethical review of human genetic research and the role of HRECs within that system. The Inquiry recommends that the NHMRC develop and implement procedures to promote consistency, efficiency, transparency and accountability in HREC review of human genetic research, through a systematic quality improvement program.

This program should address, among other things: the membership of HRECs and, in particular, the balance between institutional and non-institutional members; the expertise available to HRECs in considering proposals for human genetic research; the on-going monitoring of approved human genetic research projects; the education and training of HREC members; and standardised record keeping and reporting to the NHMRC, including in relation to commercial arrangements. The Inquiry also recommends that the NHMRC should review the need for a formal accreditation system for HRECs in respect of the ethical review of human genetic research.

Part E: Human Genetic Databases

Part E deals with the regulation of genetic samples and information in databases created specifically for use in research (human genetic research databases) and in collections formed for other purposes that may have research uses (human tissue collections). Part E also considers regulatory issues related to the law of property and the Human Tissue Acts.

Chapter 18 is concerned with genetic samples and information collected primarily for use in research and held in collections by hospitals, public and private research organisations, and in the archives of pathology laboratories. Researchers increasingly are compiling such collections to aid studies into the causes of disease, drug reactions, and the interaction between genetics and the environment. The Inquiry recommends that the NHMRC amend the National Statement to provide ethical guidance on the establishment, governance and operation of human genetic databases—and, in particular, to offer specific guidance on obtaining consent to unspecified future research.

As part of these changes to the National Statement, the Inquiry recommends that the NHMRC establish and administer a public register of human genetic research databases, develop conditions of registration, and include provisions so that no genetic research under the National Statement can be conducted using information from a database unless it is duly registered. The Inquiry also recommends that the National Statement be amended to provide guidance on the use of a ‘gene trustee’ system, where appropriate, as an additional privacy protection.

Finally, Chapter 18 recommends that the Australian Health Ministers’ Advisory Council (AHMAC) should develop nationally consistent rules governing the disclosure, for law enforcement purposes, of genetic samples and information held in human genetic research databases. These rules should be based upon the principle that

such disclosure is permissible only with the consent of the person sampled or pursuant to a court order.

Chapter 19 is concerned with genetic samples and information held in tissue collections maintained chiefly by hospitals or pathology laboratories, which have *not* been collected primarily for use in research. For example, archived collections of preserved human tissue and collections of newborn screening cards (also known as ‘Guthrie cards’) are invaluable research resources for studies into the genetic causes of disease. Genetic testing of stored tissue samples has potential uses in other contexts, including criminal or police investigations, as evidence in court proceedings and for parentage or other kinship testing. These secondary uses raise important issues of ethics, privacy and consent. The Inquiry recommends that AHMAC also develop nationally consistent rules in relation to the handling of newborn screening cards and other human tissue collections. Disclosure for law enforcement purposes should be permissible only with the consent of the person sampled (or a person authorised to consent on his or her behalf) or pursuant to a court order.

Chapter 20 considers whether privacy interests in genetic information might be protected more effectively by recognising increased property rights over genetic samples. The Inquiry concludes there should be no change to the current position whereby hospitals and pathology laboratories have a proprietary right to preserved samples, but full property rights in genetic samples are not recognised.

Each Australian State and Territory has enacted legislation that regulates the donation of human tissues and organs for transplantation and research (the Human Tissue Acts). In Chapter 20, the Inquiry examines whether amending the Human Tissue Acts to cover the handling of genetic samples might be an effective means of protecting privacy interests. Until such a review is conducted, the Inquiry believes that the regulation of the handling of genetic samples should not rely primarily on amendments to the Human Tissue Acts.

Part F: Health Services

Part F looks at systemic issues of privacy and ethical practice in the provision of medical and allied health services by doctors, genetic counsellors and other health professionals.

Historically, confidentiality has been a cornerstone of the doctor-patient relationship. However, genetic information about an individual may have important health implications for that person’s genetic relatives. Chapter 21 focuses on questions about how individual patients and their doctors (and other health professionals, including genetic counsellors) should collect and deal with genetic information about genetic relatives, derived in the course of diagnosis, treatment or counselling.

The Inquiry concludes that there may be exceptional circumstances in which health professionals should be permitted to disclose personal genetic information to genetic relatives without the consent of their patient. Consequently, the Inquiry recommends

amending the *Privacy Act* to permit such disclosures in circumstances where disclosure is necessary to lessen or prevent a serious threat to an individual's life, health, or safety, even where the genetic risk is not 'imminent' (which is the current language of the Act).

In Chapter 21, the Inquiry also recommends that the *Privacy Act* should be amended to provide that an individual has a limited right to access genetic information about first-degree genetic relatives where access is necessary to lessen or prevent a serious threat to their life, health, or safety, even where the threat is not imminent.

The collection and disclosure of family genetic information is central to the operation of genetic registers and the conduct of genetic counselling. The primary purpose of genetic registers is to identify and contact members of families who are at significantly increased risk of developing an inherited disorder or of having affected children. Information on a genetic register generally will comprise genetic information about many biological relatives and also may contain genetic samples.

Chapter 22 deals with the operation of genetic registers and examines whether the existing regulatory framework to protect the privacy of genetic information collected and held on genetic registers is adequate. Medical practitioners already have received an authorisation (a 'Public Interest Determination' or PID) from the federal Privacy Commissioner to collect family medical history information for clinical purposes without breaching the *Privacy Act*. The Inquiry recommends that organisations operating family cancer registers and other genetic registers also should receive a PID in order to continue collecting family medical history information without breaching privacy legislation.

Genetic test information may have profound medical and psychological implications for individuals. These implications will depend on the nature and context of genetic testing—the genetic condition being tested for and the reasons for testing. The results and implications of genetic testing often will be complex and difficult to understand and it is therefore critically important that individuals are provided with appropriate information about a genetic test and, in some cases, assisted in decision making through genetic counselling.

Chapter 23 discusses genetic counselling, the need for genetic counselling services, and issues related to its further development as a professional discipline. The Inquiry recommends that the Commonwealth, States and Territories should develop strategies to assess and respond to the need for increased and adequately resourced genetic counselling services throughout Australia and examine options for the further development of genetic counselling as a recognised health profession, including the possibility of new certification, accreditation or registration systems for genetic counsellors.

The Inquiry recommends that the HGCA should develop guidelines, in consultation with interested organisations, to identify genetic tests, or categories of genetic tests, that require special treatment in relation to procedures for ordering testing and ensuring

access to genetic counselling. The Inquiry also recommends approaches to ensure that present and future medical practitioners are appropriately trained and equipped in clinical genetics and in the use of relevant genetic counselling and genetic services.

Chapter 24 discusses the types of population genetic screening programs that can be undertaken and the guidelines that have been developed to regulate them. Such programs involve testing large numbers of people—who usually have no disease symptoms—for their genetic status in relation to a particular gene or condition. Examples of population genetic screening programs include the Tay-Sachs screening program that operates in Jewish schools in Sydney and Melbourne, and the ‘HaemScreen’ (haemochromatosis) program operated at some places of employment. Of course, in one sense, there is a universal screening program in Australia, since virtually all newborns are given the ‘heel prick’ test to screen for a range of genetic conditions, such as phenylketonuria, hypothyroidism and galactosaemia.

Because population genetic screening programs raise a range of privacy, ethical and other issues, the Inquiry recommends this as another area in which AHMAC should develop nationally consistent standards. Standards in this context should cover informed consent, testing standards, quality assurance, cost-benefit considerations, and reporting and data collection.

Part G: Insurance

Concern about the use of genetic information by the insurance industry was one of the main factors leading to the establishment of this Inquiry. The Inquiry has considered the evidence provided in submissions, the role of insurance in the Australian community and alternative models regarding the use of genetic information in underwriting personal insurance, such as those developed in some European countries.

The Inquiry concludes that the evidence does not support a departure at this time from the fundamental principle that has long governed the voluntary, mutually rated, personal insurance market—namely, equality of information between the applicant and the insurer. However, the Inquiry does recommend a range of reforms that target specific concerns about how insurers use genetic information, and how persons who have received an adverse underwriting decision may seek to have this reviewed.

Chapter 25 describes the framework for insurance in Australia, including key insurance concepts and the differences between various types of insurance products—particularly the difference between mutually rated and community rated insurance. Most concerns raised in submissions related to mutually rated insurance and the use of genetic information by insurers to calculate risk. The chapter also examines the exchange of information between applicants and insurers; the type and amount of genetic information currently collected by insurers; and industry policy in this area, especially the Genetic Testing Policy developed by the Investment and Financial Services Association (IFSA), the peak body of life insurers.

Chapters 26 and 27 deal with the substance of the concerns raised in submissions. The recommendations in these chapters proceed on the basis that parity of information between the applicant and the insurer is necessary in a voluntary mutually rated insurance market. Anti-discrimination legislation throughout Australia recognises that it is necessary for mutually rated insurance to differentiate among individuals on the basis of certain characteristics. The DDA, for example, includes a specific exception for discrimination in insurance where the conduct is based upon reasonable actuarial or statistical data, or is otherwise reasonable.

The Inquiry makes a number of recommendations aimed at promoting fair underwriting practices to maintain public confidence in the use of genetic information by insurers. For example, the Inquiry recommends that: the HGCA be given the specific role of making recommendations about the use of particular genetic tests for underwriting purposes; insurers be obliged to provide applicants with clear and meaningful reasons for unfavourable underwriting decisions based on genetic information; and the industry's review and appeal mechanisms for adverse underwriting decisions be expanded and improved. The HGCA is also assigned a general 'watching brief' over insurance industry use of genetic information, including family medical history.

Chapter 28 examines the privacy protection afforded to genetic information in the insurance context. Although the insurance industry holds large quantities of sensitive personal information, including health information, submissions reflected a reasonable degree of satisfaction with the industry's practices in relation to information privacy. Insurers are now covered by the private sector provisions of the *Privacy Act* and are obliged to comply with the National Privacy Principles (NPPs) in the handling of all personal information that they collect and hold.

The Inquiry considered the impact of various insurance industry practices on the nature and quality of the consent to provide genetic information in an application for insurance. It is recommended that insurers review their consent and medical authority forms to ensure that they contain sufficient information about the collection, use and disclosure of genetic information to allow applicants to make informed decisions. The Inquiry also recommends that insurers do not 'bundle' requests for consent to collect genetic information for the purpose of assessing insurance applications together with consent for other purposes.

Chapter 28 also considers the practice of collecting of family medical history from insurance applicants without the knowledge or consent of the genetic relatives to whom the information relates. It is recommended that the industry apply for a PID under the *Privacy Act* to confirm the legitimacy of this practice.

Part H: Employment

Part H deals with genetic testing and information in employment. Chapter 29 discusses the types of genetic information that might be relevant in this context, and the legal and ethical issues involved in the potential use of such information. There is little evidence that Australian employers are currently seeking access to genetic information about job

applicants or employees, although there is some evidence of this occurring overseas. However, other forms of workplace testing (such as drug and alcohol testing, and psychometric testing) that were unknown some years ago are now becoming relatively commonplace. There is little doubt that the pressures to use genetic information will increase as the reliability and availability of genetic tests increases, and as the cost of testing decreases.

Chapter 30 describes the existing regulatory framework governing discrimination in employment and considers its application in relation to employers' requests for, or use of, genetic information. The Inquiry considers that there are sound reasons of public policy for making a strong intervention into the use of genetic information by employers, and recommends that, as a general matter, employers should not collect or use genetic information in relation to job applicants or employees. However, the Inquiry acknowledges that there may be rare circumstances where such action may be necessary to protect the health and safety of workers or third parties, and this should be permitted if the action complies with stringent privacy, discrimination and other safeguards, including standards developed by the HGCA and the NOHSC.

Chapter 31 sets out three main concerns regarding the existing anti-discrimination framework. The first concern is the scope of the 'inherent requirements' of the job exemption in the DDA and other legislation. The Inquiry recommends that the assessment of an applicant or employee's ability to perform the inherent requirements of a job should not include an assessment of whether the applicant or employee will be unable to perform the inherent requirements in the future by reason of his or her genetic status, unless it is reasonable to do so in the exceptional circumstances of the particular case.

The second concern relates to the current absence of effective restraints upon the ability of employers to request genetic information from employees. The Inquiry recommends that such requests should be unlawful, except where such information is reasonably required for a purpose that does not involve discrimination—such as to ensure that a person is able to perform the inherent requirements of the job.

The third concern relates to the need for guidance in relation to the use of genetic information by employers, to ensure that test results are interpreted accurately and that employers do not use genetic information inappropriately. The Inquiry recommends that the Human Rights and Equal Opportunity Commission, in consultation with the HGCA and other stakeholders, develop guidelines to deal specifically with the collection and use of genetic information in employment. The Attorney-General also should consider the development of Disability Standards in this area.

Chapter 32 discusses employers' duties regarding occupational health and safety, and considers each of the ways in which genetic screening or monitoring could be used in this context. Briefly, employers might seek to conduct genetic testing to screen for work-related susceptibilities, to monitor workplace-induced conditions, or to screen for the purpose of protecting the safety of third parties.

Strong concerns were expressed about the potential use of genetic screening programs by employers, in particular the worry that employers might use such screening or monitoring to exclude ‘high risk’ individuals rather than taking all reasonably practicable steps to remove hazardous substances from the workplace. It was suggested that employers have economic incentives to transfer their responsibility for workplace safety onto their employees, and would do so in this way.

The Inquiry is sensitive to these concerns but also recognises the potentially beneficial use of genetic information to protect employees against the onset of serious medical conditions to which they have a particular susceptibility, or for which they exhibit early signs of development. There is also a need to take steps to protect third parties from unreasonable risks to their health and safety, especially in areas involving inherent dangers, such as public transport or the storage or transport of dangerous chemicals.

Accordingly, the Inquiry recommends that employers should be able to collect and use genetic information *in limited circumstances*, for example, where this is directly relevant to the discharge of their obligations to protect employees and third parties from serious dangers to their health or safety. This only would be available where: the employer already has taken all reasonable steps to eliminate the environmental hazard from the workplace; there is clear evidence of a connection between the working environment and onset of a genetic condition; and there is a scientifically reliable method of screening for the condition. To this end, the Inquiry also recommends that the HGCA and NOHSC develop national guidelines and codes of practice to govern the use of genetic information in the workplace, and that the HGCA provide specific advice about which tests are effective for use in these circumstances.

Chapter 33 discusses the potential use of genetic information in the context of a workers’ compensation claim, or a common law claim arising out of work-related injury or death. The Inquiry outlines the reasons that employers might seek to obtain an applicant or employee’s family medical history or genetic test results in this context. The Inquiry recommends that the HGCA should develop a policy regarding the appropriate use of genetic information in the assessment of workers’ compensation claims.

Chapter 34 identifies the existing privacy protections applying to genetic information obtained from job applicants and employees in the course of their employment, and notes the existence of the ‘employee records’ exemption in the *Privacy Act*. There appears to be no reasonable basis for the fact that the health information of public sector employees is protected but the health information of private sector employees is not. The Inquiry concludes that the employee records exemption is too broad and recommends amendment of the *Privacy Act* so that genetic information held by private sector employers about their employees is given the same high level of privacy protection afforded to the same information when held, for example, by health service providers or insurers.

Part I: Other contexts

Genetic testing provides a powerful tool for identifying or dispelling biological linkages between individuals—that is, in establishing kinship relations. Chapter 35

considers this matter in the context of parentage testing. Chapter 37 considers this matter in the context of establishing kinship relationships for immigration purposes. In Chapters 2 and 3, the Report notes that genetic information not only has a strong familial dimension, but can also contain links beyond the individual to the broader descent group or community. Chapter 36 considers some of these sensitive questions of the relationship between kinship and identity. Chapter 38 takes a different tangent, examining the potential use of genetic testing and information in sport.

Chapter 35 discusses the use of DNA testing for the purpose of determining parentage or kinship. This may arise in a variety of contexts—most commonly in family law and child support proceedings, but also in relation to succession to estates, identification of human remains, genealogical curiosity, and so on.

The Inquiry received a large number of submissions and representations from various support groups, laboratories conducting parentage testing, and private individuals—both in support of, and sharply critical of, aspects of the current regulatory framework and industry practice. In particular, many submissions were forwarded by ‘men’s rights’ or ‘father’s rights’ groups. Concerns focused on access to parentage testing; the provision of what is known as ‘motherless testing’; consent and decision making in relation to testing (in particular on behalf of children); the provision of associated counselling and support; and the need to protect against fraudulent practices both in relation to alleged ‘paternity fraud’ and fraudulent parentage testing.

The Inquiry recognises the sensitivities in this area, and the need for greater regulation of parentage testing in the public interest. The Inquiry recommends that DNA parentage testing should be conducted only by NATA-accredited laboratories, operating in accordance with the specific accreditation requirements in this area. The Inquiry also recommends that NATA should review these accreditation requirements to ensure that they meet the highest technical and ethical standards, particularly in relation to consent to testing, protecting the integrity of genetic samples, and providing information about counselling.

The Inquiry recommends that parentage testing reports should be inadmissible in proceedings under the *Family Law Act 1975* (Cth) unless the testing complies with the *Family Law Regulations 1984* (Cth). Other forms of kinship testing, such as sibship testing, are currently unregulated, and the Inquiry recommends that the NATA accreditation program be extended to these forms of testing.

One of the central concerns of the Inquiry was to ensure that the personal dignity and autonomy of the child not be overlooked in any argument about parental rights. After considerable thought, the Inquiry recommends a two-tiered system for regulating consent for children’s participation in parentage testing. A child over the age of 12 years, who is independently assessed as having sufficient maturity to do so, should be permitted to decide whether to give or withhold consent.

While the Inquiry received a number of submissions supporting continued access to ‘motherless’ testing, the Inquiry has not found these arguments compelling. Therefore, for a child under 12 years, or who is over that age but lacks sufficient maturity, the

Inquiry recommends that all those with parental responsibility for the child must make the decision on behalf of the child. In those cases in which agreement cannot be reached—for example, because a mature child or a person with parental responsibility withholds consent or is unavailable—a court may order the child’s participation, after taking the child’s interests into account.

Finally, the Inquiry recommends that NATA should develop accreditation requirements requiring laboratories to inform all persons who provide genetic samples for parentage testing of the availability of counselling both at the time the samples are submitted and at the time the results are made available.

Chapter 36 considers issues of kinship and identity, and discusses the use of genetic testing and information in Australia and overseas, in efforts to establish (or refute) membership of particular communities. The Inquiry was correct in forecasting, in earlier consultation papers, that arguments could arise about whether genetic information could or should be used as a means of establishing Aboriginal or Torres Strait Islander identity for the purposes of determining eligibility to vote in Aboriginal and Torres Strait Islander Commission (ATSIC) elections—as occurred in Tasmania in 2002. However, the Inquiry remains deeply sceptical about whether there is any proper role for genetic testing in determining Aboriginal identity, which is primarily a social and cultural construct. Accordingly, these are matters to be determined by Aboriginal and Torres Strait Islander people themselves, working through their own communities, institutions and consultation processes. The chapter provides substantial research and commentary from Australia and overseas intended to assist any further consideration of these issues, as well as to promote general community understanding of these sensitive matters.

Chapter 37 examines the law and practices that regulate the use of genetic testing for migration decision making under the *Migration Act 1958* (Cth) and associated regulations. Kinship testing may be used in migration decision making to confirm family relationships for certain types of visas or to detect possible fraud (such as sibling marriages and child trafficking) in migration applications.

The use of genetic testing by immigration authorities is primarily regulated by internal policy guidelines, rather than by legislation or regulations. The Inquiry recommends that the use of genetic testing in migration decision making should be subject to more formal control in order to ensure the protection of privacy and personal autonomy.

The Inquiry recommends that the Department of Immigration and Multicultural and Indigenous Affairs (DIMIA) review its policies and procedures regarding the provision of information to migration applicants about kinship testing. In particular, these revised policies should ensure that visa applicants are: informed early in the application process that they may be requested to undergo a genetic kinship test; provided with DIMIA’s reasons for requesting the applicant undergo a genetic test and given an opportunity to address the doubts through other evidence; and provided with information about the implications of testing and the desirability of seeking counselling.

As a general matter, health status is a relevant consideration in migration decision making. Genetic testing has not been a feature of health assessment to date. However, in common with other areas under consideration by this Inquiry, it is not difficult to foresee that this will change in future as genetic testing services increase in availability and reliability, and decrease in unit cost. The Inquiry recommends that the Department of Health and Ageing, in consultation with DIMIA and the HGCA, should develop policies on genetic tests and the use of genetic information (including family medical history) for the purpose of assessing the health requirement under migration legislation.

Chapter 38 examines the use of genetic information in sport—in particular, to identify potential elite athletes carrying ‘performance’ genes and individuals with a genetic predisposition to sports-related injury. While the use of genetic information in this way is still largely experimental, the Inquiry recommends that the Australian Sports Commission develop policies and guidelines for the use of sporting organisations and athletes.

Part J: Law Enforcement and Evidence

Part J deals with the use of genetic testing and information in criminal investigations, the identification of deceased persons and in criminal and civil proceedings. DNA profiling already is a major tool for Australian law enforcement authorities. Contrary to the practice of DNA testing in the clinical and research contexts, forensic testing is performed on the non-coding section of DNA, at a number of agreed sites (‘loci’), in order to construct an alpha-numeric DNA profile for identification purposes.

Chapter 39 discusses the current and potential uses of DNA profiling in criminal investigations and provides an overview and analysis of forensic procedures legislation in Australia, especially Part 1D of the *Crimes Act 1914* (Cth) (*Crimes Act*).

Chapter 40 focuses on concerns arising from the lack of harmonisation among Commonwealth, state and territory forensic procedures laws and practices. The Inquiry recommends that, in order to facilitate an effective national approach to sharing genetic information, the Commonwealth, States and Territories should develop national minimum standards in Australian forensic procedures legislation with respect to the collection, use, storage, destruction and index matching of forensic material (and the DNA profiles created from such material). No inter-jurisdictional sharing of information should be permitted except in accordance with these national minimum standards.

Chapter 41 discusses specific issues arising from the regulatory framework for forensic procedures. The Inquiry recommends that the legislative definition of ‘destruction’—(for genetic samples and profiles) should provide for physical destruction of genetic samples or permanent and irreversible de-identification of profiles. The Inquiry also recommends that the Commonwealth develop and publish guidelines for the conduct of ‘mass DNA screening’ programs which are conducted as part of a criminal investigation—both in relation to the authorisation process for such programs, as well as the manner in which they are conducted.

The Inquiry also asks the Commonwealth to review ‘consent’ provisions in the *Crimes Act* that apply to suspects and serious offenders so that a forensic procedure could be conducted only pursuant to an order made by a judicial officer or an authorised police officer, in accordance with existing provisions for ordering a forensic procedure. This would better reflect the inherently coercive nature of the procedures in these circumstances, and would remove potential arguments that consent given by a suspect or serious offender was not a valid informed consent.

Chapter 42 considers the use of DNA testing and information for the purpose of identification of missing and deceased persons. Subsequent to the Bali terrorist bombings in October 2002, the Commonwealth amended the *Crimes Act* to facilitate the identification of the victims. The Inquiry recommends that the Commonwealth, States and Territories should clarify the arrangements under which police officers of one jurisdiction are authorised to act on behalf of another jurisdiction in collecting, using, storing or destroying forensic material from a missing or deceased person, or a genetic relative of a missing or deceased person.

In addition, the Inquiry recommends that where information stored on a DNA database system is disclosed to Interpol or any other foreign agency, the Commonwealth must take reasonable steps to ensure that the information transferred will not be held, used or disclosed by the recipient inconsistently with Australian minimum standards.

Chapter 43 discusses issues arising from the regulation and operation of DNA database systems in the law enforcement context. To date, the Commonwealth has established two national DNA database systems—the National Criminal Investigation DNA Database (NCIDD) and the Disaster Victim Identification (DVI) Database—for the storage and matching of DNA profiles and the inter-jurisdictional sharing of information for law enforcement purposes. In order to ensure that all DNA databases established for law enforcement purposes are subject to Part 1D of the *Crimes Act*, the Inquiry recommends that the Commonwealth should amend the definition of a ‘DNA database system’.

To promote public confidence in the integrity of the national DNA database systems, the Inquiry recommends that the Commonwealth should add independent members (that is, members not affiliated with a police agency) to CrimTrac’s board of management, and provide for periodic, independent audits of the operation of all DNA database systems operating pursuant to the Act.

In Chapter 44, the Inquiry calls for better education about the use of DNA evidence for those lawyers and judges involved in criminal proceedings. The Inquiry also recommends that the judiciary develop a model jury direction regarding the need for caution in evaluating DNA evidence and the statistical calculations relating to that evidence. It is also recommended that the National Institute for Forensic Science should play a key role in providing ongoing guidance to forensic scientists and legal practitioners regarding reliable methods of DNA analysis, statistical calculation, and presentation of evidence in criminal proceedings. Similarly, in Chapter 46, it is

recommended that continuing judicial and legal education programs be developed in relation to the use of DNA evidence in *civil* proceedings.

The ‘Innocence Project’ in the United States has demonstrated the power of DNA evidence as a tool for challenging miscarriages of justice through post-conviction review—by the end of February 2002, 123 convicted offenders had been exonerated in this way, including a number on ‘death row’. In Chapter 45, the Inquiry recommends that the *Crimes Act* be amended to require the long-term retention of forensic material found at the scene of serious crimes to facilitate post-conviction analysis, and that a process be established to consider applications for post-conviction review from any person who alleges that DNA evidence may exist that would call into question his or her conviction.

List of Recommendations

Part A. Introduction

5. A Human Genetics Commission of Australia

- 5-1 The Commonwealth should establish a Human Genetics Commission of Australia (HGCA) under federal legislation as an independent statutory authority with sufficient resources to fulfil its mission.
- 5-2 As a general matter, the role of the HGCA should be to provide:
- on-going, high-level, technical and strategic advice to Australian governments about current and emerging issues in human genetics;
 - similar high-level advice on the ethical, legal and social implications arising from these developments, including consideration of any impact on human rights, and analysis of cost-benefit issues;
 - national leadership in managing the process of change in relation to human genetics, including engagement of the public on these issues;
 - relevant expertise and a consultative mechanism for the development of policy statements and national guidelines in this area, where appropriate in association with other government agencies or the relevant industries and organisations;
 - assistance with the development and coordination of community, school, university and professional education about human genetics;
 - advice and a consultative mechanism to assist relevant bodies in identifying strategic priorities for research in human genetics; and
 - a focus for the coordination and integration of various national—and perhaps regional and international—programs and initiatives.
- 5-3 The HGCA also should have specific role in:
- identifying genetic tests that have particular concerns or sensitivities attached to them, and thus may require special treatment;
 - making recommendations about the suitability of specific genetic tests (and the appropriate analysis and treatment of results) for use by the insurance industry (for example for risk-rating purposes), and by employers (for example for occupational health and safety reasons);

- performing any similar function or providing expert advice on any other matters relating to human genetics, whether on its own motion or in response to a formal reference from the responsible minister or ministers; and
 - monitoring the implementation of the broad strategies and specific recommendations identified in this Report.
- 5–4 The HGCA structure should involve at least two principal committees: (a) a Technical Committee, and (b) an Ethical, Legal and Social Implications Committee. However, this should not preclude the HGCA from establishing other committees or working groups (for example, on education, or community consultation) from time to time, as it sees fit.
- 5–5 Appointments to the HGCA should ensure a balanced and broad-based range of expertise, experience and perspectives relevant to the evaluation and delivery of genetic health services, and the use and protection of human genetic information and genetic samples. The appointments process should involve consultation with state and territory governments, relevant communities and other stakeholders.
- 5–6 The HGCA should operate in an open and transparent manner, to the greatest extent practicable, in order to promote public confidence and engage the wider community in uses of human genetic information.
- 5–7 The HGCA should be required to present an annual report to Parliament and also should be empowered to make such other reports to Parliament from time to time as it sees fit.
- 5–8 The HGCA should liaise closely with relevant government departments and authorities, as well as other key stakeholders, in order to promote a national approach to the protection of human genetic information.
- 5–9 The HGCA should be subject to a basic review two years after establishment, and then a more thorough, independent review after five years of operation.

Part B. Regulatory Framework

7. Information and Health Privacy Law

- 7–1 As a matter of high priority, the Commonwealth, States and Territories should pursue the harmonisation of information and health privacy legislation as it relates to human genetic information. This would be achieved most effectively by developing nationally consistent rules for handling all health information. (See also Recommendation 8–1 in relation to genetic samples.)

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- 7-2 States and Territories and privacy regulators should consider harmonising their privacy regimes, as applicable, in a manner consistent with the Recommendations in this Report. (See also Recommendations 7-4 to 7-7, 8-1 to 8-4, 21-1 to 21-3, and 22-1.)
- 7-3 The Commonwealth, States and Territories should take into account the Recommendations in this Report in developing the proposed National Health Privacy Code. (See also Recommendations 7-4 to 7-7, 8-1 to 8-4, 21-1 to 21-3, and 22-1.)
- 7-4 The Commonwealth should amend s 6 of the *Privacy Act 1988* (Cth) (*Privacy Act*) to define ‘health information’ to include genetic information about an individual in a form which is or could be predictive of the health of the individual or any of his or her genetic relatives. (See also Recommendation 8-2 in relation to genetic samples.)
- 7-5 The Commonwealth should amend s 6 of the *Privacy Act* to define ‘sensitive information’ to include human genetic test information.
- 7-6 The Commonwealth should amend the *Privacy Act* to provide that ‘health information’ includes information about an individual who has been dead for 30 years or less. These amendments should include provision for decision making by next-of-kin or an authorised person in relation to the handling of a deceased individual’s health information. (See also Recommendation 8-2 in relation to genetic samples.)
- 7-7 The Commonwealth should amend the *Privacy Act* to ensure that all small business operators that hold genetic information are subject to the provisions of the Act. (See also Recommendation 8-2 in relation to genetic samples.)

8. Privacy of Genetic Samples

- 8-1 The Commonwealth, States and Territories should enact legislation to provide legally enforceable privacy standards for handling genetic samples, including in relation to the collection, storage, use and transfer of samples. The standards should be consistent with those that apply to the handling of genetic information derived from the analysis of genetic samples under existing information and health privacy legislation such as the *Privacy Act 1988* (Cth) (*Privacy Act*).
- 8-2 The Commonwealth should amend the *Privacy Act* to extend the coverage of the Information Privacy Principles and National Privacy Principles (or similar privacy principles) to identifiable genetic samples. This may be done by:
- (a) defining ‘personal information’ and ‘health information’ to include bodily samples from an individual whose identity is apparent or can reasonably be ascertained from the sample; and

- (b) defining a 'record' to include a bodily sample.
- 8-3 The Commonwealth should amend the *Privacy Act* to provide that an individual has a right to access his or her own bodily samples, through a nominated medical practitioner, for the purpose of medical testing, diagnosis or treatment. The right of access should be limited to a right to obtain access to part of the sample. Access may be refused where:
- (a) it is not physically possible to provide part of a sample;
 - (b) providing part of a sample means that the remaining portion is insufficient for the purposes of the organisation retaining it; or
 - (c) releasing a sample to an individual raises public health concerns.
- 8-4 The Commonwealth should amend the *Privacy Act* to provide that an individual has a right to access bodily samples of his or her first-degree genetic relatives, through a nominated medical practitioner, where access is necessary to lessen or prevent a serious threat to his or her life, health, or safety, even where the threat is not imminent. The right of access should be limited to a right to obtain access to part of the sample. Where an organisation subject to the *Privacy Act* receives a request for access, the organisation should be obliged to seek consent from the genetic relative, where practicable, before determining whether to provide access. Access may be refused where:
- (a) it is not physically possible to provide part of a sample;
 - (b) providing part of a sample means that the remaining portion is insufficient for the purposes of the organisation retaining it;
 - (c) releasing a sample to an individual raises public health concerns; or
 - (d) providing access would have an unreasonable impact upon the privacy of the individual from whom the sample comes.

9. Anti-Discrimination Law

- 9-1 Discrimination on the ground of genetic status should continue to be dealt with under the framework of existing federal, state and territory anti-discrimination laws, subject to the legislative amendments and other safeguards recommended in this Report.
- 9-2 The Commonwealth should amend the objects clause of the *Disability Discrimination Act 1992* (Cth) (DDA) to clarify that the Act applies to discrimination in relation to past, present, possible future or imputed disability, including discrimination on the ground of genetic status.

- 9-3 In order to provide a consistent approach to addressing discrimination on the basis of genetic status, the Commonwealth, in consultation with the Human Genetics Commission of Australia and other stakeholders, should:
- amend the definitions of ‘disability’ in the DDA and ‘impairment’ in the regulations made under the Human Rights and Equal Opportunity Commission Act 1986 (Cth) (HREOC Act) to clarify that the legislation applies to discrimination based on genetic status;
 - amend the definition of ‘impairment’ in the regulations made under the HREOC Act to clarify the application of the legislation to a disability that may exist in the future; and
 - define ‘disability’ in the *Workplace Relations Act 1996* (Cth) by reference to the definition of ‘disability’ in the DDA.
- 9-4 The Commonwealth should amend the definition of ‘impairment’ in the regulations made under the HREOC Act to include discrimination on the basis of association with a person who has an impairment or disability.
- 9-5 The States and Territories should consider harmonising their anti-discrimination legislation, and other relevant laws, in a manner consistent with the recommendations in this Report.

Part C. Genetic Testing

11. Regulating Access to Genetic Testing

- 11-1 In order to complement existing pathology laboratory accreditation arrangements, the Commonwealth, States and Territories should enact legislation to require laboratories to: (a) be accredited for any genetic test that they conduct for medical, diagnostic or treatment purposes; and (b) comply with the relevant accreditation standards. The legislation should make provision for exemptions in appropriate circumstances, such as for genetic tests performed by research laboratories.
- 11-2 While the primary focus of laboratory accreditation should remain on matters of technical proficiency and scientific reliability, the National Pathology Accreditation Advisory Council (NPAAC) should continue to develop ethical standards for medical genetic testing, in consultation with the Human Genetics Commission of Australia and the National Health and Medical Research Council.
- 11-3 NPAAC, in consultation with the National Association of Testing Authorities, Australia (NATA) and the Royal College of Pathologists of Australasia (RCPA), should examine how compliance with its accreditation standards in relation to consent, counselling and other ethical considerations in medical genetic testing should be assessed as part of the NATA/RCPA accreditation process.

- 11-4 NATA, in consultation with the RCPA, should develop training programs to equip its officers and peer assessors to verify compliance with NPAAC accreditation standards relating to consent, counselling and other ethical considerations.
- 11-5 The Commonwealth should amend the *Therapeutic Goods Act 1989* (Cth) (*Therapeutic Goods Act*) and regulations made under that Act to enable the Therapeutic Goods Administration (TGA) to regulate more effectively in vitro diagnostic devices used in genetic testing provided directly to the public.
- 11-6 The Commonwealth should amend the *Therapeutic Goods Act* and regulations made under that Act to enable the TGA to regulate DNA identification test kits used in genetic testing provided directly to the public, including for parentage and other kinship testing.
- 11-7 The HGCA should develop codes of practice and advice relating to technical and ethical standards for genetic testing services provided directly to the public, including advice to the TGA or its statutory advisory committees.

12. A New Criminal Offence

- 12-1 The Standing Committee of Attorneys-General should develop a model criminal offence relating to non-consensual genetic testing, for enactment into Commonwealth, state and territory law. Criminal liability should attach to any individual or corporation that, without lawful authority, submits a sample for genetic testing, or conducts genetic testing on a sample, knowing (or recklessly indifferent to the fact) that the individual from whom the sample has been taken did not consent to such testing.

Part D. Human Genetic Research

14. Enforcing Compliance with the National Statement

- 14-1 The National Health and Medical Research Council, as part of its review of the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) in the 2003–2005 triennium, should review the mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.

15. Human Genetic Research and Consent

- 15-1 The National Health and Medical Research Council (NHMRC), as part of its review of the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) in the 2003–2005 triennium, should amend the National Statement to provide that Human Research Ethics Committees (HRECs) must report annually to the Australian Health Ethics Committee (AHEC) with respect to human genetic research proposals for which waiver of

consent has been granted under the National Statement. Until such time as the National Statement has been so amended, the NHMRC should exercise its existing authority to request information from HRECs to require them to report annually to AHEC with respect to such human genetic research proposals.

- 15-2 The NHMRC, as part of its review of the National Statement in the 2003–2005 triennium, should ensure that the provisions of the National Statement relating to waiver of consent and reporting of decisions are consistent with privacy laws and, in particular, with guidelines issued under s 95 and s 95A of the *Privacy Act 1988* (Cth).
- 15-3 The NHMRC, in strengthening the level of training and other support provided to HRECs in accordance with Chapter 17 of this Report, should ensure that adequate attention is given to: (a) the interpretation of the waiver of consent provisions of the National Statement; and (b) HREC decision making in relation to such waiver.
- 15-4 The NHMRC, as part of its review of the National Statement in the 2003–2005 triennium, should amend the National Statement to provide clear guidance to researchers about obtaining consent to unspecified future human genetic research.

16. Encouraging Best Practice in Human Genetic Research

- 16-1 The National Health and Medical Research Council (NHMRC) should develop information and advice for the preparation of human genetic research protocols, including examples and practical guidance on:
- (a) the mechanisms for coding or de-identifying genetic samples and information used in research, and the relative advantages and disadvantages of each approach in different research contexts;
 - (b) the use of independent intermediaries, in appropriate cases, to hold codes linking genetic samples or information with the identifiers;
 - (c) the discharge of legal and ethical obligations to inform research participants about the health implications of testing of genetic samples; and
 - (d) disclosure by researchers to research participants of information about actual or anticipated commercial arrangements connected with human genetic research proposals.
- 16-2 The NHMRC should develop information and advice for preparing consent forms for human genetic research, including examples and practical guidance on such matters as:
- (a) graduated consent options;

- (b) disclosure by researchers about actual or anticipated commercial arrangements;
- (c) ownership or property interests in genetic samples or information;
- (d) methods of protecting the privacy interests of participants; and
- (e) withdrawal of consent by participants.

17. Strengthening Review by HRECs

- 17-1 The National Health and Medical Research Council (NHMRC) should develop and implement procedures to promote consistency, efficiency, transparency and accountability in the review of human genetic research by Human Research Ethics Committees (HRECs). In developing such procedures, the NHMRC should initiate a systematic quality improvement program that addresses:
- (a) consolidation of ethical review by region or subject-matter;
 - (b) the membership of HRECs and, in particular, the balance between institutional and non-institutional members;
 - (c) the need for expertise of HRECs in considering proposals for human genetic research;
 - (d) on-going monitoring of approved human genetic research projects;
 - (e) the education and training of HREC members;
 - (f) payment of HREC members for their work in reviewing research proposals;
 - (g) independent audit of HREC processes; and
 - (h) standardised record keeping and reporting to the NHMRC, including in relation to commercial arrangements.
- 17-2 In the course of developing a quality improvement program for HRECs in accordance with Recommendation 17-1, the NHMRC should review the need for an accreditation system for HRECs in their ethical review of human genetic research.
- 17-3 As part of the process of strengthening HREC review of human genetic research, each relevant institution and organisation should provide adequate resources to enable its HREC to fulfil its institutional responsibilities and achieve the standards set in accordance with Recommendations 17-1 and 17-2.

Part E. Human Genetic Databases

18. Human Genetic Research Databases

- 18-1 The National Health and Medical Research Council (NHMRC), as part of its review of the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) in the 2003–2005 triennium, should amend the National Statement to provide ethical guidance on the establishment, governance and operation of human genetic research databases. The amendments (whether by means of a new chapter or otherwise) should include specific guidance on obtaining consent to unspecified future research. (See also Recommendation 15-4.)
- 18-2 The NHMRC should establish and administer a public register of human genetic research databases. The National Statement, as revised in accordance with Recommendation 18-1, should establish conditions of registration and provide that no genetic research under the National Statement can be conducted using information from a database unless it is duly registered.
- 18-3 The NHMRC, in revising the National Statement in accordance with Recommendation 18-1, should provide guidance on the circumstances in which the use of an independent intermediary is to be a condition of: (a) registration of a human genetic research database; or (b) approval by an Human Research Ethics Committee of research involving a human genetic research database.
- 18-4 The Australian Health Ministers' Advisory Council, in consultation with state and territory Attorney-General's Departments and police services, the Human Genetics Commission of Australia and the NHMRC, should develop nationally consistent rules governing the disclosure, for law enforcement purposes, of genetic samples and information held in human genetic research databases. These rules should provide for disclosure only: (a) with the consent of the sampled person or a person authorised to consent on his or her behalf; or (b) pursuant to a court order.

19. Human Tissue Collections

- 19-1 The Australian Health Ministers' Advisory Council (AHMAC), in consultation with the Human Genetics Commission of Australia (HGCA), the National Health and Medical Research Council (NHMRC) and key professional bodies, should develop nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, newborn screening cards. In particular, and in consultation with state and territory Attorney-General's Departments and police services, AHMAC should develop nationally consistent rules governing disclosure of newborn screening cards for law enforcement purposes. These rules should provide for disclosure only: (a) with the consent of the person sampled or a person authorised to consent on his or her behalf; or (b) pursuant to a court order.

- 19–2 AHMAC, in consultation with the HGCA, the NHMRC and key professional bodies, should review the need for nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, other human tissue collections—including collections of pathology samples and banked tissue.

20. Ownership of Samples and the Human Tissue Acts

- 20–1 The proprietary rights in preserved samples, which are currently enjoyed by hospitals and others under the common law, should continue to be upheld on a case-by-case basis. Legislation should not be enacted to confer full proprietary rights in human genetic samples.
- 20–2 Pending any comprehensive review of relevant laws, the regulation of the collection, storage, access to, or use of genetic samples (whether for the purposes of human genetic research or otherwise) should rely primarily on the *Privacy Act 1988* (Cth) as amended in accordance with the Recommendations in Chapter 8, rather than on amendment of the Human Tissue Acts.

Part F. Health Services

21. Health Professionals and Family Genetic Information

- 21–1 The Commonwealth should amend the *Privacy Act 1988* (Cth) (*Privacy Act*) to permit a health professional to disclose genetic information about his or her patient to a genetic relative of that patient where the disclosure is necessary to lessen or prevent a serious threat to an individual’s life, health or safety, even where the threat is not imminent.
- 21–2 The National Health and Medical Research Council (NHMRC), in consultation with the Office of the Federal Privacy Commissioner, should develop guidelines for health professionals dealing with disclosure of genetic information to the genetic relatives of their patients. The guidelines should address the circumstances in which disclosure to genetic relatives is ethically justified or required, and the need for patients to be counselled about the disclosure of information in these circumstances. The guidelines should be made pursuant to either new provisions of the *Privacy Act* (amended consistently with Recommendation 21–1) or s 7 of the *National Health and Medical Research Council Act 1992* (Cth).
- 21–3 The Commonwealth should amend the *Privacy Act* to provide that an individual has a right to access genetic information about first-degree genetic relatives where such access is necessary to lessen or prevent a serious threat to the individual’s life, health, or safety, even where the threat is not imminent. Where an organisation subject to the *Privacy Act* receives a request for access, the organisation should be obliged to seek consent, where practicable, before determining whether to provide access. The right of access should be exercisable only through a nominated medical practitioner or genetic

counsellor and may be refused where providing access would have an unreasonable impact upon the privacy of the individual whose information is sought or other individuals. (See also Recommendation 8–4.)

- 21–4 In developing the guidelines referred to in Recommendation 21–2, the NHMRC should include advice to health professionals in dealing with requests for access to genetic information by the genetic relatives of their patients.

22. Genetic Registers and Family Genetic Information

- 22–1 Organisations operating genetic registers should seek a Public Interest Determination (PID) under the *Privacy Act 1988* (Cth), where applicable, to ensure that they can continue to collect family medical history information from registrants without breaching the National Privacy Principles. The PID process should review whether any other acts or practices of genetic registers, including those involving the use or disclosure of personal information, may justify exemption under the PID.
- 22–2 The National Health and Medical Research Council (NHMRC) should review the *Guidelines for Genetic Registers and Associated Use of Genetic Material* with particular regard to the de-identification of information. In conducting its review, the NHMRC should ensure that the Guidelines are consistent with privacy laws.

23. Genetic Counselling and Medical Education

- 23–1 As a matter of priority, the Commonwealth, States and Territories should develop strategies to assess and respond to the need for increased and adequately resourced genetic counselling services.
- 23–2 The Commonwealth, States and Territories should examine options for the further development of genetic counselling as a recognised health profession, including the use of certification, accreditation or registration systems for genetic counsellors.
- 23–3 The Human Genetics Commission of Australia (HGCA) should develop genetic testing and counselling practice guidelines, in consultation with the Human Genetics Society of Australasia, state clinical genetics services, and other interested organisations. These guidelines should identify genetic tests, or categories of genetic tests, that require special treatment in relation to procedures for ordering testing and ensuring access to genetic counselling. (See also Recommendation 5–3.)
- 23–4 The HGCA should work with the Australian Medical Council, the Committee of Deans of Australian Medical Schools, and the Committee of Presidents of Medical Colleges to develop an integrated approach to medical education and training in human genetics. This approach should ensure that present and

future medical practitioners are appropriately trained and equipped in clinical genetics and in the use of relevant genetic counselling and genetic services.

- 23–5 The HGCA should work collaboratively with key professional and educational bodies to design and enhance education and training programs aimed at improving genetic health services provided by medical practitioners and other health professionals.

24. Population Genetic Screening

- 24–1 The Australian Health Ministers' Advisory Council, in cooperation with the National Health and Medical Research Council, the Human Genetics Commission of Australia and key professional bodies, should develop national standards in relation to the development and implementation of:
- (a) population genetic screening programs—covering such matters as informed consent, counselling, testing standards, quality assurance, cost-benefit considerations, and reporting and data collection; and
 - (b) newborn screening programs—promoting both universal participation and informed decision making by parents.

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Part G. Insurance

26. Genetic Discrimination in Insurance

- 26–1 As a general matter, there should be no departure from the fundamental principle underlying the market in voluntary, mutually rated insurance, namely, equality of information between the applicant and the insurer. However, where the underwriting of insurance involves the use of human genetic information, the insurance process should be subject to the Recommendations in this Report. (See Chapters 27 and 28).
- 26–2 The Human Genetics Commission of Australia, in consultation with peak industry bodies and regulators, should keep a watching brief on developments in the insurance industry in relation to the use of human genetic information, both in Australia and overseas, with a view to reviewing Australian insurance practices as the need arises.

27. Improving the Underwriting Process

- 27–1 The Human Genetics Commission of Australia (HGCA) should, as a matter of priority, establish procedures to assess and make recommendations on whether particular genetic tests should be used in underwriting mutually rated insurance, having regard to their scientific reliability, actuarial relevance and reasonableness.

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- 27-2 The Investment and Financial Services Association (IFSA) and the Insurance Council of Australia (ICA) should develop mandatory policies for their members to ensure that, once the HGCA has made a recommendation in relation to the use of a particular genetic test in underwriting, that test is used only in conformity with the recommendation. As a transitional arrangement, insurers should be permitted to continue using genetic tests in underwriting in accordance with industry policies, until such time as the HGCA makes a recommendation in relation to those tests.
- 27-3 IFSA and the ICA should require their members to state, on relevant insurance application forms, that not all genetic test results have to be disclosed and that applicants may obtain further information about this from the insurer. In addition, IFSA and the ICA should require their members to provide, upon request, accurate information to applicants in relation to those genetic tests that the HGCA has recommended not be used in underwriting in accordance with Recommendation 27-1.
- 27-4 IFSA and the ICA, in consultation with the HGCA and the Institute of Actuaries of Australia, should develop and publish policies for their members on the use of family medical history for underwriting mutually rated insurance.
- 27-5 The Commonwealth should amend the *Insurance Contracts Act 1984* (Cth) to clarify the nature of the obligation of an insurer to provide written reasons for an unfavourable underwriting decision upon the request of an applicant. Where such a decision is based on genetic information, including family medical history, the insurer should be required to give reasons that are clear and meaningful and that explain the actuarial, statistical or other basis for the decision.
- 27-6 IFSA and the ICA should require their members to inform applicants of their statutory entitlement to reasons for an adverse underwriting decision based on genetic information, including family medical history. IFSA and the ICA should also develop mandatory policies for their members about appropriate mechanisms for providing sensitive information to applicants in response to a request for reasons.
- 27-7 IFSA and the ICA should develop mandatory policies for their members regarding the provision of reasons by an insurer to an applicant following an unfavourable underwriting decision based on family medical history. These policies should ensure that the reasons given are clear and meaningful and that they explain the actuarial, statistical or other basis for the decision.
- 27-8 The Commonwealth should amend the *Disability Discrimination Act 1992* (Cth) and related legislation to clarify the nature of the information required to be disclosed by an insurer to the Human Rights and Equal Opportunity Commission in the course of resolving a complaint. The legislation should ensure that the complainant is entitled to access to the information so disclosed.

- 27-9 IFSA and the ICA should expand the jurisdiction of the Financial Industry Complaints Service Ltd (FICS) and Insurance Enquiries and Complaints Ltd (IEC) to allow those organisations to review underwriting decisions involving the use of genetic information, including family medical history. The amended rules should ensure that the complaint handling processes are:
- timely and efficient;
 - carried out by suitably qualified individuals with a demonstrated understanding of insurance law and anti-discrimination law, underwriting practice, and clinical genetics;
 - binding on the insurer but not on the complainant; and
 - available in respect of a substantial majority of complaints, having regard to the monetary sum in question.
- 27-10 IFSA, the ICA and other relevant bodies should review their policies and practices in relation to training and education of members regarding the collection and use of genetic information in insurance.
- 27-11 The National Finance Industry Training Advisory Body, in consultation with IFSA, the ICA and the HGCA, should review relevant competency standards and the Financial Services Training Package to incorporate an appropriate level of competence regarding the collection and use of genetic information in insurance.

28. Insurance and Genetic Privacy

- 28-1 Insurers should review their consent forms, including medical authority forms, to ensure that they contain sufficient information about the collection, use and disclosure of genetic information to allow applicants to make an informed decision about whether to proceed with their application and consent to the collection of the information. In undertaking this review, insurers should consult with the Human Genetics Commission of Australia and the Office of the Federal Privacy Commissioner.
- 28-2 In reviewing consent and medical authority forms in accordance with Recommendation 28-1, insurers should ensure that consent to collect genetic information for the purpose of assessing an application for insurance is not bundled together with consent for other purposes. The provision of insurance should not be made conditional on the giving of consent to other, unrelated or secondary uses of the genetic information.
- 28-3 Insurers should seek a Public Interest Determination under the *Privacy Act 1988* (Cth) in relation to the practice of collecting genetic information from applicants about their genetic relatives for use in underwriting insurance policies in relation to those applicants.

Part H. Employment

30. Genetic Discrimination in Employment

- 30-1 Employers should not collect or use genetic information in relation to job applicants or employees, except in the limited circumstances where this is consistent with privacy, anti-discrimination, and occupational health and safety legislation, as amended in accordance with the Recommendations in this Report. (See Chapters 31 to 34.)

31. Inherent Requirements of the Job

- 31-1 The Commonwealth should amend the *Disability Discrimination Act 1992* (Cth) (DDA), the *Human Rights and Equal Opportunities Commission Act 1984* (Cth) and the *Workplace Relations Act 1996* (Cth) to provide that, except where it is reasonable to do so, the assessment of an applicant or employee's ability to perform the inherent requirements of a job should not include an assessment of whether he or she will be unable to perform the inherent requirements in the future on the basis of his or her genetic status.
- 31-2 Where genetic information is used to assess an applicant or employee's ability to perform the inherent requirements of a job, employers should develop clearly defined job descriptions that identify these inherent requirements. Employers should also develop policies to ensure that genetic information is used for these purposes only in relevant and reasonable circumstances.
- 31-3 The Commonwealth should amend the DDA to prohibit an employer from requesting or requiring genetic information from a job applicant or employee except where the information is reasonably required for a purpose that does not involve unlawful discrimination, such as ensuring that a person is able to perform the inherent requirements of the job.
- 31-4 The Human Rights and Equal Opportunity Commission, in consultation with the Human Genetics Commission of Australia and other stakeholders, should develop guidelines dealing with the collection and use of genetic information in employment. The Attorney-General should consider the development of Disability Standards in this area pursuant to the DDA.

32. Occupational Health and Safety

- 32-1 The Human Genetics Commission of Australia (HGCA) should establish procedures to assess and make recommendations on whether particular genetic tests should be used in employment for screening for susceptibility to work-related conditions. In assessing particular genetic tests, the HGCA should consider whether:
- there is strong evidence of a clear connection between the working environment and the development of the condition;

- the condition may seriously endanger the health or safety of employees; and
 - the test is a scientifically reliable method of screening for the condition.
- 32–2 The HGCA and the National Occupational Health and Safety Commission (NOHSC) should collaborate with other stakeholders to develop national guidelines for the conduct of genetic screening for susceptibility to work-related conditions. The guidelines should indicate:
- that genetic screening of job applicants and employees for susceptibility to work-related conditions should not be conducted if the danger can be eliminated or significantly reduced by reasonable measures taken by the employer to reduce the environmental risk;
 - that employers should use genetic tests only where they have been recommended for that purpose by the HGCA;
 - how genetic test results are to be interpreted;
 - that screening should not be conducted on a job applicant until the applicant has been made an offer of employment;
 - that screening should be conducted on a voluntary basis except in those rare circumstances in which the HGCA has recommended that screening be mandatory;
 - the circumstances in which family medical history may be collected and used;
 - what provision should be made for genetic counselling of those undergoing testing;
 - appropriate responses by employers where genetic screening reveals relevant susceptibilities; and
 - what measures should be taken to ensure the confidentiality of screening results.
- 32–3 NOHSC should consider adopting the national guidelines on the conduct of genetic screening for susceptibility to work-related conditions as a national code of practice. NOHSC should ensure that the National Priority Action Plans developed under the *National OHS Strategy 2002–2012* reflect these developments.
- 32–4 Within the framework of the National Hazardous Substances Regulatory Package, NOHSC, in consultation with the HGCA and other stakeholders, should develop a national code of practice for the conduct of genetic monitoring of employees exposed to hazardous substances in the workplace. Under this code of practice, genetic monitoring of employees should be conducted only where:

- there is strong evidence of a connection between the working environment and the development of the condition;
 - the condition may seriously endanger the health or safety of employees; and
 - there is a scientifically reliable method of screening for the condition.
- 32–5 The HGCA and NOHSC should collaborate with other stakeholders to develop national guidelines for the collection and use of genetic information from applicants and employees for the protection of third party safety. The guidelines should indicate that genetic information from an applicant or employee should not be collected or used for the protection of third party safety if the danger can be eliminated or significantly reduced by other reasonable measures taken by the employer. Where this is not possible, genetic information should be collected or used only where:
- the applicant or employee’s condition poses a real risk of serious danger to the health or safety of third parties; and
 - there is a scientifically reliable method of screening for the condition.
- 32–6 NOHSC should consider adopting the national guidelines on the collection and use of genetic information for the protection of third party safety as a national code of practice. NOHSC should ensure that the National Priority Action Plans developed under the *National OHS Strategy 2002–2012* reflect these developments.

33. Workers’ Compensation

- 33–1 The Human Genetics Commission of Australia, in consultation with the Heads of Workplace Safety and Compensation Authorities, should develop a policy regarding the appropriate use of genetic information in the assessment of workers’ compensation claims.

34. Employment and Genetic Privacy

- 34–1 The Commonwealth should amend the *Privacy Act 1988 (Cth) (Privacy Act)* to ensure that employee records are subject to the protections of the Act, to the extent that they contain genetic information.
- 34–2 The Commonwealth Attorney-General’s Department and the Department of Employment and Workplace Relations, in their pending inter-departmental review of the employee records exemption, should consider whether the *Privacy Act* should be amended to ensure that employee records are subject to the protections of the Act, to the extent that they contain health information other than genetic information.

Part I. Other Contexts

35. Parentage Testing

- 35-1 The Commonwealth should enact legislation to provide that DNA parentage testing in Australia is conducted only by laboratories accredited by the National Association of Testing Authorities, Australia (NATA), and only in accordance with NATA accreditation requirements.
- 35-2 NATA should review its accreditation requirements for DNA parentage testing to ensure that they meet the highest technical and ethical standards, particularly in relation to consent to testing, protecting the integrity of genetic samples, and providing information about counselling.
- 35-3 The Commonwealth should review Part IIA of the *Family Law Regulations 1984* (Cth) (FL Regulations) to ensure that the requirements for parentage testing meet the highest technical and ethical standards, particularly in relation to consent to testing, protecting the integrity of genetic samples, and providing information as to counselling. In so doing, the Commonwealth should have regard to the accreditation requirements for parentage testing developed by NATA in accordance with Recommendation 35-2.
- 35-4 The Commonwealth should enact legislation to provide that parentage testing reports are not admissible in proceedings under the *Family Law Act 1975* (Cth) (FLA) unless the testing complies with the relevant provisions of the FL Regulations. The States and Territories should consider enacting parallel legislation to ensure that parentage testing reports are not admissible in state or territory proceedings unless the testing complies with NATA accreditation requirements.
- 35-5 NATA should develop accreditation requirements that require laboratories to be satisfied that the sample of each adult donor has been supplied for parentage testing with his or her consent. Provision should also be made for obtaining consent from the deceased's next-of-kin or other authorised person in relation to a sample from a deceased person.
- 35-6 The Commonwealth should amend the FL Regulations to insert a prescribed consent form in relation to parentage testing for each adult donor indicating that the sample has been supplied with his or her consent. Provision should also be made for obtaining consent from the deceased's next-of-kin or other authorised person in relation to a sample from a deceased person.
- 35-7 The Commonwealth should enact legislation to provide that where a child: (a) has attained 12 years of age; and (b) has sufficient maturity to make a free and informed decision, testing of the child's genetic sample can be performed only with the written consent of the child or pursuant to a court order. The child's maturity, and the voluntariness of the child's consent, should be assessed by an independent professional, being a family and child counsellor as defined under the FLA, a social worker or a psychologist.

- 35–8 NATA should develop accreditation requirements to ensure that laboratories conducting DNA parentage tests obtain the written consent of each mature child in accordance with Recommendation 35–7.
- 35–9 The Commonwealth should enact legislation to provide that where a child is:
- (a) under 12 years of age; or
 - (b) 12 years of age or over but less than 18 years of age and does not have sufficient maturity to make a free and informed decision whether to submit a genetic sample for parentage testing;
- such testing can be performed only with the written consent of all persons with parental responsibility for the child, or pursuant to a court order. Where one person with parental responsibility withholds consent or cannot reasonably be contacted, a court should be authorised to make a decision on behalf of the child.
- 35–10 NATA should develop accreditation requirements to ensure that laboratories conducting DNA parentage tests obtain, in relation to each child’s sample, the written consent of all persons with parental responsibility for the child, in accordance with Recommendation 35–9.
- 35–11 NATA should develop accreditation requirements that require laboratories performing DNA parentage tests to inform all persons who provide genetic samples of the availability of counselling, both at the time the samples are submitted for testing and at the time the results are available.
- 35–12 NATA should extend its accreditation program to cover DNA kinship testing other than parentage testing (for example, sibling testing). NATA should apply the requirements for parentage testing, as amended by the Recommendations in this Report, to other kinship testing, in so far as they are applicable.

37. Immigration

- 37–1 The Department of Immigration and Multicultural and Indigenous Affairs (DIMIA) should review its policies and procedures on kinship testing. In particular, the revised policies should ensure that:
- (a) visa applicants are advised at an early stage in the application process that they may be asked to undergo genetic testing to prove an asserted kinship relationship;
 - (b) where DIMIA doubts the veracity of documentary evidence submitted to establish the existence of a kinship relationship, visa applicants should be provided with adequate reasons and given an opportunity to address the doubts by undergoing genetic testing or providing other evidence;

- (c) information in community languages is disseminated to visa applicants about the potential implications of the test and the desirability of seeking counselling;
 - (d) in relation to offshore testing, the panel doctor who takes a sample for kinship testing offers the applicant counselling, or information about the availability of counselling;
 - (e) DIMIA has adequate procedures for preventing identity fraud; and
 - (f) consent is obtained for the disclosure of genetic test results to third parties, including sponsors.
- 37-2 In implementing Recommendation 37-1, policies and procedures for conducting genetic kinship testing for the purpose of migration decision making should be formalised through a Minister's direction made under s 499 of the *Migration Act 1958* (Cth), amendments to the Procedures Advice Manual, or both, as appropriate.
- 37-3 The Department of Health and Ageing, in consultation with DIMIA and the Human Genetics Commission of Australia, should develop policies on genetic tests and the use of genetic information (including family medical history) for the purpose of assessing the health requirement under migration legislation. These policies should include detailed guidelines for Medical Officers of the Commonwealth on the use of genetic information.

38. Sport

- 38-1 The Australian Sports Commission (ASC) should monitor the use of genetic testing and genetic information for identifying or selecting athletes with a view to developing policies and guidelines for sports organisations and athletes. The policies and guidelines should be developed in consultation with the Human Genetics Commission of Australia (HGCA), the Human Rights and Equal Opportunity Commission (HREOC), the Office of the Federal Privacy Commissioner (OFPC), and other stakeholders.
- 38-2 The ASC should develop policies and guidelines for sports organisations and athletes on the use of genetic information in relation to predisposition to sports-related illness or injury. The policies and guidelines should be developed in consultation with the HGCA, HREOC, OFPC and other stakeholders.

Part J. Law Enforcement and Evidence

40. Harmonisation of Forensic Procedures Legislation

- 40-1 In order to facilitate an effective national approach to sharing genetic information for law enforcement purposes, the Commonwealth, States and Territories should collaborate to develop adequate national minimum

standards in Australian forensic procedures legislation with respect to the collection, use, storage, destruction and index matching of forensic material, and the DNA profiles created from such material.

- 40-2 The Commonwealth, States and Territories should not engage in inter-jurisdictional sharing of genetic information—whether on a bilateral basis or through a national DNA database system—unless there is legislation requiring that any information transferred to that jurisdiction will be treated in accordance with the national minimum standards developed under Recommendation 40-1.
- 40-3 In order to facilitate an effective national approach to sharing genetic information the States and Territories should amend their forensic procedures legislation in a manner consistent with the recommendations made in this Report in relation to the *Crimes Act 1914* (Cth).
- 40-4 For the purpose of achieving greater transparency, the Commonwealth, States and Territories should publish all ministerial agreements for sharing genetic information, as well as protocols for inter-jurisdictional matching.

41. Criminal Investigations

- 41-1 The Commonwealth should consider amending the *Crimes Act 1914* (Cth) (*Crimes Act*) to:
- (a) remove the consent provisions in relation to suspects and serious offenders so that a forensic procedure only can be conducted on these persons pursuant to an order made by a judicial officer or an authorised police officer in accordance with the *Crimes Act*; and
 - (b) provide that, once the appropriate authority has made an order for a compulsory forensic procedure, the person who is the subject of the order should be able to choose the method by which the sample is taken.
- 41-2 The Commonwealth should amend the *Crimes Act* to provide that:
- (a) the prescribed information about the nature, purpose and consequences of a forensic procedure should be given to a suspect, serious offender or volunteer in a form that is capable of being easily understood by the person receiving the information;
 - (b) a child or incapable person who is a volunteer, suspect or serious offender should be given the prescribed information in a form that is capable of being easily understood by that child or incapable person, as far as circumstances permit; and

- (c) in addition to information provided to a parent or guardian, the prescribed information also should be given to a child or incapable person who is a volunteer.
- 41-3 The Commonwealth should amend the *Crimes Act* to provide that a forensic procedure may be carried out on a child volunteer of 12 years or more only: (a) with the consent of the child and his or her parent or guardian; or (b) pursuant to a magistrate's order under s 23XWU of the *Crimes Act*.
- 41-4 The Commonwealth should make separate provision for the collection, use, storage, index matching and destruction of forensic material, and profiles obtained from that material, for each main category of volunteer, whether by amending Part 1D of the *Crimes Act* or through regulations.
- 41-5 The Commonwealth should amend the *Crimes Act* to specify that a known victim of crime must be treated as a volunteer, and to require that all reasonable measures be taken to:
- (a) separate the DNA belonging to a victim of crime from a crime scene sample where the latter contains mixed samples;
 - (b) ensure that a victim's DNA profile is not stored in the crime scene index of a DNA database system; and
 - (c) ensure that a victim's DNA profile is not matched against the crime scene index of a DNA database system.
- 41-6 The Commonwealth should develop and publish guidelines for the conduct of mass screening programs in relation to both the process for approving the initiation of programs and the manner in which they are conducted.
- 41-7 The Commonwealth should amend the *Crimes Act*, or regulations made thereunder, to provide that forensic analysis of genetic samples for use by law enforcement authorities should be conducted only by laboratories accredited by National Association of Testing Authorities, Australia (NATA) in the field of forensic science.
- 41-8 The Commonwealth should amend the *Crimes Act* to provide that forensic material obtained pursuant to Part 1D must be destroyed as soon as practicable after a DNA profile has been obtained from the material.
- 41-9 The Commonwealth should amend the *Crimes Act* so that the provisions limiting use and disclosure of information held on a DNA database system also apply to forensic material.
- 41-10 The Commonwealth should amend the *Crimes Act* to define the destruction of forensic material and information obtained from it in terms of physical destruction of samples and permanent and irreversible de-identification of profiles.

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- 41-11 The Commonwealth should amend the *Crimes Act* to assign ultimate responsibility for managing the destruction of forensic material and any information obtained from it.
- 41-12 The Commonwealth should develop formal policies and procedures to:
- (a) enable a volunteer (or parent or guardian) to specify, from a range of options, the retention period for his or her forensic material and any information obtained from it; and
 - (b) establish a process for persons to obtain confirmation that their forensic material, and any information obtained from it, has been destroyed.
- 41-13 The Commonwealth should amend the *Crimes Act* to provide that, with the exception of crime scene samples, law enforcement officers may collect genetic samples only from: (a) the individual concerned, pursuant to Part 1D; or (b) a stored sample, with the consent of the individual concerned (or someone authorised to consent on his or her behalf), or pursuant to a court order.

42. Identification of Deceased Persons

- 42-1 The Commonwealth should amend the *Crimes Act 1914* (Cth) (*Crimes Act*) to delete reference to the DNA profiles of genetic relatives of missing persons from the definition of the ‘missing persons index’.
- 42-2 The Commonwealth, States and Territories should clarify the arrangements under which police officers of one jurisdiction are authorised to act on behalf of another jurisdiction in collecting, using, storing or destroying forensic material from a missing or deceased person (or from a genetic relative of a missing or deceased person).
- 42-3 The Commonwealth should amend Division 11A of Part 1D of the *Crimes Act* to provide that where information stored on a DNA database system is accessed by, or disclosed to, a person for a ‘permitted purpose’, the information may be used only for that purpose.
- 42-4 The Commonwealth should amend s 23YUD of the *Crimes Act*, which regulates inter-jurisdictional sharing, to extend its coverage beyond criminal investigations to include the identification of missing or deceased persons.
- 42-5 Where information stored on a DNA database system is disclosed to Interpol or any foreign agency, the Commonwealth must take reasonable steps to ensure that the information transferred will not be held, used or disclosed by the recipient inconsistently with the national minimum standards established in accordance with Recommendation 40-1.

43. DNA Database Systems

- 43-1 The Commonwealth should amend the *Crimes Act 1914* (Cth) (*Crimes Act*) to provide that forensic material taken from a suspect, and any information obtained from its analysis, must be destroyed as soon as practicable after the person has been eliminated from suspicion, or police investigators have decided not to proceed with a prosecution against that person in relation to that investigation. However, in any event, the forensic material and information must be destroyed no later than: (a) 12 months after the material was taken or the information obtained; or (b) the period stipulated in an order made under s 23YD of the *Crimes Act*.
- 43-2 The Commonwealth should amend the definition of a ‘DNA database system’ in the *Crimes Act* to mean a database (whether in computerised or other form and however described) containing identifiable DNA profiles maintained for law enforcement purposes.
- 43-3 The Commonwealth should expand CrimTrac’s board of management to include independent members, such as nominees of the Office of the Federal Privacy Commissioner and the Commonwealth Ombudsman, legal academics and ethicists.
- 43-4 The Commonwealth should amend the *Crimes Act* to provide for a periodic audit, by an independent body, of the operation of all DNA database systems operating pursuant to the Act. The audit should include the forensic laboratories participating in the DNA database system and the audit report should be made publicly available.
- 43-5 In its annual report to Parliament, the Australian Federal Police should provide information on the number and category of samples obtained pursuant to Part 1D of the *Crimes Act* in that year; the authority under which these samples were obtained; and compliance with the required destruction dates for those samples and profiles.

44. Criminal Proceedings

- 44-1 The National Judicial College of Australia and the Law Council of Australia (through its constituent professional associations) should develop and promote continuing legal education programs for judges and legal practitioners, respectively, in relation to the use of genetic information in criminal proceedings.
- 44-2 In order to provide better guidance for judges and juries, the judiciary should develop a model jury direction for use where DNA evidence has been admitted in criminal proceedings.

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- 44-3 The National Institute of Forensic Science, in consultation with members of the criminal justice and science communities, should provide ongoing guidance to forensic scientists and legal practitioners regarding reliable methods of DNA analysis, statistical calculation, and presentation of evidence in criminal proceedings.
- 44-4 The Commonwealth should amend the *Crimes Act 1914* (Cth) to specify that the prosecution has a duty to provide defendants with reasonable pre-trial notice of all relevant crime scene samples in order to give them an opportunity to have such samples independently analysed.

45. Post-Conviction Use of DNA Evidence

- 45-1 The Commonwealth should amend the *Crimes Act 1914* (Cth) to require the long-term retention of forensic material found at the scene of serious crimes to facilitate post-conviction analysis.
- 45-2 The Commonwealth should establish a process to consider applications for post-conviction review from any person who alleges that DNA evidence may exist that calls his or her conviction into question.

46. Civil Proceedings

- 46-1 The National Judicial College of Australia and the Law Council of Australia (through its constituent professional associations) should develop and promote continuing legal education programs for judges and legal practitioners, respectively, in relation to the use of genetic information in civil proceedings.

Implementation Schedule

This schedule lists the action required of different bodies to implement the recommendations in this Report. The required action is identified in summary form; full details may be found in the List of Recommendations and in the corresponding chapter. The schedule does not list bodies whose role in relation to a recommendation is only to be consulted by another body that has primary responsibility for implementing the recommendation.

Attorney-General (Cth)

- 31-4 Consider the development of Disability Standards dealing with the collection and use of genetic information in employment.

Attorney-General's Department (Cth)

- 34-2 Consider, in the pending inter-departmental review of the employee records exemption, whether the *Privacy Act* should be amended to ensure that employee records are subject to the protections of the Act in so far as they contain health information other than genetic information.

Australian Federal Police

- 43-5 Provide information in the AFP's annual report to Parliament on the number and category of samples obtained pursuant to Part 1D of the *Crimes Act*; the authority under which these samples were obtained; and compliance with the required destruction dates for samples and the profiles obtained from them.

Australian Health Ministers' Advisory Council

- 18-4 Develop nationally consistent rules governing the disclosure, for law enforcement purposes, of genetic samples and information held in human genetic research databases.
- 19-1 Develop nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, newborn screening cards.
- 19-2 Review the need for nationally consistent rules in relation to other human tissue collections.
- 24-1 Develop national standards in relation to the development and implementation of population genetic screening programs and newborn screening programs.

Australian Medical Council

- 23–4 Work with the Committee of Deans of Australian Medical Schools, the Committee of Presidents of Medical Colleges and the HGCA to develop an integrated approach to medical education and training in human genetics.

Australian Sports Commission

- 38–1 Monitor the use of genetic testing and genetic information for identifying or selecting athletes with a view to developing policies and guidelines for sports organisations and athletes.
- 38–2 Develop policies and guidelines for sports organisations and athletes on the use of genetic information in relation to predisposition to sports-related illness or injury.

Committee of Deans of Australian Medical Schools

- 23–4 Work with the Australian Medical Council, the Committee of Presidents of Medical Colleges and the HGCA to develop an integrated approach to medical education and training in human genetics.

Committee of Presidents of Medical Colleges

- 23–4 Work with the Australian Medical Council, the Committee of Deans of Australian Medical Schools and the HGCA to develop an integrated approach to medical education and training in human genetics.

Commonwealth Government

- 5–1 to 5–7 Establish the HGCA, with the powers, functions, structure and composition identified in Recommendations 5–2 to 5–7.
- 5–9 Review the HGCA two and five years after establishment.
- 7–1 Harmonise, in conjunction with the States and Territories, information and health privacy legislation as it relates to human genetic information.
- 7–4 Amend the *Privacy Act* to define ‘health information’ to include all genetic information about an individual in a form which is predictive of health.
- 7–5 Amend the *Privacy Act* to define ‘sensitive information’ to include human genetic test information.
- 7–6 Amend the *Privacy Act* to provide that ‘health information’ includes information about an individual who has been dead for 30 years or less.

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- 7-7 Amend the *Privacy Act* to ensure that all small business operators that hold genetic information are subject to the Act.
- 8-1 to 8-3
Enact legislation that provides legally enforceable privacy standards for the handling of genetic samples, by amending the *Privacy Act* to extend its coverage to identifiable genetic samples.
- 8-4 Amend the *Privacy Act* to provide that individuals have a limited right to access the bodily samples of their first-degree genetic relatives.
- 9-2 Amend the objects clause of the DDA to clarify that the Act applies to discrimination in relation to past, present, possible future or imputed disability, including discrimination on the ground of genetic status.
- 9-3, 9-4 Amend the DDA, *HREOC Act*, and the *Workplace Relations Act* to provide a consistent approach to discrimination on the basis of genetic status and discrimination on the basis of association with another person.
- 11-1 Enact legislation, in conjunction with the States and Territories, to require laboratories that conduct genetic testing to be accredited.
- 11-5, 11-6
Amend the *Therapeutic Goods Act* and regulations made under that Act to enable the TGA to regulate more effectively genetic testing products provided directly to the public, including for parentage and other kinship testing.
- 21-1 Amend the *Privacy Act* to permit a health professional to disclose genetic information about his or her patient to a genetic relative of that patient where the disclosure is necessary to lessen or prevent a serious threat to an individual's life, health or safety, even where the threat is not imminent.
- 21-3 Amend the *Privacy Act* to provide that individuals have a limited right to access genetic information about first-degree genetic relatives.
- 23-1, 23-2
Develop, in conjunction with the States and Territories, strategies to assess and respond to the need for increased and adequately resourced genetic counselling services throughout Australia, and examine options for the further development of genetic counselling as a recognised health profession.
- 27-5 Amend the *Insurance Contracts Act* to clarify that insurers are required to provide clear and meaningful reasons for an unfavourable underwriting decision and to explain the actuarial, statistical or other basis for the decision, upon the request of an applicant.

- 27-8 Amend the DDA to clarify the nature of the information required to be disclosed by an insurer to HREOC in the course of resolving a complaint and to ensure that the complainant is entitled to access to the information so disclosed.
- 31-1 Amend the DDA, the HREOC Act and the *Workplace Relations Act* to provide that the assessment of an applicant or employee's ability to perform the inherent requirements of a job should not include an assessment of whether he or she will be unable to perform the inherent requirements in the future on the basis of his or her genetic status.
- 31-3 Amend the DDA to prohibit an employer from requesting or requiring genetic information from a job applicant or employee except where the information is reasonably required for a purpose that does not involve unlawful discrimination.
- 34-1 Amend the *Privacy Act* to ensure that employee records are subject to the protections of the Act, to the extent that they contain genetic information.
- 35-1 Enact legislation to provide that DNA parentage testing in Australia is conducted only by laboratories accredited by NATA, and only in accordance with NATA accreditation requirements.
- 35-3 Review Part IIA of the *Family Law Regulations* to ensure that the requirements for parentage testing meet the highest technical and ethical standards.
- 35-4 Enact legislation to provide that parentage testing reports are not admissible in relation to proceedings under the *Family Law Act* unless the testing complies with the relevant provisions of the *Family Law Regulations*.
- 35-6 Amend the *Family Law Regulations* to insert a prescribed consent form in relation to parentage testing for each adult donor indicating that the sample has been supplied with his or her consent.
- 35-7 Enact legislation to provide that where a child: (a) has attained 12 years of age; and (b) has sufficient maturity to make a free and informed decision, testing of the child's genetic sample can be performed only with the written consent of the child or pursuant to a court order.
- 35-9 Enact legislation to require that, where a child does not have sufficient maturity to make a free and informed decision whether to submit a genetic sample for parentage testing, such testing can be performed only with the written consent of all persons with parental responsibility for the child, or pursuant to a court order.

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- 40-1, 40-2 Develop, in conjunction with the States and Territories, national minimum standards in Australian forensic procedures legislation with respect to the collection, use, storage, destruction and index matching of forensic material and DNA profiles.
- 40-4 Publish all ministerial agreements for sharing genetic information and protocols for inter-jurisdictional matching.
- 41-1 Consider amending the *Crimes Act* to remove the consent provisions in relation to suspects and serious offenders and provide that, once the appropriate authority has made an order for a compulsory forensic procedure, the person who is the subject of the order should be able to choose the method by which the sample is taken.
- 41-2 Amend the *Crimes Act* to provide that the prescribed information about the nature, purpose and consequences of a forensic procedure should be given to a suspect, serious offender or volunteer in a form that is capable of being easily understood by the person receiving the information.
- 41-3 Amend the *Crimes Act* to provide that a forensic procedure may be carried out on a child volunteer of 12 years or more only: (a) with the consent of the child and his or her parent or guardian, or (b) pursuant to a magistrate's order.
- 41-4 Provide separately for the collection, use, storage, index matching and destruction of forensic material, and profiles obtained from that material, for each main category of volunteer, whether by amending Part 1D of the *Crimes Act* or through regulations.
- 41-5 Amend the *Crimes Act* to specify that a known victim of crime must be treated as a volunteer.
- 41-6 Develop and publish guidelines for the conduct of mass screening programs.
- 41-7 Amend the *Crimes Act*, or regulations made thereunder, to provide that forensic analysis of genetic samples for use by law enforcement authorities should be conducted only by laboratories accredited by NATA.
- 41-8 Amend the *Crimes Act* to provide that forensic material obtained pursuant to Part 1D must be destroyed as soon as practicable.
- 41-9 Amend the *Crimes Act* so that the provisions limiting use and disclosure of information held on a DNA database system apply also to forensic material.
- 41-10 Amend the *Crimes Act* to define the destruction of forensic material and information obtained from it in terms of physical destruction of samples and permanent and irreversible de-identification of profiles.

- 41-11 Amend the *Crimes Act* to assign ultimate responsibility for managing the destruction of forensic material and any information obtained from it.
- 41-12 Develop formal policies and procedures to enable a volunteer to specify the retention period for his or her forensic material and any information obtained from it. Establish a process for persons to obtain confirmation that their forensic material, and any information obtained from it, has been destroyed.
- 41-13 Amend the *Crimes Act* to provide that, with the exception of crime scene samples, law enforcement officers may lawfully collect a genetic sample for law enforcement purposes only from (a) the individual concerned, pursuant to Part 1D of the *Crimes Act*; or (b) a stored sample, with the consent of the person sampled or pursuant to a court order.
- 42-1 Amend the *Crimes Act* to delete reference to the DNA profiles of genetic relatives of missing persons from the definition of the ‘missing persons index’.
- 42-2 Clarify the arrangements under which police officers of one jurisdiction are authorised to act on behalf of another jurisdiction in collecting, using, storing or destroying forensic material from a missing or deceased person.
- 42-3 Amend Division 11A of Part 1D of the *Crimes Act* to provide that where information stored on a DNA database system is accessed or disclosed for a ‘permitted purpose’, the information may be used only for that purpose.
- 42-4 Amend s 23YUD of the *Crimes Act* to extend its coverage beyond criminal investigations to include the identification of missing or deceased persons.
- 42-5 Take reasonable steps, where information stored on a DNA database system is disclosed to a foreign law enforcement agency, Interpol or any other foreign agency, to ensure that the information will not be handled inconsistently with the national minimum standards established in accordance with Recommendation 40-1.
- 43-1 Amend the *Crimes Act* to provide that forensic material taken from a suspect, and any information obtained from its analysis, must be destroyed as soon as practicable after the person has been eliminated from suspicion.
- 43-2 Amend the definition of a ‘DNA database system’ in the *Crimes Act* to mean a database containing identifiable DNA profiles maintained for law enforcement purposes.
- 43-3 Expand the CrimTrac’s board of management to include independent members.
- 43-4 Amend the *Crimes Act* to provide for a periodic audit, by an independent body, of the operation of all DNA database systems operating pursuant to the Act.

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- 44-4 Amend the *Crimes Act* to specify that the prosecution has a duty to provide defendants with reasonable pre-trial notice of all relevant crime scene samples.
- 45-1 Amend the *Crimes Act* to require the long-term retention of forensic material found at the scene of serious crimes.
- 45-2 Establish a process to consider applications for post-conviction review from any person who alleges that DNA evidence may exist that calls his or her conviction into question.

Department of Employment and Workplace Relations (Cth)

- 34-2 Consider, in the pending inter-departmental review of the employee records exemption, whether the *Privacy Act* should be amended to ensure that employee records are subject to the protections of the Act to the extent that they contain health information other than genetic information.

Department of Health and Ageing (Cth)

- 37-3 Develop policies on genetic tests and the use of genetic information, including family medical history, for the purpose of assessing the health requirement under migration legislation.

Department of Immigration and Multicultural and Indigenous Affairs (Cth)

- 37-1, 37-2 Review policies and procedures on kinship testing for immigration decision making purposes and formalise these procedures through a Minister's direction or through amendment to the Procedures Advice Manual, or both, as appropriate.

Employers

- 30-1 Should not collect or use genetic information in relation to job applicants or employees except in the limited circumstances where this is consistent with privacy, anti-discrimination, and occupational health and safety legislation, as amended in accordance with this Report.
- 31-2 Develop clearly defined job descriptions that identify inherent requirements, where genetic information is used to assess an applicant or employee's ability to perform a job, and policies to ensure that genetic information is used for these purposes only in relevant and reasonable circumstances.

Genetic registers

- 22–1 Seek a PID under the *Privacy Act*, where applicable, to ensure that they can continue to collect family medical history information from registrants without breaching the NPPs.

Human Genetics Commission of Australia

- 5–2 When established in accordance with Recommendation 5–1, provide:
- ongoing, high-level, technical and strategic advice to Australian governments about current and emerging issues in human genetics;
 - similar high-level advice on the ethical, legal and social implications arising from these developments, including consideration of any impact on human rights, and analysis of cost-benefit issues;
 - national leadership in managing the process of change in relation to human genetics, including engagement of the public on these issues;
 - relevant expertise and a consultative mechanism for the development of policy statements and national guidelines in this area, where appropriate in association with other government agencies or the relevant industries and organisations;
 - assistance with the development and coordination of community, school, university and professional education about human genetics;
 - advice and a consultative mechanism to assist relevant bodies in identifying strategic priorities for research in human genetics; and
 - a focus for the coordination and integration of various national—and perhaps regional and international—programs and initiatives.
- 5–3 Have a specific role in:
- identifying genetic tests that have particular concerns or sensitivities attached to them, and thus may require special treatment;
 - making recommendations about the suitability of specific genetic tests for use by the insurance industry and by employers;
 - performing any similar function or providing expert advice on any other matters relating to human genetics, whether on its own motion or in response to a formal reference from the responsible minister or ministers; and

- monitoring the implementation of the broad strategies and specific recommendations identified in this Report.

5–6 to 5–8

Operate in an open and transparent manner, to the greatest extent practicable, and liaise closely with relevant government departments and authorities, as well as other key stakeholders, in order to promote a national approach to the protection of human genetic information.

11–7 Develop codes of practice and advice relating to technical and ethical standards for genetic testing services provided directly to the public.

23–3 Develop genetic testing and counselling practice guidelines, which identify genetic tests, or categories of genetic tests, requiring special treatment in relation to procedures for ordering testing and ensuring access to genetic counselling.

23–4 Work with the Australian Medical Council, the Committee of Deans of Australian Medical Schools, and the Committee of Presidents of Medical Colleges to develop an integrated approach to medical education and training in human genetics.

23–5 Work with key professional and educational bodies to design and enhance education and training programs aimed at improving genetic health services provided by medical practitioners and other health professionals.

26–2 Keep a watching brief on developments in the insurance industry in relation to the use of human genetic information, both in Australia and overseas, with a view to reviewing Australian insurance practices as the need arises.

27–1 Establish procedures to assess and make recommendations on whether particular genetic tests should be used in underwriting mutually rated insurance, having regard to their scientific reliability, actuarial relevance and reasonableness.

32–1 Establish procedures to assess and make recommendations on whether particular genetic tests should be used in employment for screening for susceptibility to work-related conditions.

32–2 Develop, in collaboration with NOHSC, national guidelines for the conduct of genetic screening for susceptibility to work-related conditions.

32–5 Develop, in collaboration with NOHSC, national guidelines for the collection and use of genetic information from applicants and employees for the protection of third party safety.

33–1 Develop a policy regarding the appropriate use of genetic information in the assessment of workers' compensation claims.

Human Rights and Equal Opportunity Commission

- 31-4 Develop guidelines dealing with the collection and use of genetic information in employment.

Insurance Council of Australia

- 27-2 Develop mandatory policies for their members to ensure that, once the HGCA has made a recommendation in relation to the use of a particular genetic test in underwriting, that test is used by members in underwriting only in conformity with the recommendations of the HGCA.
- 27-3 Require members to state, on relevant insurance application forms, that not all genetic test results have to be disclosed and that applicants may obtain further information from the insurer.
- 27-4 Develop and publish policies for members on the use of family medical history for underwriting mutually rated insurance.
- 27-6 Require members to inform applicants of their statutory entitlement to reasons for an adverse underwriting decision based on genetic information, including family medical history.
- 27-7 Develop mandatory policies for members regarding the provision of reasons by an insurer to an applicant following an unfavourable underwriting decision based on family medical history. The policies should ensure that the reasons given are clear and meaningful and that they explain the actuarial, statistical or other basis for the decision.
- 27-9 Expand the jurisdiction of the IEC to allow that organisation to review underwriting decisions involving the use of genetic information.
- 27-10 Review ICA policies and practices in relation to training and education of members regarding the collection and use of genetic information in insurance.

Insurers

- 28-1, 28-2 Review consent forms to allow applicants to make informed decisions about whether to proceed with their application and consent to the collection of genetic information.
- 28-3 Seek a PID under the *Privacy Act* in relation to the practice of collecting genetic information from applicants about their genetic relatives for use in underwriting insurance policies in relation to those applicants.

Investment and Financial Services Association

- 27-2 Develop mandatory policies for their members to ensure that, once the HGCA has made a recommendation in relation to the use of a particular genetic test in underwriting, that test is used by members in underwriting only in conformity with the recommendations of the HGCA.
- 27-3 Require members to state, on relevant insurance application forms, that not all genetic test results have to be disclosed and that applicants may obtain further information from the insurer.
- 27-4 Develop and publish policies for members on the use of family medical history for underwriting mutually rated insurance.
- 27-6 Require members to inform applicants of their statutory entitlement to reasons for an adverse underwriting decision based on genetic information, including family medical history.
- 27-7 Develop mandatory policies for members regarding the provision of reasons by an insurer to an applicant following an unfavourable underwriting decision based on family medical history. The policies should ensure that the reasons given are clear and meaningful and that they explain the actuarial, statistical or other basis for the decision.
- 27-9 Expand the jurisdiction of the FICS to allow that organisation to review underwriting decisions involving the use of genetic information.
- 27-10 Review IFSA policies and practices in relation to training and education of members regarding the collection and use of genetic information in insurance.

Judiciary

- 44-2 Develop a model jury direction for use where DNA evidence has been admitted in criminal proceedings.

Law Council of Australia

- 44-1 Develop and promote continuing legal education programs for legal practitioners in relation to the use of genetic information in criminal proceedings.
- 46-1 Develop and promote continuing legal education programs for legal practitioners in relation to the use of genetic information in civil proceedings.

National Association of Testing Authorities, Australia

- 11–4 Develop training programs to equip NATA officers and peer assessors to verify compliance with NPAAC accreditation standards relating to consent, counselling and other ethical considerations.
- 35–2 Review accreditation requirements for DNA parentage testing to ensure that they meet the highest technical and ethical standards.
- 35–5 Develop accreditation requirements that require laboratories to be satisfied that the sample of each adult donor has been supplied for parentage testing with his or her consent.
- 35–8 Develop accreditation requirements to ensure that laboratories conducting DNA parentage tests obtain the required child consent in accordance with Recommendation 35–7.
- 35–10 Develop accreditation requirements to ensure that laboratories conducting DNA parentage tests obtain, in relation to each child’s sample, the written consent of all persons with parental responsibility for the child, in accordance with Recommendation 35–9.
- 35–11 Develop accreditation requirements that require laboratories performing DNA parentage tests to inform all persons who provide genetic samples of the availability of counselling.
- 35–12 Extend the accreditation program to cover DNA kinship testing other than parentage testing and apply the requirements for parentage testing, as amended by the Recommendations in this Report, to other kinship testing, in so far as they are applicable.

National Finance Industry Training Advisory Body

- 27–11 Review relevant competency standards and the Financial Services Training Package to ensure that they incorporate an appropriate level of competence regarding the collection and use of genetic information in insurance.

National Health and Medical Research Council

- 14–1 Review the mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.
- 15–1, 15–2 Amend the National Statement to provide that HRECs must report annually to AHEC with respect to human genetic research proposals for which waiver of consent has been granted under the National Statement and review the National Statement to ensure the provisions of the National Statement

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- relating to waiver of consent and reporting of decisions are consistent with privacy laws.
- 15-3 Ensure that adequate training and other support is given in relation to HREC interpretation of the waiver of consent provisions of the National Statement and decision making in relation to waiver.
- 15-4 Amend the National Statement to provide clear guidance to researchers about obtaining consent to unspecified future human genetic research.
- 16-1 Develop information and advice for the preparation of human genetic research protocols.
- 16-2 Develop information and advice for preparing consent forms for human genetic research, including examples and practical guidance.
- 17-1, 17-2
Develop and implement procedures to promote consistency, efficiency, transparency and accountability in HREC review of human genetic research, through a systematic quality improvement program, and review the need for an accreditation system for HRECs.
- 18-1, 18-2
Amend the National Statement to provide ethical guidance on the establishment, governance and operation of human genetic research databases; and establish and administer a public register of human genetic research databases.
- 18-3 Amend the National Statement to: establish conditions of registration for human genetic research databases; provide that no genetic research can be conducted using information from an unregistered database; and provide guidance on circumstances in which the use of a gene trustee system is required.
- 21-2, 21-4
Develop guidelines for health professionals dealing with the disclosure of genetic information to, and access to genetic information by, the genetic relatives of their patients.
- 22-2 Review the NHMRC Guidelines for Genetic Registers with particular regard to the de-identification of information and consistency with privacy laws.

National Institute of Forensic Science

- 44-3 Provide ongoing guidance to forensic scientists and legal practitioners regarding reliable methods of DNA analysis, statistical calculation, and presentation of evidence in criminal proceedings.

National Judicial College of Australia

- 44-1 Develop and promote continuing legal education programs for judges in relation to the use of genetic information in criminal proceedings.
- 46-1 Develop continuing legal education programs for judges in relation to the use of genetic information in civil proceedings.

National Occupational Health and Safety Commission

- 32-2 Develop, in collaboration with the HGCA, national guidelines for the conduct of genetic screening for susceptibility to work-related conditions.
- 32-3 Consider adopting the national guidelines on the conduct of genetic screening for susceptibility to work-related conditions as a national code of practice.
- 32-4 Develop a national code of practice for the conduct of genetic monitoring of employees exposed to hazardous substances in the workplace.
- 32-5 Develop, in collaboration with the HGCA, national guidelines for the collection and use of genetic information from applicants and employees for the protection of third party safety.
- 32-6 Consider adopting the national guidelines on the collection and use of genetic information for the protection of third party safety as a national code of practice.

National Pathology Accreditation Advisory Council

- 11-2, 11-3
Develop ethical standards for medical genetic testing and examine how compliance with accreditation standards on ethical matters should be assessed as part of the accreditation process.

Research institutions and organisations

- 17-3 Provide adequate resources to enable HRECs to fulfil their institutional responsibilities and achieve standards set in accordance with Recommendations 17-1 and 17-2.

Standing Committee of Attorneys-General

- 12-1 Develop a model criminal offence relating to non-consensual genetic testing, for enactment into Commonwealth, state and territory law.

State and Territory Governments

7-1 to 7-3

Harmonise, in conjunction with the Commonwealth government, information and health privacy legislation as it relates to human genetic information, in a manner consistent with the recommendations in this Report.

8-1

Enact legislation that provides legally enforceable privacy standards for the handling of genetic samples, including in relation to collection, storage, use and transfer.

9-5

Harmonise anti-discrimination legislation, and other relevant laws, in a manner consistent with the recommendations in this Report.

11-1

Enact legislation, in conjunction with the Commonwealth government, to require laboratories that conduct genetic testing to be accredited.

23-1, 23-2

Develop, in conjunction with Commonwealth, strategies to assess and respond to the need for increased and adequately resourced genetic counselling services throughout Australia, and examine options for the further development of genetic counselling as a recognised health profession.

35-4

Consider enacting legislation to provide that parentage testing reports are not admissible in state or territory proceedings unless the testing complies with NATA accreditation requirements.

40-1, 40-2

Develop, in conjunction with the Commonwealth, national minimum standards in Australian forensic procedures legislation with respect to the collection, use, storage, destruction and index matching of forensic material and DNA profiles.

40-3

Amend forensic procedures legislation in a manner consistent with the recommendations made in this Report in relation to the *Crimes Act*.

40-4

Publish all ministerial agreements for sharing genetic information and protocols for inter-jurisdictional matching.

42-2

Clarify the arrangements under which police officers of one jurisdiction are authorised to act on behalf of another jurisdiction in collecting, using, storing or destroying forensic material from a missing or deceased person.

Part A. Introduction

1. Introduction to the Inquiry

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An ALRC–AHEC Joint Inquiry

1.1 In August 2000, the Attorney-General of Australia, the Hon Daryl Williams AM QC MP, and the then Minister for Health and Aged Care, the Hon Dr Michael Wooldridge MP, announced that an inquiry into genetic testing and information would be established, to be conducted jointly by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) of the National Health and Medical Research Council (NHMRC).¹ On 5 February 2001, the same Ministers announced that Terms of Reference had been settled and signed,² signalling the formal start of the joint inquiry (the Inquiry).

1.2 Following representations from the ALRC, AHEC and others, the responsible Ministers approved an extension of time to the final reporting date to 31 March 2003, expressly in order to afford greater opportunities for public participation in the Inquiry’s consultative processes.³

1 Attorney-General and Minister for Health and Aged Care, ‘Gene Technology’, *Joint News Release*, 9 August 2000.

2 Attorney-General and Minister for Health and Aged Care, ‘Genetic Privacy’, *Joint News Release*, 7 February 2001.

3 Daryl Williams AM QC MP, *Correspondence*, 25 January 2002. The original reporting date was 30 June 2002. The Minister for Health and Ageing is now the Hon Senator Kay Patterson.

1.3 The Government's decision to opt for a joint inquiry reflects the wide array of legal and ethical concerns surrounding the field of human genetic samples and information. The ALRC has experience in dealing with legal issues that involve important ethical and social dimensions, and well-tested processes for engaging in effective community consultation.⁴ The Commission has worked on medico-legal issues before—most notably in relation to alcohol, drugs and driving,⁵ human tissue transplants,⁶ and informed consent to medical procedures.⁷ On 17 December 2002, the Attorney-General announced that the ALRC's next inquiry, under fresh terms of reference, would look at issues relating to gene patenting and human health.⁸

1.4 Given the nature of the current Inquiry, it was considered that the addition of the specialist expertise of AHEC would be valuable to the success of the Inquiry. AHEC is a principal committee of the NHMRC, advising the parent body on ethical issues relating to health, and developing guidelines for the proper conduct of medical research involving humans.⁹ The Minister for Health and Ageing has asked AHEC also to play a role in the promotion of community debate on health ethics issues, monitor the work of Human Research Ethics Committees (HRECs), and monitor and advise on international developments in health ethics. AHEC's membership is specified in its establishing legislation, and draws on experts in philosophy, the ethics of medical research, public health and social science research, clinical medical practice and nursing, disability, law, religion and health consumer issues.¹⁰

1.5 Some of AHEC's recent work of particular relevance to this Inquiry includes the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement),¹¹ *Guidelines for Genetic Registers and Associated Genetic Material*,¹² and an information paper *Ethical Aspects of Human Genetic Testing*.¹³ AHEC also has produced a Handbook to assist HRECs in applying the National Statement,¹⁴ and is currently engaged in the process of reviewing the guidelines governing the use of Assisted Reproductive Technologies in Australia, as well as participating in the NHMRC working group developing guidelines for xenotransplantation.

4 The functions of the ALRC are set out in the *Australian Law Reform Commission Act 1996* (Cth) s 21.

5 Australian Law Reform Commission, *Alcohol, Drugs and Driving*, Report 4 (1976), AGPS, Canberra.

6 Australian Law Reform Commission, *Human Tissue Transplants*, Report 7 (1977), AGPS, Canberra.

7 Australian Law Reform Commission, *Informed Decision-Making in Medical Procedures*, Report 50 (1989), AGPS, Canberra.

8 Attorney General and Minister for Health and Ageing, 'Inquiry into Human Genetic Property Issues', *News Release*, 17 December 2002.

9 The functions of AHEC are set out in the *National Health and Medical Research Council Act 1992* (Cth) s 35.

10 *Ibid*, s 36.

11 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

12 National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra.

13 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, NHMRC, <www.nhmrc.gov.au/issues/humangenetics.htm>, 19 February 2003.

14 National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), National Health and Medical Research Council, Canberra.

Advisory committees

1.6 It is standard operating procedure for the ALRC to establish a broad-based, expert Advisory Committee to assist with the development of its inquiries. In this particular case, the Advisory Committee established by the ALRC and AHEC included leaders in the areas of genetic and molecular biological research; medicine; clinical genetics and genetic counselling; community health; indigenous health; health administration and community education; insurance and actuarial practice; and privacy and anti-discrimination laws. A separate Working Group on Law Enforcement and Evidence also was established, with some overlapping membership, including experts on genetic testing, forensic medicine, DNA profiling, policing and trial practice (civil and criminal, defence and prosecution). As always, attention has been paid to achieving a measure of gender, geographical and interest group balance. Full membership details are provided in the List of Participants, above.

1.7 The Advisory Committee and the Working Group met several times during the course of the Inquiry to provide general advice and assistance to the ALRC and AHEC. Such bodies have particular value in helping the Inquiry maintain a clear focus and arrange its priorities, as well as in providing quality assurance in the research and consultation effort, and commenting upon draft materials and reform proposals. However, ultimate responsibility for this Report and its recommendations remains with the relevant Commissioners of the ALRC and Members of AHEC.

1.8 During the course of the Inquiry, the ALRC and AHEC established communications with other bodies that have undertaken parallel projects or research, such as the Ethics Committee of the Human Genome Organisation (HUGO), the Bio-Ethics Committee of UNESCO, the United Kingdom's Human Genetics Commission, the Organisation for Economic Cooperation and Development's Working Party on Information Security and Privacy, the United States Equal Employment Opportunity Commission, and the Ontario (Canada) Ministry of Health and Long-Term Care.

Defining the scope of the Inquiry

The Terms of Reference

1.9 The Terms of Reference for the Inquiry are set out in full above. They ask the ALRC and AHEC to have regard to the broader landscape, including 'the rapid advances in human genetic technology' and 'emerging issues about the control of, ownership of, and intellectual property rights in relation to human genetic samples and information'. The Terms of Reference also acknowledge the breadth of contexts in which the use of genetic information may be relevant, and of potential concern. These include employment; health, including medical research, pharmaceuticals and health administration; insurance and superannuation; and law enforcement.

1.10 The 'action' part of the Terms of Reference specifically asks the ALRC and AHEC to inquire into and report on whether, and to what extent, a regulatory framework is required to:

- protect the privacy of human genetic samples and information;
- provide protection from inappropriate discriminatory use of human genetic samples and information; and
- reflect the balance of ethical considerations relevant to the collection and uses of human genetic samples and information in Australia.

1.11 This is to be done in a way that has regard to the range of Australian ethical opinion on application of human genetic information, as well as the benefits and potential benefits of the scientific and medical applications of the new technology. The Terms of Reference also note the ‘global dimensions of issues relating to research, regulation and the protection of interests’.

1.12 As suggested by the Terms of Reference, the specific drivers for the establishment of the Inquiry were concerns about privacy and discrimination, especially in the contexts of employment and insurance,¹⁵ as well as ethical and other oversight of medical and scientific research, clinical practice, and the collection and use of genetic databases. All of these matters have remained central to the Inquiry’s research and consultation efforts.

Matters outside of this Inquiry

1.13 The Issues Paper (see below) described the manner in which the Inquiry has applied the Terms of Reference in practice, and included a brief discussion of a number of related matters that were considered to be outside of the scope of the present project. These matters include the regulation of genetically modified organisms; access to assisted reproductive technology; embryonic stem cell research and human reproductive cloning; and gene patenting.¹⁶ As noted above, the ALRC will be inquiring into matters relating to gene patenting and human health under fresh terms of reference, with a reporting date of 30 June 2004.¹⁷

Issues Paper 26

1.14 On 15 November 2001, the Inquiry released its first community consultation paper—the 441-page Issues Paper on the *Protection of Human Genetic Information* (IP 26). The primary role of IP 26 was to provide sufficient background material to promote informed community discussion and debate about these new and important

15 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.24]–[2.41].

16 Ibid [1.29]–[1.78]. The ALRC and AHEC wrote to the Attorney-General and the then Minister for Health and Aged Care noting that the gene patenting issue is a matter of considerable importance, and suggesting that this should be the subject of a separate inquiry under fresh terms of reference. In his letter to the ALRC of 11 January 2002, the Attorney-General agreed that this matter is beyond the scope of the current joint Inquiry, and indicated that the government would give consideration to the establishment of a separate inquiry.

17 The Terms of Reference for this Inquiry and related materials may be found on the ALRC’s website: Australian Law Reform Commission, *Gene Patenting*, ALRC, <www.alrc.gov.au/inquiries/current/patenting/index.htm>, 11 March 2003.

issues.¹⁸ To this end, IP 26 posed 68 questions (many of them multi-part questions), to highlight the matters under consideration and to provide a focus for comment and submissions to the Inquiry.

1.15 Nearly 3000 hard copies of IP 26 were distributed within Australia and overseas. The document may be downloaded without charge from the ALRC's website at <www.alrc.gov.au>, and is available in hard copy upon request. Surveys of the ALRC website indicate that IP 26 has been one of the ALRC's most heavily downloaded documents, confirming the very high level of interest in this Inquiry.

Discussion Paper 66

1.16 On 28 August 2002, the Inquiry released a second major community consultation document, this time a 992-page Discussion Paper on the *Protection of Human Genetic Information* (DP 66). The principal differences between DP 66 and IP 26 were that the latter contained the benefits of: (a) further and deeper research; (b) many additional consultations; and (c) the information and views contained in the large number of submissions received (see below). Most importantly, DP 66 replaced the many questions asked in IP 26 with a series of specific proposals for reform (as well as some new questions) for public consideration and comment. Nearly 3000 hard copies of DP 66 were distributed within Australia and overseas. This document also may be downloaded without charge from the ALRC's website at <www.alrc.gov.au>, and is available in hard copy upon request.

Community consultation processes

1.17 One of the most important features of ALRC inquiries is the commitment to public engagement and community consultation. The ALRC-AHEC media release in February 2001, which responded to the Terms of Reference, expressly recognised that widespread public consultation would be a key feature of the genetic information Inquiry. As mentioned above, the reporting deadline for this Inquiry was extended expressly to facilitate and accommodate the high level of public participation. The Inquiry acknowledged from the beginning that while it was essential to become familiar with the latest developments and projected advances in this cutting-edge area of scientific research, in Australia and overseas:

we also recognise that this is an area of broad community interest and concern—so it is equally important that we consult widely and provide all Australians with an opportunity to have their say.¹⁹

18 An executive summary of IP 26 was posted on the ALRC's website, and a short brochure was prepared to provide basic information about major issues, and the means of obtaining further material or participating in the Inquiry. The Inquiry also experimented with a give-away postcard promotion announcing the establishment of the Inquiry and providing contact details for further information.

19 ALRC-AHEC, 'Public Consultation a Priority on Genetic Information', *Media Release*, 7 February 2001. See also B Opeskin, 'Engaging the Public: Community Participation in the Genetic Information Inquiry' (2002) 80 *Reform* 53.

Public forums

1.18 The Inquiry organised a number of well-publicised, and generally well-attended, public forums in November–December 2001 and February–April 2002, in all capital cities and a number of major regional centres, as follows:

- Melbourne, 22 November 2001;
- Hobart, 26 November 2001;
- Perth, 3 December 2001;
- Adelaide, 5 December 2001;
- Brisbane, 12 December 2001;
- Byron Bay/Lismore, 15 December 2001;
- Newcastle, 25 February 2002;
- Canberra, 26 February 2002;
- Wollongong, 4 March 2002;
- Sydney, 12 March 2002;
- Parramatta, 13 March 2002;
- Darwin, 18 March 2002;
- Alice Springs, 19 March 2002;
- Townsville, 3 April 2002; and
- Cairns, 4 April 2002.

1.19 The format of these public forums involved a set presentation from representatives of the ALRC and AHEC, explaining the nature and processes of the Inquiry and highlighting the major issues, followed by comments and questions from the floor. Some meetings also featured a presentation from a leading genetic scientist, where a suitable person was available. Meetings tended to go for a minimum of two hours (the advertised length), and some lasted for up to three hours in order to ensure that everyone in attendance was given an opportunity to be heard.

Other meetings

1.20 Members of the Inquiry also participated in a number of well-attended meetings and workshops, held at the invitation of community, professional and industry organisations or public authorities. These included the Human Genetics Society of Australasia (HGSA), the Committee for the Economic Development of Australia, the Victorian Genetic Services Network, the Institute of Actuaries of Australia, the Anti-Discrimination Board of NSW, the Law Association for Asia and the Pacific, Royal North Shore Hospital Sydney, the University of NSW School of

Community Medicine, Bronte Public School, the Chatswood (NSW) Rotary Club, Southern Cross University's Byron Bay Summer Law School. The Inquiry also participated in a number of seminars and conferences aimed at lawyers and others interested in the regulation of biotechnology.

1.21 The Inquiry also arranged a very large number of targeted meetings with key stakeholders and interested parties. By the end of the process, 185 such meetings had taken place around Australia. These included meetings with:

- clinical genetics services, clinical geneticists, genetic counsellors and genetics educators—including the Queensland Clinical Genetics Service, the Public Health Genetics Working Group Victoria, the Committee of Deans of Australian Medical Schools, the Westmead Hospital Familial Cancer Service, and the Australian Medical Association;
- genetic support groups and disability advocacy groups—including the Genetics Support Council WA;
- organisations concerned with health consumer education and advocacy—including Liberty Victoria, the Australian Consumers Association (CHOICE), and the NSW Council for Civil Liberties;
- organisations concerned with indigenous health, medical and legal issues—including the Aboriginal sub-committee of the HREC of the Menzies School of Health and Research, the Central Australia Aboriginal Congress, the Central Australia Aboriginal Legal Aid Service, Professor Larissa Behrendt (University of Technology, Sydney), Professor Marcia Langton (University of Melbourne), and the Australian Institute of Aboriginal and Torres Strait Islander Studies;
- leading genetic research laboratories—including the Queensland Medical Research Institute, the Murdoch Childrens Research Institute, and the Peter MacCallum Cancer Institute;
- family cancer registers, pathology laboratories, genetic testing laboratories, and institutions holding newborn screening cards—including Genetic Technologies Pty Ltd, Sydney IVF, WA Research Tissue Network, Children's Hospital at Westmead, and NSW Biochemical Genetics & Newborn Screening Services;
- federal, state and territory departments and agencies—including the Attorney-General's Department SA, the Department of Human Services SA, the Australian Securities and Investments Commission, the National Occupational Health and Safety Commission, the Department of Defence, Comcare Australia, the Department of Employment and Work Place Relations, the National Pathology Accreditation Advisory Council, the Queensland Government, the Department of Immigration and Multicultural and Indigenous Affairs (DIMIA), the Office of Gene Technology Regulator, the Therapeutic Goods Administration, and the Australian Health Ministers' Advisory Council Group on Gene Patenting and Genetic Testing;

- peak employer groups, employee groups and trade unions—including the Australian Chamber of Commerce and Industry, the Australian Council of Trade Unions, and the Construction Forestry Mining and Energy Union;
- senior persons involved in the insurance industry—including officers of the Investment and Financial Services Association, the Insurance Council of Australia; the Financial Industry Complaints Service, the Institute of Actuaries of Australia, the Australian Securities and Investments Commission, and the Australian Prudential Regulation Authority;
- federal, state and territory police officers and forensic services units—including the National Institute for Forensic Services, CrimTrac, and the Australian Federal Police;
- organisations concerned with privacy protection—including the Office of the Federal Privacy Commissioner and Privacy NSW; and
- organisations concerned with human rights protection—including the federal Human Rights and Equal Opportunity Commission and the NSW Anti-Discrimination Board.

1.22 Members of the Inquiry also made presentations in 2002 to the Einstein Institute for Science, Health and the Courts (EINSHAC) Conference in Rome (primarily concerned with educating judicial officers about genetic science and the use of expert evidence in this area), the Seventh International HUGO Human Genome Meeting in Shanghai (the major international conference for genetic researchers), the Genetics and Law Conference in London, and the Health and Law Policy Program in Toronto.

1.23 Fourteen meetings were held overseas in Denmark, Sweden, and the Netherlands with representatives from insurers, peak employer and employee bodies, and research ethics committees. After the release of DP 66, meetings were also held:

- in Chicago, with the executive of the National Conference of Commissioners on Uniform State Law;
- in New York, with Professor Dorothy Nelkin (New York University), Dr Thomas Murray (The Hastings Center) and Professor Adrienne Asch (Wellesley College);
- in Washington, DC, with Paul Steven Miller (Commissioner, Equal Employment Opportunity Commission); Associate Professor Kevin FitzGerald (Bioethicist and Oncologist, Georgetown University Medical Center); Barbara Fuller (Senior Policy Analyst, National Human Genome Research Institute); Dr Kathy Hudson, Gail Javitt and Susannah Baruch (Director and Policy Analysts, Genetics and Public Policy Center, Johns Hopkins University); Joanne Hustead (Senior Counsel, Health Privacy Project, Institute for Health Care Research and Policy, Georgetown University); the Hon Justice Gladys Kessler (United States

District Court for the District of Columbia); Professor Karen Rothenberg (Dean, University of Maryland School of Law); Franklin Zweig (President, EINSHAC); Dr Sherrie Hans, Consultant to the Assistant Secretary of Health, and John P Fanning, Privacy Advocate, United States Department of Health and Human Services;

- in Minneapolis, with Dr Jeffrey Kahn (Director, Center for Bioethics, University of Minnesota), and executive members of the National Conference of Commissioners on Uniform State Law;
- in Toronto with Dr Ron Carter (Chair, Ontario Genetics Advisory Committee); Phil Jackson (Director, Strategic Health Policies, Ontario Ministry of Health and Long Term Care); Barbara Slater (Manager, Health Sciences Policy Unit, Ontario Ministry for Health and Long-Term Care); Professor Trudo Lemmens and Professor Colleen Flood (Health and Law Policy Program, University of Toronto); and
- in London with participants in the Genetics and Law Conference; Dr Mark Bale (UK Human Genetic Commissioner); and Professor Martin Partington (Law Commission of England and Wales).

Other means of promoting community engagement

1.24 The ALRC reworked sections of IP 26 in a plain language format for publication in the *Hot Topics* series produced by the State Library of New South Wales' Legal Information Access Centre. *Hot Topics* aims to highlight areas of the law that are subject to change or public debate, with an audience including high school legal studies teachers. *Hot Topic 36: Human Genetic Information* was launched on 10 July 2002 at the State Library of New South Wales, with a panel discussion and question and answer session,²⁰ before an audience of about 100 people.

1.25 There has been strong media interest in the Inquiry from the beginning, and this has been sustained throughout the life of the project. The launch of IP 26 and DP 66 attracted substantial media coverage, including television reports. The public forums also received substantial radio and television coverage in most of the cities in which they were held. The media also have referred to the Inquiry for comment on parallel developments in other jurisdictions, such as the release of papers by the United Kingdom's Human Genetics Commission. As of mid-February 2003, the Inquiry had participated in about 170 separate interviews for radio, television, newspapers or magazines.

20 Moderated by JJJ FM's Adam Spencer, with panellists Professor David Weisbrot of the ALRC, Professor Ron Trent of Royal Prince Alfred Hospital, genetic counsellor Annie Evans, and Kirsten Edwards of the UTS Innocence Project.

1.26 A number of scientific, professional and popular journals, such as *Nature*, *Australian Doctor*, *Australian Biotechnology News* and *HQ* also have provided coverage of particular issues being handled by the Inquiry, as have journals directed at judges, privacy practitioners, lawyers, industrial relations specialists, and insurance industry personnel.

Written submissions

1.27 The Inquiry has strongly encouraged interested persons and organisations to make written submissions to help advance the policy-making process. By mid-July 2002, when DP 66 was being finalised, the Inquiry had received 168 written submissions.²¹ Between the publication of DP 66 and the completion of the manuscript for this Report, another 148 written submissions were received by the Inquiry, bringing the total number of submissions to 316.

1.28 The submissions varied substantially in size and style, ranging from short notes written by individuals or families providing personal views and experiences, to large, well-researched documents prepared by government departments and agencies, research centres and laboratories, genetic support groups, industry bodies and professional associations. It is not merely being polite to state that the Inquiry has found all of the submissions to be very valuable in shaping its views and formulating its final recommendations. Much of the information provided in the institutional submissions probably could be obtained through direct approaches to the relevant bodies. However, the insights and experiences offered in the personal submissions are not readily available elsewhere, and the Inquiry deeply appreciates the time and trouble people have gone to in order to provide these. Some of this material is also deeply personal and sensitive, and in recognition of this the Inquiry left open the option of individuals or groups lodging submissions that were confidential in whole or in part. Of the total of 316 submissions, 34 have been treated as confidential (as requested).

Other assistance

1.29 Apart from those listed above as formal participants, the Inquiry received valuable assistance from a wide variety of individuals and organisations, in Australia and overseas, who provided advice, comments or research materials. The Inquiry expresses special thanks to: Sam Ahlin, Commonwealth Attorney-General's Department; Dr Mary Anderlik, Institute for Bioethics, Health Policy and Law, University of Louisville; Associate Professor Ian Anderson, Melbourne University; John Anderson, CrimTrac; Professor Lori Andrews, Director of the Institute of Science, Law and Technology, Chicago-Kent College of Law; Dr Mark Bale, Secretary, UK Human Genetics Commission; Douglas Barry, Office of the Federal Privacy Commissioner; Lorana Bartels, NSW Attorney-General's Department; Sara

21 Most submissions were received in response to IP 26, but a smaller number of preliminary submissions were received based upon the Terms of Reference and early publicity about the Inquiry. Roughly 200 separate documents were received by the Inquiry, however, where regular correspondents wrote what the Inquiry considered to be 'staged submissions', these were aggregated and treated as a single submission for ease of reference.

Bravini, Immigration Advice and Rights Centre; Julia Cabassi, Anti-Discrimination Board of NSW; Professor Ron Carter, Chair, Ontario Advisory Committee on Genetics; Laurette Chao, Migration Institute of Australia; Helen Cheung, Commonwealth Attorney-General's Department; Alan Clayton; Professor Simon Cole, University of California–Irvine; Anne Deegan, Commonwealth Department of Health and Ageing; Dr Carina Dennis, Australasian Correspondent, *Nature*; Lisa Devereux, Tissue Bank Manager, Peter MacCallum Cancer Institute; Alan Doble, Institute of Actuaries of Australia and Munich Reinsurance; Dr Carl Elliott, Center for Bioethics, University of Minnesota; Stewart Ellis, Comcare; Conor Flanagan, Legal Policy, Victoria Police; Professor Colleen Flood, University of Toronto; Barbara Fuller, National Human Genome Research Institute; John Gaudin, Office of the NSW Privacy Commissioner; Brendan Gogarty, Centre for Law and Genetics, University of Tasmania; Jackie Hickman, DIMIA; Dr Kathy Hudson, Genetics and Public Policy Center, Johns Hopkins University; Eilis Hughes, Genetic Health Services Victoria; Valerie Hurt, National Institutes of Health; Joanne Hustead, Health Privacy Project, Georgetown University; Graeme Innes, Human Rights and Equal Opportunity Commission; Phil Jackson, Director, Strategic Health Policies Branch, Ontario Ministry of Health and Long-Term Care; Professor Richard Johnstone, National Research Centre for Occupational Health and Safety Regulation, the Australian National University; Dr Eric Juengst, Center for Biomedical Ethics, Case Western Reserve University; Dr Josephine Johnston, Department of Bioethics, Dalhousie University; Dr Jeffrey Kahn, Director, Center for Bioethics, University of Minnesota; Dr Kathy King, DIMIA; Richard Konarski, DIMIA; Jocelyn Kula, Therapeutic Goods Administration; Dr Sonia Le Bris, Health Canada; Professor Stephen Leeder, Dean, University of Sydney Medical School; Professor Trudo Lemmens, University of Toronto; Timothy Leshan, National Human Genome Research Institute; Dr Richard Linsk, University of Michigan; Barbara Luby, Office of the Federal Privacy Commissioner; Professor Darryl Macer, Tsukuba University; Professor Angus MacDonald (Heriot-Watt University); Dr John MacMillan, Director, Queensland Clinical Genetics Service; Agnieszka Martin, DIMIA; Dr Nick Martin, Queensland Institute of Medical Research; David Mason, Human Rights and Equal Opportunity Commission; Professor John Mattick, Director, Australian Genome Research Facility; Professor Alexander McCall Smith, University of Edinburgh and Deputy Chair, Human Genetics Commission; Rita Maclachlan, Therapeutic Goods Administration; David Mico, IFSA; Paul Steven Miller, Commissioner, US Equal Employment Opportunity Commission; John Mobbs, CrimTrac; Dr Thomas Murray, Director, the Hastings Center; Professor Roxanne Mykitiuk, Osgoode Hall Law School, York University; Professor Dorothy Nelkin, New York University; Bruce Newey, Centre for Law and Genetics, University of Tasmania; Dr Ainsley Newson, London IDEAS Genetics Knowledge Park; Dr Dianne Nicol, Centre for Law and Genetics, University of Tasmania; Catherine Paice, DIMIA; Maritta Parsell, NATA; Dr Kenneth Pritzker, Pathologist-in-Chief, Mount Sinai Hospital, Toronto; Professor Sandy Raeburn, Centre for Medical Genetics, University of Nottingham; Nicole Rose, NSW Innocence Panel; Professor Karen Rothenberg, Dean, University of Maryland School of Law; Professor Mark Rothstein, Director, Institute for Bioethics, Health Policy and Law, University of Louisville; Professor Mary Segal, University of Pennsylvania School of Medicine; Dr Tom Shakespeare,

Bioscience Centre, University of Newcastle (England); Katy Skinner, DIMIA; Barbara Slater, Manager, Health Sciences Policy Unit, Ontario Ministry for Health and Long-Term Care; Ian Smith, Genetic Technologies Corporation; Meena Sripathy, Immigration Advice and Rights Centre; Dr Alison Stewart, Chief Knowledge Officer, Public Health Genetics Unit, Strangeways Laboratory, Cambridge; Professor Grant Sutherland, Women and Children's Hospital, Adelaide; Geraldine Taylor, DIMIA; Dr Luke Taylor, AIATSIS; Dr Susan Treloar, Queensland Institute of Medical Research; Dr Peter Vodicka, DIMIA; and Professor Dorothy Wertz, University of Massachusetts.

Essentially Yours

1.30 The Inquiry has decided to name this final Report *Essentially Yours*. In part, this reflects the fact that every good pun on the word 'gene' already has been used. More seriously, the Inquiry wished to use a title that clearly signified some of the central tenets of this Inquiry: that fundamental human dignity requires that individuals have a high level of control over their own genetic material, and the information derived from that material; and that human genetic information is personal, sensitive, and deserving of a high level of legal protection—except where there are compelling, countervailing reasons.

1.31 The Inquiry has recognised from the beginning that justice in this complex area is not susceptible to a simple vindication of individual rights. Rather, this is an area in which strong, competing, and even directly conflicting, interests often will arise in practice. In IP 26, the Inquiry noted that:

The major challenge for this inquiry is ... to develop policies to recommend to government which meet the public appreciation of the need to foster innovations in genetic research and practice that serve humanitarian ends, while providing sufficient reassurance to the community that such innovations are subject to proper ethical scrutiny and legal control.

Although relatively easy to articulate, achieving the proper balance is difficult in practice, since various interests will compete and clash across the spectrum of activity.²²

1.32 For example, as discussed throughout this Report, human genetic information also has a strong familial dimension—an individual's genetic information will usually reveal information about, and have implications for, his or her parents, grandparents, siblings, children, and generations to come. Thus, there may be circumstances in which an individual's presumptive right to privacy, and to the confidentiality of the doctor-patient relationship, may be called into question by the competing needs of genetic relatives.

1.33 Similarly, a balance must be struck in a number of other areas—such as in the compulsory acquisition of DNA samples by law enforcement authorities, or in the ability of HRECs to waive individual consent requirements for human genetic

22 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.15]–[2.16].

research—in such a way as to recognise and accommodate broad social interests rather than individual ones. The creative ambiguity inherent in the word ‘essentially’—used in the title *Essentially Yours*—imports the suggestion that it would be misguided to become pre-occupied with absolutes, while ignoring competing interests in this area.

1.34 Finally, there was never any chance that this Report could have been entitled *Essentially You*. In IP 26²³ and DP 66,²⁴ and again in this Report, the Inquiry has made clear its strong opposition to any notion of ‘genetic essentialism’ informing public policy in this area. The idea of genetic essentialism (or sometimes ‘genetic determinism’ or ‘geneticisation’) is that human beings are no more than the sum total of their genes, and thus that ‘genes are everything’. As discussed in further detail in Chapters 3 and 36, such an approach improperly denies the critical importance of environmental (including cultural) factors, and their constant interaction with genetic factors, in shaping human destiny.

The organisation of this Report

1.35 The organisation of this Report largely follows that of DP 66, with the material divided into ten substantial Parts, each of which contains a number of chapters.²⁵ Given the length of this Report, it is hoped that the use of smaller and more targeted chapters will allow readers to identify and focus upon those parts of the Report that most concern them.

1.36 As usual, the prefatory materials contain both an Executive Summary and an aggregated list of the final recommendations, which may be as much as many readers will need or want. For the first time in one of its reports, the ALRC also has included an ‘Implementation Schedule’, in order to highlight the body responsible for implementing each recommendation, with the ultimate aim of facilitating the adoption of the Inquiry’s recommendations.

1.37 Under the *Australian Law Reform Commission Act 1996* (Cth), s 23, reports presented to the Attorney-General must be tabled in Parliament within 15 sitting days, after which they become public documents. All ALRC reports are available on the Commission’s website, at <www.alrc.gov.au>, and may be downloaded without charge. Participants in the Inquiry, including those who have made submissions, will be provided with a copy of this Report; other interested parties may purchase the Report, in hard copy or CD-Rom format, from the ALRC.

23 Ibid [2.134]–[2.142].

24 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [6.105]–[6.119].

25 Some chapters in DP 66 have been split or consolidated, so that chapter numbers in this Report are not necessarily the same as those in DP 66.

2. Genetics and Human Health: A Primer

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2.1 This chapter is intended to serve as a basic primer on genetics and human health, to provide the scientific background for this Report.¹ Among other things, the chapter describes the emerging understanding of genetic science in the wake of the Human Genome Project, including: the nature of DNA, RNA, genes and chromosomes; patterns of inheritance of genetic traits; the interplay among genes and between genetics and the environment; and the implications of the ‘New Genetics’ for human health.

DNA, RNA, genes and chromosomes²

2.2 Every cell in the human body contains a nucleus, with the exception of red blood cells, which lose this structure as they mature. Within the nucleus are tightly coiled threadlike structures known as chromosomes (see Figure 1). Every chromosome has a long arm and a short arm, with a pinch point known as a ‘centromere’. Humans normally have 23 pairs of chromosomes, one member of each pair derived from the mother and one from the father (see Figure 2). One of those pairs consists of the sex chromosomes—with two X chromosomes determining femaleness, and one X and one Y determining maleness. The other 22 chromosomes are known as ‘autosomes’.

1 Chapter 2 of IP 26 also contained a genetics primer to assist public understanding of the issues involved in this Inquiry.

2 This section is drawn from a number of sources including: R Hawley and C Mori, *The Human Genome: A User's Guide* (1999) Harcourt Academic Press, Burlington; M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London; R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone; The Cooperative Research Centre for Discovery of Genes for Common Human Diseases, *GeneCRC*, <www.genecrc.org/>, 18 February 2003; and an ALRC in-house seminar on ‘Introduction to Genetic Testing’, presented by Dr Richard Linsk, University of Michigan, 20 February 2001.

2.3 Each chromosome has within it, arranged end-to-end, hundreds or thousands of genes (see Figure 3),³ each with a specific location, consisting of the inherited genetic material known as DNA. Some chromosomes are significantly larger than others, and some are more densely packed with genes. Under the standard system of identification, scientists have numbered these autosomes from 1–22 in size order (that is, the number of base pairs),⁴ with chromosome 1 being the largest (279 million base pairs, and an estimated 2968 genes).⁵ Of the sex chromosomes, the X (163 million base pairs and an estimated 1184 genes) is similar in size to chromosome 7, while the Y is the smallest chromosome (51 million base pairs and an estimated 231 genes).⁶

2.4 DNA (deoxy-ribo-nucleic acid) is so called because it consists of a large acid molecule mainly found in the nucleus (*nucleic*) to which many sugar groups (*ribo*) that are missing an oxygen molecule (*deoxy*) are attached. DNA contains a code that directs the ‘expression’ or production of proteins, which form much of the structure of the cell and control the chemical reactions within them. The DNA of each gene is characterised by a unique sequence of bases which, when arranged in triplets (codons) in various orders, represent the ‘genetic code’.⁷

2.5 There are many different definitions for a ‘gene’, but one of the most commonly accepted is that a gene contains all of the information required to determine the expression of a specific protein or a chain of amino acids (a ‘polypeptide’). Sometimes a polypeptide can form a complete protein on its own (as in the case of insulin), but in most cases a number of polypeptides combine to create a single functional protein (as in the case of collagen and globin).

2.6 There are four basic building blocks (nucleotides) for DNA: Adenine (A) and Guanine (G), which are known as ‘purines’; and Thymine (T) and Cytosine (C), which are known as ‘pyrimidines’. These nucleotides link together to form long polynucleotide chains (see Figure 4). A DNA molecule consists of two of these chains, linked together by hydrogen bonds, running in opposite directions. Linkage of the chains follows a strict rule, known as ‘complementary base pairing’:

- the base A can only pair with the base T, and vice versa; and
- the base G can only pair with the base C, and vice versa.

3 As noted above, recent work by the Human Genome Project and related research mapping the human genome suggests that human beings have about 30,000–40,000 genes.

4 However, this numbering system contains an historical anomaly: chromosome 22 originally was thought to be smaller than chromosome 21, but it is now known to be slightly larger.

5 See the US Department of Energy’s ‘Gene Gateway’ website: US Department of Energy, *Chromosome FAQs*, <www.ornl.gov/hgmis/posters/chromosome/faqs.html>, 13 February 2003.

6 It is believed that 150–200 million years ago, the X and Y chromosomes were about the same size, but that during evolution the Y chromosome has shrunk at the rate of five genes per million years: see S Jones, *Y: The Descent of Men* (2002) Time Warner Books, London.

7 For an excellent popular account of modern genetics, see M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London.

2.7 There are over three billion of these base pairs of DNA making up the human genome. The two chains link together in a ladder-like shape, twisted into the now famous ‘double helix’ shape first described by Watson and Crick in 1953,⁸ with sugars and phosphates forming the sides or backbone of the ladder and the base pairs forming the rungs (see Figure 5).

2.8 In humans, genes comprise only a small proportion of the DNA in a cell. Up to 98% of DNA consists of ‘non-coding’ regions—popularly, but incorrectly, referred to as ‘junk DNA’—which are full of repeat sequences, pseudogenes and retroviruses. There are no non-coding portions of DNA in bacteria—there are only genes, each one expressing a specific protein. In recent years, genetic scientists have increasingly come to believe that it is non-coding DNA that may be the basis for the complexity and sophistication of the human genome, which permits only 30,000 genes to produce about 200,000 proteins. A leader in this field, Professor John Mattick AO, Director of the Institute for Molecular Biology at the University of Queensland (and a Member of the Australian Health Ethics Committee), has surmised that non-coding DNA forms:

a massive parallel processing system producing secondary signals that integrate and regulate the activity of genes and proteins. In effect, they co-ordinate complex programs involved in the development of complex organisms.⁹

2.9 Proteins are critical components of all cells, determining colour, shape and function. Proteins can have a structural role (such as keratin, from which hair is made), or a functional role in regulating the chemical reactions that occur within each cell (such as the enzymes involved in producing energy for the cell). Proteins are themselves made up of a chain of amino acids. Within the DNA there is a code that determines which amino acids will come together to form that particular protein. The genetic code for each amino acid, consisting of three base sequences, is virtually identical across all living organisms.¹⁰

2.10 Different genes are switched on and off in different cells, leading to different proteins being made or expressed with different structures, appearances and functions—leading to the production of brain cells, nerve cells, blood cells, and so on. Contemporary stem cell research is based around the idea that it should be possible to learn how to use gene switches to coax stem cells into developing into the specialised cell or tissue needed for therapeutic purposes. Research recent has also begun to focus on ‘epigenetic’ changes to the human genome—subtle modifications to the genome that do not alter the DNA sequence, but may play a role in modulating gene

8 James Watson and Francis Crick, building upon work by Linus Pauling and RB Corey, and ‘stimulated by a knowledge of the general nature of the unpublished experimental results and ideas of Dr MHF Wilkins, Dr RE Franklin and their co-workers at King’s College, London’: see J Watson and F Crick, ‘A Structure for Deoxyribose Nucleic Acid’ (1953) 171 *Nature* 737. In 1962 Watson and Crick were awarded the Nobel Prize for this work.

9 G O’Neill, ‘Ghost in the machine’, *The Bulletin*, 11 March 2003, 55.

10 There are 64 different possible codons (given the four letters in the building blocks), and no codon can code for more than one amino acid. As there are only 20 different types of amino acids, some codons must encode the same amino acid. See R Hawley and C Mori, *The Human Genome: A User’s Guide* (1999) Harcourt Academic Press, Burlington, 32.

expression. This may explain, for example, why many diseases appear only later in life, and why one twin may develop a genetic-linked disease while the other does not.¹¹

2.11 When the instructions in a gene are to be read, the DNA comprising that gene unwinds and the two strands separate. An enzyme called RNA polymerase allows a complementary copy of one strand of the DNA to be made. This copy is made from RNA nucleotides, and is called ‘messenger RNA’ (or mRNA) because it serves to carry the coded genetic information to the protein-producing units in the cell, called ribosomes.¹² This process of reading the message in the DNA is called ‘transcription’. On the ribosomes, the amino acids are assembled in the precise order coded for in the mRNA.¹³ The process of converting the message encoded in the RNA (mRNA) to protein using the ribosome is called ‘translation’. When the whole message has been translated, the long chain of amino acids folds itself up into a distinctive shape that depends upon its sequence, and is now known as a ‘protein’ (see Figure 6).¹⁴

Genetic difference: genotype and phenotype

2.12 All humans have the same basic set of about 32,000–35,000 genes, according to the latest best estimates.¹⁵ This is far lower than the early estimates of 200,000, and even the relatively recent estimates of 100,000 used at the start of the Human Genome Project. This figure is similar for the mouse—and, at least for some people, uncomfortably close to the figures for the round worm (19,000), the fruit fly (13,000) and mustard cress (25,000). As has been widely reported, the human genome is more than 98% identical to that of chimpanzees, and 97% identical to that of gorillas.¹⁶

2.13 Genes may come in different versions, known as *alleles*. These alleles arise when there is a change in the ordering of the bases described above—in effect, a ‘typographical error’ in the code, involving the change of a single letter, the inversion of two letters, the deletion or insertion of a phrase (a ‘codon’), or the repetition of a phrase. This change in the sequence may cause no harm (a ‘polymorphism’), or it may make the gene faulty (a ‘mutation’) in the way it directs (expresses) the production of protein.

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- 11 See C Dennis, ‘Altered states’ (2003) 421 *Nature* 686, for a summary of recent research into epigenetics.
- 12 RNA also carries the linear code and employs the same building block letters as DNA, except that it uses U (for uracil) in place of T (for thymine).
- 13 Transfer RNA molecules (tRNA) also play a key role in carrying specific amino acids to the ribosome to be linked to the growing polypeptide or protein.
- 14 See M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 9.
- 15 E Lander, ‘Genomic Information: Driving a Revolution in Bio-Medicine’ (Paper presented at Seventh International Conference of the Human Genome Organisation, Shanghai, 14 April 2002).
- 16 The principal genetic difference is that the other primates have 24 pairs of chromosomes, rather than the 23 pairs that characterise human beings. This appears to be the result of the fusion of two medium-sized ape chromosomes to become human chromosome 2, the second largest of the human chromosomes. Human chromosome 2 is not only the same size as the two ape chromosomes put together, but it also contains the same pattern of bands: M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 24.

2.14 Although any two human beings will be 99.9% genetically identical, the precise DNA sequence of about 6.2 billion letters (3.1 billion base pairs) differs in each person's genetic code. The remaining 0.1% of difference is thought to comprise more than 10 million common single-letter genetic variations (and a larger number of rare variants). The rate of variation is very low in humans (one single nucleotide polymorphism per 1300 bases) compared with other species, including other primates—suggesting a small species with a small 'starter population'.¹⁷

2.15 These facts explain both the striking similarities among all people, which are the result of our common inheritance, and the many individual differences found even within a nuclear family.

2.16 Some genetic variations make little or no difference to health, for example hair colour. However, some mutations do affect basic functioning:

Mutations are permanent and inheritable changes in the ability of a gene to encode its protein. Much like typographical errors, which can change the meaning of a word, or even render a sentence as gibberish, such changes in gene structure can have severe effects on the ability of a gene to encode its protein. Some mutations prevent any protein from being produced, some produce a non-functional or only partially functional protein, and some produce a faulty or poisonous version of the protein.¹⁸

2.17 For example, Huntington's disease (HD) is caused by a mutation to a gene that lies on chromosome 4, in which the triplet 'CAG' repeats an abnormally large number of times. Most people have 10–35 repeats; 40 or more repeats mean that the person will develop HD at some time, with a larger number of repeats leading to earlier and more severe onset. The complete lack of this triplet, together with other mutations, will cause another rare but serious disease, Wolf-Hirschhorn syndrome.¹⁹

2.18 The unique combination of alleles found in a particular individual's genetic make-up is said to constitute that person's *genotype*. The observable physical characteristics of this genotype, as determined by the interaction of both genetic makeup and environmental factors, is said to constitute that person's *phenotype*. This includes such features as eye colour and hair colour, determined genetically,²⁰ as well as height and weight—determined by genetic factors as well as by diet and other environmental influences.

Patterns of inheritance

2.19 Because mutations can affect the functioning and expression of the alleles of genes, resulting in particular traits or characteristics, it is possible to follow the pattern of inheritance of the different alleles of a gene in a family. For most genes, two copies

17 E Lander, 'Genomic Information: Driving a Revolution in Bio-Medicine' (Paper presented at Seventh International Conference of the Human Genome Organisation, Shanghai, 14 April 2002).

18 R Hawley and C Mori, *The Human Genome: A User's Guide* (1999) Harcourt Academic Press, Burlington, 6.

19 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 55.

20 At least initially—hair colour and eye colour now can be modified cosmetically and, of course, hair colour can change naturally over time.

are found in the one individual. If the two copies are the same allele, the individual is said to be *homozygous*. If there are two different alleles for that gene present, the individual is referred to as *heterozygous* for that gene. The exceptions to this are traits coded for by genes that are found on the X chromosome, which are discussed below.

Traits that follow a pattern of recessive inheritance

2.20 As noted above, autosomes are the chromosomes that do not determine sex—in humans, this means all of the chromosomes except for the X and Y. Everyone has two copies of the autosomes and therefore two copies of the genes carried on these chromosomes. A *recessive* trait is one that is expressed only if an individual is homozygous for a mutated copy of that gene—ie, he or she must have two copies of the mutated allele coding for it, one inherited from the mother and one from the father. Two parents who themselves do not express a particular trait nevertheless may have a child with the trait if each parent is a heterozygous *carrier* for the mutated allele—ie, each parent has one copy of the recessive mutated allele and one copy of a normally functioning allele. Where both parents are carriers, each child has a one-in-four chance of inheriting both abnormal alleles and so developing a clinical disorder (see Figure 7).

2.21 To provide a prosaic example, two brown-eyed parents with recessive alleles for blue eyes can produce a blue-eyed child.²¹ Other examples of autosomal recessive (AR) conditions include cystic fibrosis, haemochromatosis, β -thalassaemia, and Tay–Sachs disease (TSD) (see Table 2–1).

Traits that follow a pattern of dominant inheritance

2.22 A dominant trait is one that is manifest when a person has only one mutated allele in a particular gene pair. An affected person may have inherited the mutated allele from either parent or, as the result of a new mutation, may be the first person in the family to have it. The children of persons who have a dominant mutated allele have a one-in-two chance of inheriting that allele and trait (see Figure 8). Examples of autosomal dominant (AD) traits include HD, myotonic dystrophy, hereditary non-polyposis colorectal cancer, Marfan syndrome, familial adenomatous polyposis, and early onset familial Alzheimer’s disease (see Table 2–1, below).

Traits that follow a pattern of X-linked inheritance

2.23 X-linked traits are determined by genes found on the X chromosome. Since males have an X and a Y chromosome, they only have one copy of each of the genes found on the X chromosome and will always express a mutated copy of one of these genes (see Figure 9). Since a woman has two X chromosomes, having a recessive mutated allele on one X chromosome may not cause the trait to be expressed because

21 It should be noted, however, that the development of eye colour is complex and involves more than one gene. For a discussion of the genetics of hair colour, eye colour and other physical characteristics, see R van Oorschot and others, ‘Beyond DNA Databases: Physical Identification Using DNA’ (Paper presented at DNA Evidence: Prosecuting Under the Microscope International Conference, Adelaide, 10 September 2001).

she will have a normally functioning allele on the other X chromosome. X-linked conditions (XL) caused by recessive genes include haemophilia, Fragile X mental retardation and Duchenne muscular dystrophy (see Table 2–1).

The importance of penetrance

2.24 ‘Penetrance’ is the term used to describe the degree of likelihood (based on clinical studies) that an individual carrying a particular genetic trait that *could* cause a disorder *will* actually develop it.²² This can vary from very low to very high. For instance, it is possible to speak of the penetrance of each particular mutation (or combination of mutations) causing cystic fibrosis (CF). For the mutation known as ‘ $\Delta F508$ ’, the penetrance is high—about 99%. For other alleles, the penetrance is lower—but this calculation is also dependent upon the definition of the disease.²³

2.25 Mutation in the so-called ‘breast cancer gene’, BRCA1, which is found in up to 1% of women in certain populations, is another example. Its presence is said to increase the risk of developing breast cancer by a factor of five. However, only 60–85% of women with a BRCA1 mutation will develop breast cancer during their lifetimes (that is, 60–85% penetrance). In other words, 15–40% will not do so.

2.26 HD is an example of a condition with a very high penetrance, approaching 100%. Those who test positive for the HD mutation will almost always develop the disease if they live long enough.²⁴ However, even for HD, some people may develop the disease very late in life, or die of something else before they manifest symptoms.²⁵

2.27 Table 2–1 contains further information about penetrance for a range of medical conditions. However, the figures must be read in context. As discussed above, the penetrance and severity of genetic disorders may not be the same for all persons.

Genetics and human health

2.28 It is now common in reporting about health issues for BRCA1 and BRCA2 to be used as a form of shorthand for ‘breast cancer’. This is highly misleading: *everyone* has the BRCA1 and BRCA2 genes, which in their correct form have a role in suppressing the growth of tumours in breast and ovarian tissue. Increased risk of breast cancer is due to inheriting the *mutated alleles* of these genes (including from the father, it should be noted, contrary to popular myth), which removes their protective capacity.

22 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 47.

23 In the case of cystic fibrosis, for example, clinicians must consider whether male infertility in the absence of any other clinical signs is a ‘condition’, a ‘disease’, or nothing of significance.

24 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 58. See also M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 55–66.

25 There is on-going research to determine the extent to which an enriched environment (mental stimulation, physical activity etc) may delay the onset of HD and other degenerative diseases. See A van Dellen and others, ‘Environmental Effects on Huntington’s Disease in Transgenic Mice’ (2000) 404 *Nature* 721, 721–722, discussed on the ABC Radio program ‘The Science Show’, 15 April 2000: The Science Show, *Huntington’s Disease Study*, Radio National, <www.abc.net.au/rn/science/ss/stories/s117687.htm>, 19 February 2003.

2.29 Ridley has pointed out that the tendency to identify a specific gene as the cause of disease obscures the vital role of genes in human health:

Open any catalogue of the human genome and you will be confronted not with a list of human potentialities, but a list of diseases, mostly named after pairs of obscure central-European doctors. ... The impression given is that genes are there to cause diseases. ...

Yet to define genes by the diseases they cause is about as absurd as defining organs of the body by the diseases they get: livers are there to cause cirrhosis, hearts to cause heart attacks and brains to cause strokes. It is a measure, not of our knowledge but of our ignorance, that this is the way the genome catalogues read. It is literally true that the only thing we know about some genes is that their malfunction causes a particular disease. This is a pitifully small thing to know about a gene, and a terribly misleading one. It leads to the dangerous shorthand that runs as follows: 'X has got the Wolf-Hirschhorn gene'. Wrong. We all have the Wolf-Hirschhorn gene, except, ironically, people who have Wolf-Hirschhorn syndrome. Their sickness is caused by the fact that the gene is missing altogether. In the rest of us the gene is a positive, not a negative force. The sufferers have the mutation, not the gene.²⁶

2.30 Medical conditions or diseases linked to genes can be classified in a number of ways.²⁷ Importantly for this Inquiry these include:

- Monogenic (or single gene) disorders;
- Polygenic (or multi-gene) disorders; and
- Multifactorial disorders.

2.31 In addition, there are chromosomal disorders (such as Down syndrome)²⁸ and somatic cell disorders (such as cancer), in which the genetic abnormality was not present at conception but was acquired during life and is found only in specific cells rather than in all cells in the body.²⁹

2.32 Table 2–1, at the conclusion of this Chapter, lists a number of more common genetic disorders. The table describes the prevalence of the disorder, the mutations involved, the pattern of inheritance, the age of onset, and the opportunities for diagnosis, prevention and treatment.³⁰

26 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 54–55.

27 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 37.

28 *Ibid.*, 69–70.

29 *Ibid.*, 210–211.

30 The inquiry thanks Associate Professor Eric Haan, a member of the Advisory Committee, for the preparation of this Table.

Monogenic disorders

2.33 A monogenic disorder is one in which a mutation in one or both alleles of a single gene is the main factor in causing a genetic disease. Much of our early understanding about genetic influences on health is derived from the observation and study of monogenic disorders such as HD. However, such diseases are relatively rare:

Huntington's disease is at the far end of the spectrum of genetics. It is pure fatalism, undiluted by environmental variability. Good living, good medicine, healthy food, loving families or great riches can do nothing about [it]. Your fate is in your genes.³¹

2.34 The rarity of monogenic disorders is an important consideration in developing a policy framework for the protection of human genetic samples and information. Monogenic disorders allow relatively accurate inferences to be drawn about a person's future health from their current genetic status. However, this is not the case with the vast majority of genetic disorders, where the relationship between genetic status and disease is highly complex and contingent.

Polygenic disorders and haplotyping

2.35 We are increasingly aware that the vast majority of medical conditions with some genetic link involve either the complex interaction of a number of genes (polygenic) or the complex interaction between genes and the environment (multifactorial disorders).³² As Ridley has stated:

Unless you are unlucky enough to have a rare and serious genetic condition, and most of us do not, the impact of genes upon our lives is a gradual, partial, blended sort of thing. You are not tall or a dwarf, like Mendel's pea plants, you are somewhere in between. You are not wrinkled or smooth, but somewhere in between. This comes as no great surprise, because just as we know it is unhelpful to think of water as a lot of little billiard balls called atoms, so it is unhelpful to think of our bodies as the products of single, discrete genes.³³

2.36 According to the Human Genome Database,³⁴ as of 29 December 2002, 14,014 genes have been mapped to individual chromosomes, of which 1,639 have been identified as being involved in a genetic disorder. It may be that most of the simple linkages have already been made, since the rate of discovery has slowed dramatically despite better technology: of the last 3,783 genes to have been mapped, only 17 have been identified with a genetic disorder.

2.37 Following the success of the Human Genome Project, the 'next big thing', according to Dr Francis Collins, director of the United States National Human Genome

31 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 64.

32 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 55, 211.

33 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 66.

34 An international collaboration in support of the Human Genome Project. See the excellent website hosted by the Hospital for Sick Children in Toronto, Canada, which contains regularly updated tables containing details of 'Genetic Disorders by Chromosome', as well as a 'Display Map' to view genetic disorders mapped to a chromosome. See Hospital for Sick Children, *Reports and Statistics*, <www.gdb.org/gdb/report.html>, 18 February 2003.

Research Institute, is to produce a Human Haplotype Map. Haplotypes (or haplotype blocks) refer to a number of closely-linked alleles along a region of a chromosome, which tend to be inherited together.³⁵

2.38 According to the US National Human Genome Research Institute:

The elucidation of the entire human genome has made possible our current effort to develop a haplotype map of the genome. The haplotype map, or 'HapMap', will be a reference work that catalogs the genetic variations of most importance to health and disease.

The DNA sequence of any two people is some 99.9 percent identical. The variations, however, may greatly affect an individual's disease risk. Sites in the DNA sequence where individuals differ at a single DNA base are called single nucleotide polymorphisms (SNPs). Sets of nearby SNPs on the same chromosome are inherited in blocks. This pattern of SNPs on a block is a haplotype. Blocks may contain a large number of SNPs, but a relatively few SNPs can be enough to uniquely identify a haplotype. The HapMap is a map of these haplotype blocks, including the specific SNPs that identify the haplotypes.

The HapMap will enable researchers to quickly compare a patient's genetic patterns with known patterns, and thus determine if that patient is at risk for particular diseases. In addition, people with the same disease may respond differently to the same drug treatments; the HapMap will enable researchers to examine drug efficacy in specific diseases with genetic patterns. Finally, haplotype mapping will reveal the role of variation in individual responses to environmental factors.³⁶

2.39 In theory, there could be large numbers of haplotypes in a chromosome region; however, recent research suggests that there are a smaller number of common haplotypes—perhaps as few as four or five common patterns across all populations—which would permit researchers to shortcut their work dramatically by testing for genetic predispositions for such complex diseases as cancer, diabetes, hypertension, and Alzheimer's block-by-block, rather than letter-by-letter.³⁷ Researchers involved in the work supporting this 'common variant hypothesis' have drawn three important conclusions:

First, the human genome can be objectively parsed into simple haplotype blocks each averaging 11,000 to 22,000 DNA letters but only four or five different variations in the letters. Second, the blocks are similar across individuals from Africa, Europe, and Asia, suggesting that a map of haplotypes will have broad utility for most people.

35 Haplotype and allele frequencies are affected by cellular-level processes as well as by population-level processes: National Human Genome Research Institute, *Developing a Haplotype Map of the Human Genome for Finding Genes Related to Health and Disease*, <www.genome.gov/page.cfm?pageID=10001665>, 20 February 2003.

36 See the National Human Genome Research Institute website: Ibid.

37 See S Gabriel and others, 'The Structure of Haplotype Blocks in the Human Genome' (2002) 296 *Science* 2225; discussed in C Morton, *The Next Big Thing in Mining the Genome: A Simpler Tool for Finding Disease Genes and Spotting Genetic Variation*, Harvard University Focus, <www.focus.hms.harvard.edu/2002/June7_2002/genomics.html>, 21 July 2002.

Third, the haplotype blocks appear to capture about 90 percent of genetic variation in a region of the human genome.³⁸

2.40 According to Mark Daly, one of the authors:

This study is a significant step toward developing a more powerful statistical approach to studying complex human disease. Genetics has not made tremendous inroads in complex disease, even though great effort has been put in during the last 10 to 15 years. As a community, we've come to understand that complex diseases are not caused by single high-penetrance genes, but from more modest risk factors common in populations.

This study provides a great deal of hard data scientists can use to go back and refine and improve models of population biology and molecular evolution. For once, genetics worked out to be easier than it could have been. But now we have to do the disease studies, which will not be simple.³⁹

Multifactorial disorders and the environment

2.41 In the case of multifactorial disorders, inheriting a mutated allele for particular conditions means that the person is susceptible or predisposed to develop the condition. Other factors such as diet or exposure to certain environmental factors are necessary to ensure the expression of the trait or condition. Most of the important and common medical problems in humans are multifactorial, including heart disease, hypertension, psychiatric illness (such as schizophrenia), dementia, diabetes, and cancers.

2.42 For much of the latter part of the last century, the prevailing orthodoxy was that 'nurture' (environment) is far more important than 'nature' (genes) in influencing human development,⁴⁰ at least outside of the basic inherited physical traits. The pace and weight of genetic research in recent times, however, appears to have tipped common wisdom in the other direction—perhaps too far in the direction of genetic exceptionalism and determinism (see Chapter 3).

2.43 In fact, the picture is far more complex. A person is not the sum of a column of traits and behaviours determined by individual genes. Instead, it is better to think of a person as comprising all of the product of his or her genes; the intricate interaction of those genes; and the elaborate interaction between that genetic legacy and environmental factors.

2.44 Even a simple reference to 'the environment' understates the dynamic and multifaceted nature of this relationship. At the most simple level, the quality of the

38 C Morton, *The Next Big Thing in Mining the Genome: A Simpler Tool for Finding Disease Genes and Spotting Genetic Variation*, Harvard University Focus, <www.focus.hms.harvard.edu/2002/June7_2002/genomics.html>, 21 July 2002. However, for a contrary view of the promise of haplotype research, see J Couzin, 'New Mapping Project Splits the Community' (2002) 296 *Science* 1391.

39 C Morton, *The Next Big Thing in Mining the Genome: A Simpler Tool for Finding Disease Genes and Spotting Genetic Variation*, Harvard University Focus, <www.focus.hms.harvard.edu/2002/June7_2002/genomics.html>, 21 July 2002.

40 See S Rose, L Kamin and R Lewontin, *Not in Our Genes: Biology, Ideology and Human Nature* (1985) Pantheon.

environment—a nutritious diet, access to good health care, opportunities for exercise—will allow the full expression of genetically inherited traits, such as height. Over a lifetime, other aspects of the physical environment also will shape human health and development—for example, air and water pollution, endemic disease, workplace safety, drought and war. Choice and chance also play an important role—smoking and skydiving pose dangers to health unrelated to genetic inheritance, and a high speed, head-on car accident will always trump good genes.

2.45 As Ridley has put it:

You had better get used to such indeterminacy. The more we delve into the genome the less fatalistic it will seem. Grey indeterminacy, variable causality and vague predisposition are the hallmarks of the system ... because simplicity piled upon simplicity creates complexity. The genome is as complicated and indeterminate as ordinary life, because it is ordinary life. This should come as a relief. Simple determinism, whether of the genetic or environmental kind, is a depressing prospect for those with a fondness for free will.⁴¹

2.46 The environment is also full of social constructs that affect our well-being and the opportunities to reach our full potential. If a community prohibits women from receiving higher education or bars certain racial groups from employment through discrimination, then inherent intellectual ability will count for little. Similarly, if a community is pre-occupied with idealised (and atypical) body images, then this may contribute to severe eating disorders and ill health in otherwise healthy young women, notwithstanding genes that code for good health.

Disease or protective trait?

2.47 There is a tendency to label many genetic variations as ‘diseases’ or ‘disorders’—but historically, some of these mutations served to *enhance* the prospects of survival in certain environmental contexts. The following examples involve autosomal recessive conditions in which the genetic ‘abnormality’ does not cause significant clinical problems for the carrier (but would do so in a child who inherits affected genes from both parents).⁴²

- *β-thalassaemia* is common in the Mediterranean area and in many parts of Southeast Asia. The genetic defect involves impairment in the synthesis of a protein (β-globin) found in red blood cells. The carrier state, present in as many as 1 in 10 people in some populations, affords protection against malaria because carriers have pale and small red blood cells that do not provide the malaria parasite with a good environment in which to grow. Carriers tend to have very mild anaemia (not enough to cause serious health problems) but the homozygous affected person has severe anaemia, which usually requires life-long blood transfusion.

41 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 75.

42 Information provided by Advisory Committee members Professor Ron Trent and Dr Kristine Barlow-Stewart. See also R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 10–11.

- *Tay–Sachs disease* (TSD) is ten times more common in the Ashkenazi (Central and eastern European) Jewish community than in non-Jews or Sephardic (Middle Eastern) Jews, resulting in a carrier frequency of about 1 in 30 people. It is a neurological degenerative disease that usually results in death by the age of four or five. Carriers of the mutated allele for TSD do not have any symptoms of the condition, but it is thought that the carrier state provided protection against tuberculosis in the cramped conditions of the ghettos in which the Jewish population had to live in times past.
- *Cystic fibrosis* (CF) is common in many ethnic groups but particularly among Caucasians—about one in 25 of whom are carriers of the mutated allele for CF. The defect in CF involves movements of chloride across cells and causes severe problems in lung and pancreatic functions for those with the disease. Those people who are carriers of the mutated allele for CF do not move chloride (ie salt) across their membranes as well as those who are not carriers, and so are at less risk of dying from diarrhoea. Over the many thousands of years of evolution, this would have been a useful mutation to carry when cholera and dysentery were endemic. Carriers generally do not have the symptoms of CF (in fact carrier status only can be determined through a DNA test)⁴³ but a child inheriting the CF mutated allele from both parents may develop severe health problems (although CF is very variable in its severity).
- *Sickle cell anaemia* is caused by a mutation in the haemoglobin gene, and is common among persons from Africa and the Mediterranean area. The carrier state affords protection against malaria, however, because carriers have abnormal red blood cells that die soon after being infected with the malaria parasite, compared with normal red blood cells, which continue to work and to provide an environment in which the malaria parasite can grow. In an evolutionary sense, being a carrier for sickle cell disease is a good thing if one lives in a region in which there is endemic malaria.

2.48 The link between an individual's genetic status and the expression of a genetic disorder, and the link between the expression of a disorder and particular health outcomes, are discussed in detail throughout this Report. In particular, the following chapter examines some of the difficulties in interpreting genetic information in a way that is reliable and relevant for the many contexts in which genetic information is or may be used, now and in the future.

43 However, a man who is a carrier of a mutated allele and has a polymorphism in the other allele may not have outwards symptoms of CF, but may be infertile.

Table 2–1 Genetic disorders and genetic testing

Disorder	Prevalence	Gene(s)	Inheritance	Age of onset	The mutations				Prevention/ Surveillance	Treatment
					% affected families with mutation in this gene(s)	Mutations / mutational mechanism	Common mutations / mutational mechanisms	Penetrance of mutation		
Alzheimer disease — early onset	1:2,500	PS1, PS2, APP	AD	40s-50s	30-40%	Base substitution, deletion, other	Many different mutations	PS1: 100% by 65y PS2: <100% by 80y	None	None
Charcot-Marie-Tooth disease type 1A	1:2,500	PMP22	AD	First two decades	100%	Duplication, base substitution	Duplication: 98%	~100%	None	Physiotherapy, podiatry, modification of physical environment
Cystic fibrosis	1:2,500	CFTR	AR	Birth-first years	100%	Base substitution, deletion, other (over 1000)	ΔF508: 65–75% in Caucasian populations	100% when homozygous	Early diagnosis by newborn screening.	Physiotherapy, antibiotics, diet,
Duchenne muscular dystrophy	1:3,500 boys	Dystrophin	XLR	First years	100%	Deletion, duplication, base substitution	Deletion: 65-70%	100%	None	Physiotherapy, orthotics, mobility aids
Factor V Leiden	Heterozygotes 1:30 Homozygotes 1:4,000	Factor V	AD	Very variable	100% (Factor V Leiden is a form of Factor V with a specific mutation)	Base substitution	100%	Heterozygotes: relative risk 3-4 for venous thrombosis Homozygotes: relative risk 80-100 for venous thrombosis	Prevention of thrombosis in those with phenotypic resistance to activated protein C	Standard treatment of thrombosis

Disorder	Prevalence	Gene(s)	Inheritance	Age of onset	The mutations				Prevention/ Surveillance	Treatment
					% affected families with mutation in this gene(s)	Mutations / mutational mechanism	Common mutations / mutational mechanisms	Penetrance of mutation		
Familial adenomatous polyposis (FAP)	1:3,500 <1% of all colorectal cancer	APC	AD	Polyps from teens, colon cancer from 20s	100%	Base substitution, deletion, other	Many different mutations	100% for polyps and colorectal cancer in families with classical FAP. <100% for 'attenuated FAP'	Surveillance by sigmoidoscopy from 10-15 years. Colectomy once polyps appear.	Standard treatment if cancer develops
Familial breast cancer	5-10% of all breast and ovarian cancer	BRCA1, BRCA2 0.1–0.2% of women in the general population and 1–2% of women of Jewish descent have a mutation in one of these genes.	AD	Very variable 20s–80s	Up to 50% of families meeting specific criteria for familial breast cancer	Base substitution, deletion, other	Many different mutations	60–85% for breast cancer, 30-60% for ovarian cancer	Breast examination, mammography, prophylactic mastectomy or oophorectomy, ovarian cancer surveillance	Standard treatment if cancer develops
Fragile X mental retardation	1:4,000 boys 1:2,000 girls	FMR1	XL	Birth	100%	Triplet repeat expansion	CCG triplet repeat expansion: 100%	100% in males and 60% in females for copy number >230	None	Educational and behavioural support

Disorder	Prevalence	Gene(s)	Inheritance	Age of onset	The mutations				Prevention/ Surveillance	Treatment
					% affected families with mutation in this gene(s)	Mutations / mutational mechanism	Common mutations / mutational mechanisms	Penetrance of mutation		
Haemochromatosis	>1:600	HFE	AR	From ~ 30s	>95%	Base substitution	Homozygosity for C282Y in >90%	Estimated to be 50–60% when homozygous	Surveillance for iron overload	Venesection
Haemophilia A	1:10,000 boys	Factor 8 C	XL	First months when severe	100%	Inversion, base substitution, deletion and insertion	Inversion intron 22: 45% of severe haemophilia A	100%	Prophylaxis with Factor 8	Factor 8
Hereditary non-polyposis colorectal cancer (HNPCC)	5-10% of all colorectal cancer	MLH1 MSH2 MSH6 PMS1 PMS2	AD	Very variable 20s-80s	Up to 60% of families meeting the definition of HNPCC	Base substitution, deletion, other	Many different mutations	70-90% for any cancer. In men: colorectal cancer 80-90%. In women, colorectal cancer 30-80%, endometrial cancer 40%, ovarian cancer 10-20%	Colonoscopy, colectomy once cancer develops, endometrial and ovarian cancer surveillance	Standard treatment if cancer develops
Huntington's disease	1:20,000	IT15	AD	Variable but mostly 35–55y	100%	Triplet repeat expansion	CAG triplet repeat expansion: 100%	36-39 copies: <100% >39 copies: 100%	None	Supportive

Disorder	Prevalence	Gene(s)	Inheritance	Age of onset	The mutations				Prevention/ Surveillance	Treatment
					% affected families with mutation in this gene(s)	Mutations / mutational mechanism	Common mutations / mutational mechanisms	Penetrance of mutation		
Myotonic dystrophy	1:20,000	DMPK	AD	Very variable. Birth to 80s	100%	Triplet repeat expansion	CAG triplet repeat expansion: 100%	50-150: mild effects 100-1,000: classical adult onset form 500->2,000: congenital form	Surveillance for complications	Treatment of complications
β -thalassaemia	Depends on ethnic background and control programs. Heterozygote frequency as high as 1 in 10 in some populations	β -globin	AR	First months	100%	Base substitution, deletion, other	Many different mutations	100% when homozygous	None	Blood transfusion, iron chelation
Werdnig-Hoffmann disease	1:10,000	SMN	AR	First 6 months	100%	Deletion, base substitution	Deletions: 98%	100% when homozygous	None	Supportive

3. Coming to Terms with Genetic Information

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3.1 Chapter 2 provided a basic primer on genetics and human health, which described the emerging understanding of genetic science in the wake of the Human Genome Project. This chapter considers the nature of the genetic information that may be obtained about a person as a result of these emerging technologies. In particular, the chapter considers the ways in which genetic information is similar to, and different from, other kinds of personal information, and the policy implications of this.

What is ‘genetic information’?

Apparent or inferential information

3.2 In one sense, almost all information about a person’s health and physical well-being can be called ‘genetic information’. A casual glance reveals information about a person’s gender, race, height, weight, and other features that are related, in whole or in part, to that person’s genetic inheritance.

3.3 Doctors and insurers have been using general family medical histories for over 100 years to draw inferences about the present and future health of individuals. Medical and health practitioners were making clinical observations about genetic conditions long before the technology was developed to test directly for such conditions. Similarly, information that a person has high blood pressure, high cholesterol levels, diabetes, or cancer may also provide information about that person’s genetic inheritance.

Genetic testing and genetic information

3.4 In Chapter 10, the Inquiry notes that there are a number of types of scientific tests that may reveal genetic information. Most famously, these now include tests that amplify selected segments of a person's DNA or RNA using polymerase chain reaction technology, and then analyse the targeted gene sequences in search of 'markers' known to be associated with particular genetic traits, conditions or disorders.

3.5 However, other biochemical tests of non-genetic substances (for example, ordinary blood tests for cholesterol) as well as some medical imaging processes may provide strong indicators of particular genetic disorders, particularly in combination with other tests or clinical observations.

3.6 For the same reasons that the Inquiry is reluctant to adopt a hard and fast definition of 'genetic test',¹ we are likewise reluctant to specify a precise or exhaustive definition of 'genetic information'.² Instead, the Inquiry's strong preference is to consider the context to determine whether the use of genetic-related information requires any special handling or protection.

The primacy of context

3.7 As technology continues to progress, and DNA testing becomes cheaper, quicker, more accurate and much more prevalent, there will inevitably be more pressure placed on institutional and individual safeguards that are intended to protect privacy, prevent discrimination and uphold ethical best practice. Consequently, many of the recommendations made in this Report relate to 'genetic information' gained from DNA (or related) testing, or tissue samples which may be subject to such testing.

3.8 However, this is not invariably the case, and the ambit of the Inquiry must vary according to the circumstances—especially in accordance with the potential mischief that is being addressed.

3.9 For example, the relatively inexpensive heel prick tests—performed on virtually all newborns in Australia—involves the measurement of proteins in the blood spots collected to detect such genetic-linked disorders as phenylketonuria, galactosemia, congenital hypothyroidism, cystic fibrosis and a number of rare metabolic disorders. Newborn screening tests do not involve DNA analysis; nevertheless there are some important issues for this Inquiry in relation to consent to such testing. Even more importantly, newborn screening cards contain blood samples from which genetic information later may be drawn, and the mass testing program over the past 30 years potentially has resulted in a very large, if disorganised, national genetic database.³

1 See Ch 10.

2 The UK Human Genetics Commission also prefers this approach. See Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 33.

3 See Pt E.

3.10 Similarly, nuchal translucency is an imaging procedure that is sometimes performed on a human foetus in utero, to determine the likelihood that the baby will have the genetic condition of Down syndrome. Although nuchal translucency does not involve any DNA testing or analysis, the procedure does provide important genetic information—which must be presented to the parents accompanied by appropriate genetic counselling and support.⁴

3.11 In the context of risk-rating for insurance purposes, an insurer may wish to collect and use ‘old-fashioned’ genetic information, in the form of family medical history, as well as information drawn from DNA tests. Sound policy and practice must be developed in relation to both sources.⁵ Employers may be interested in whether a worker has a predisposition to a genetic-linked disease or disorder that may be triggered by agents (for example, chemicals or dusts) found in the workplace.⁶

3.12 Law enforcement officials may be concerned to secure a ‘DNA profile’—a genetic ‘fingerprint’—for use in identifying an individual.⁷ The use of DNA for personal identification is also at the centre of parentage testing.⁸ By way of contrast, scientific researchers generally work with de-identified (or anonymised) samples, since they are seeking trends and correlations across broader populations, rather than conclusions about known individuals.⁹ Similarly, epidemiologists and public health officials are concerned with detecting broad trends and have little use for individual details.

3.13 Even in the context of the clinical uses, the sensitivities surrounding genetic information can vary greatly with the circumstances.¹⁰ For example, genetic information can relate to a condition that is clinically apparent—such as when a genetic test is performed to confirm a diagnosis with respect to someone who has already exhibited signs or symptoms of a particular disorder. In these circumstances, genetic tests are not distinctly different in nature from other forms of diagnostic testing (such as blood tests, MRI or CAT-scans), but they may be more accurate and convenient. For example, it may be preferable to use a genetic test to diagnose cystic fibrosis in an infant than to use a less accurate sweat test; a genetic test for haemochromatosis is far less difficult and painful for the patient than a liver biopsy.¹¹

3.14 Genetic information also can relate to a condition that is latent—such as when a genetic test is performed on someone who is apparently free of a disorder, in order to predict the likelihood that he or she will, or may, develop the disorder in the

4 Pt F. See also G Parasivam, *Submission G140*, 19 March 2002; Australian Federation of Right to Life Association, *Submission G082*, 17 January 2002.

5 Pt G.

6 Pt H.

7 Pt J.

8 Pt I.

9 Pt D.

10 Pt F.

11 See Human Genetics Society of Australasia, *Submission 6 to Senate Legal and Constitutional Legislation Committee Inquiry into the Provisions of the Genetic Privacy and Non-Discrimination Bill 1998*, 11 May 1998.

future, or may be a carrier for the disease or disorder. As noted in Chapter 10, such ‘predictive testing’ is called ‘presymptomatic testing’ where an individual’s family medical history suggests that he or she may have the genetic disorder, but the symptoms have not yet become manifest.

3.15 Predictive tests raise greater ethical and social concerns than testing conducted for immediate clinical reasons,¹² requiring among other things:

- careful thought about whether testing ought to be performed where no treatment is available, or where the patient is a child;
- much more care in interpretation, both by health professionals and the individuals concerned;
- considerably more attention to collateral uses, and the possibility of breaches of privacy or unfair discrimination; and
- the provision of adequate pre- and post-test counselling and support services.

Is genetic information special?

3.16 A central concern for the Inquiry is whether genetic information is so fundamentally different from other forms of personal health information that it requires special regimes to regulate its collection, use and disclosure. Three aspects of genetic information call for special comment: it is ubiquitous, familial and often predictive.

The ubiquity dimension

3.17 Information about a person’s identity and genetic status can be gathered from ‘the tiny bits of genetic material we scatter around us without much thought’, such as ‘the cells mixed in our saliva and the bulbs at the base of the hairs we continuously shed’.¹³ This is what makes genetic information a potent force for police investigations, where ‘the saliva on a licked postage stamp can help solve a major crime’.¹⁴

3.18 Every cell in a person’s body, with the exception of sex cells and mature blood cells, contains all of his or her genetic code. Unlike other forms of personal health data, a person’s genetic code is not transitory—genetic information lasts for life. The testing of any biological sample any time can thus reveal the full complement of a person’s genetic information.

12 C Jamieson, *Genetic Testing for Late Onset Diseases: In-depth Thematic Analysis of Policy and Jurisdictional Issues*, Health Canada, <www.hc-sc.gc.ca/iacb-dgiac/arad-draa/english/rmdd/wpapers/thematic.pdf>, 20 February 2003.

13 T Murray, ‘Genetic Exceptionalism and “Future Diaries”: Is Genetic Information Different From Other Medical Information?’ in M Rothstein (ed), *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) Yale University Press, New Haven, 60.

14 Ibid.

3.19 There is also potential for stored genetic samples to be re-tested as new tests are developed, or as our understanding of genetic conditions advances. Thus, genetic information is unusual to the extent that it may be obtained by testing material that is readily available—indeed, virtually ubiquitous—without the knowledge or consent of the person in question.

3.20 While there appears to be a community consensus in favour of the use of genetic material for identification in the law enforcement context, there would be no such comfort in the thought that hairs, or saliva taken from a glass, or mucous or drops of blood from a discarded handkerchief, might be subjected to DNA analysis by an employer, an insurer, a government official, a journalist or a medical researcher, without that person's knowledge or consent, or some other lawful authority.

3.21 This Report considers whether, and to what extent, protective mechanisms need to be put into place in each of the various contexts to avoid the misuse of genetic information. Parts of this Report also address specific concerns about taking and submitting another person's tissue samples for DNA analysis, without that person's consent or some other lawful authority.¹⁵

The familial dimension

3.22 Each person's genetic information is unique,¹⁶ but it can also reveal information about, and therefore have implications for, that person's blood relatives, including those in preceding and succeeding generations. Although some genetic mutations arise spontaneously and can thus be said to be truly individual, most genetic information flows from 'before the cradle to after the grave'. Sometimes these implications may even extend beyond the family to larger groups of closely-linked people with common ancestry, for example, indigenous, ethnic or ethno-religious communities.

3.23 Moving in the other direction, it may be possible to draw inferences about the genetic information of an individual who belongs to a family or a group, if information is already known about other members of the family or group. Similarly, genetic information is capable of revealing 'family secrets', including information about parentage (for example, non-paternity or misattributed parentage), adoption, or the use of artificial reproductive technology.

3.24 Demonstrating that an individual is a carrier of a mutated allele for cystic fibrosis means that one of that person's biological parents is also a carrier, and that his or her siblings may be affected or may also be carriers.¹⁷

3.25 The familial nature of genetic information poses certain ethical questions and challenges, both for individuals and families, as well as for those persons and institutions that handle this information, such as medical practitioners, scientific

15 See Ch 12, Ch 35.

16 Even 'identical twins' have minute differences in their genetic code.

17 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.75]–[2.86] regarding patterns of inheritance.

researchers, hospitals, family cancer registers, and others. As sensitive health information, an instinctive reaction is to provide a high level of privacy protection for genetic information. However, to the extent that genetic information has a familial dimension, it can be argued that it is ‘shared’ information, with other family members having rights—or at least interests—in information that may have implications for their own health.¹⁸ Precisely because genetic information is familial in nature, much of it will come as no surprise; indeed, it can often provide great relief to those who receive the data. It is relatively rare that individuals learn of a risk through genetic testing that they did not already anticipate.

3.26 At the same time, some family members may wish to assert a ‘right not to know’ the results of a test taken by a family member to determine the presence or absence of a serious genetic disorder, such as Huntington’s disease, preferring to organise their lives without the shadow of such information.

3.27 As with so many of the issues considered in this Report, resolving such tensions is not easy. It is not a matter of simply vindicating individual rights since the core of the problem is that while each individual’s position may be perfectly understandable, the competing positions must ultimately be subject to some sort of test that carefully balances individual, familial and societal interests.

The predictive dimension

3.28 Other questions for this Inquiry arise from the fact that, until now, individuals and society have not had to deal with predictive information of such quantity. There is no single community view about access to and use of predictive genetic information by family members and people or organisations outside the family.

3.29 Information generated by DNA testing can be very precise, indicating whether a particular allele or mutation is or is not present.¹⁹ However, as discussed in Chapter 2, this precision will often prove unhelpful when it comes to predicting future health. Genetic information tends to be about possibilities rather than certainties, because only a proportion of those people with a particular disease-related mutation will go on to develop the disorder.

3.30 There are greater pressures to discover, gain access to and use genetic information than is the case for traditional health information. Its predictive nature makes it of particular interest in situations where information about a person’s future, even though imprecise, could be incorporated into decision making by the individual or by others, such as employers, insurance companies or public health authorities.

3.31 On the one hand, genetic information has the potential to empower people to make better choices about health and medical care for themselves and their families.²⁰ On the other hand, there are growing concerns that predictive genetic information and

18 Disclosure to genetic relatives is discussed at length in Ch 21.

19 Assuming that scientific research already has confirmed the particular ‘markers’ that are being tested for.

20 See G Parasivam, *Submission G140*, 19 March 2002.

its implications may be misunderstood or misapplied by others who are permitted access to the information, resulting in adverse consequences for the person concerned.

3.32 This may be true even within the context of health care. Genetic support groups, for example, have related instances to the Inquiry in which genetic test information or the nature of a particular genetic condition is poorly understood or poorly communicated by medical practitioners. The Commonwealth Department of Health and Ageing submitted that:

There is ... an identified need for general practitioners to be better educated about specific issues relating to genetic testing and the handling of genetic information. A needs assessment among Victorian general practitioners ... has identified that current knowledge of genetics among general practitioners is poor and that education and training is needed in technical, clinical and counselling aspects of genetic testing as well as ethical and privacy of information issues.²¹

3.33 In the way of the modern world, people take the name of the genetic condition they believe they may have (based on family history, or a genetic test result, or perhaps even a self-assessment based upon apparent symptoms), type this into an Internet search engine, download a great deal of technical information—and probably scare themselves witless.

3.34 A fascinating literature has emerged in recent years suggesting that ‘even among highly educated [people], the ability to solve basic numeracy problems is, on average, relatively poor’.²² Research studies demonstrate that even well-educated people have considerable difficulty in understanding or weighing risk or opportunity when presented with figures about relative probability. For example, participants strongly tend to choose a 9-in-100 chance (that is, 9%) of winning a gamble over a 1-in-10 chance (10%).²³ Similarly, participants rated cancer as riskier when described as ‘kills 1,286 out of 10,000 people’ (that is, 12.86% risk of mortality) than when described as ‘kills 24.14 out of 100 people’ (24.14%).²⁴ It appears that people intuitively react more to the large raw numbers than to the relative percentages. This even extends to the consideration of visual cues:

People assess quantity or probability by using the numerosity of the stimulus object as a judgmental clue. In their size-estimation task, a circle was judged as bigger when it was displayed as an array of numerous pieces in a pizza-slice shape.²⁵

21 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

22 I Lipkus, G Samsa and B Rimer, ‘General Performance on a Numeracy Scale Among Highly Educated Samples’ (2001) 21 *Medical Decision Making* 37, 37–44.

23 V Denes-Raj and S Epstein, ‘Conflict Between Intuitive and Rational Processing: When People Behave Against Their Better Judgment’ (1994) 66 *Journal of Personality and Social Psychology* 819, 819–829.

24 K Yamagishi, ‘When a 12.86% Mortality is More Dangerous than 24.14%: Implications for Risk Communication’ (1997) 11 *Applied Cognitive Psychology* 495, 495–506.

25 *Ibid.*, 504. Other studies agree that ‘The traditional use of proportions to express risk in genetic counselling lacks scientific basis. Rates were easier to understand than proportions, regardless of respondents’ age, language and education’: D Grimes and G Snively, ‘Patients’ Understanding of Medical Risks: Implications for Genetic Counselling’ (1999) 93 *Obstetrics & Gynecology* 910, 910.

3.35 Community and professional education can minimise misunderstanding of and over-reaction to genetic information. This will require further research and clear thinking about how best to communicate information about risk and probability in clinical settings and genetic counselling,²⁶ as well as in other contexts in which use might be permitted in certain circumstances, such as insurance, employment and forensic contexts.²⁷

3.36 The worst result for Australians would be to allow genetic information to be used in such a way as to stereotype people about their future ability to function and the probability that disease will occur, rather than relying on evidence of actual disease and ability.²⁸ This would create the real risk of establishing a new ‘genetic underclass’ of people who are fit and able, but are locked out of securing insurance, employment or access to other goods, services and entitlements.

3.37 The Commonwealth Department of Health and Ageing has submitted that:

Instances of discrimination based on an individual’s genetic information are known to have occurred. To some degree, such discrimination appears to be due to a significant overestimation of the reliability and predictive capacity of genetic information and to limited knowledge about the interaction between genetic and other environmental factors. These issues need to be actively addressed.²⁹

3.38 In respect of insurance, the Anti-Discrimination Board of NSW submitted that:

Assessing the degree of risk on the basis of genetic information is by no means clear cut. The nature of the information varies significantly depending upon factors such as whether the information indicates a predisposition to a disorder that is dominant or recessive and the fact that the degree of symptom expression and time of onset will vary between individuals. These factors will influence the relevance of predictive genetic information when applied to risk rating for insurance purposes.³⁰

3.39 In respect of employment, the Australian Council of Trade Unions submitted that:

26 See D Grimes and G Snively, ‘Patients’ Understanding of Medical Risks: Implications for Genetic Counselling’ (1999) 93 *Obstetrics & Gynecology* 910, 910–914; I Lipkus and others, ‘Relationships Among Breast Cancer Perceived Absolute Risk, Comparative Risk, and Worries’ (2000) 9 *Cancer Epidemiology, Biomarkers & Prevention* 973, 973–975; B Biasecker and J Garber, ‘Testing and Counselling Adults for Heritable Cancer Risk’ (1995) 17 *Journal of the National Cancer Institute Monographs* 115, 115–118; A Edwards and others, ‘Presenting Risk Information — A Review of the Effects of “Framing” and Other Manipulations on Patient Outcomes’ (2001) 6 *Journal of Health Communications* 61, 61–82.

27 See Parts G, H, J.

28 E Draper, ‘The Screening of America: The Social and Legal Framework of Employers’ Use of Genetic Information’ (1999) 20 *Berkeley Journal of Employment & Labour Law* 286, 290–291.

29 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002, 3.

30 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002. See also M Otlowski, *Submission G159*, 24 April 2002.

If genetic testing was allowed, it is certain that it would be misused. First, it would be used to screen out employees who might have only a slightly higher predisposition to acquire a condition than the general population. Many employers would not distinguish between a predisposition and a certainty, while the possibility of false negatives and positives would not necessarily be taken into account.³¹

3.40 Yamagishi also has pointed out that the inability to make proper risk assessments is not merely a problem for individual patients, their families and health care professionals, there is also a broader social dimension. Public officials and policy-makers often make judgments about spending priorities and the systemic use of resources to meet certain risks—while failing to give proper regard to evidence that deploying scarce resources to meet other, far more probable, risks would be more effective and efficient in terms of reducing harm overall.³²

Is genetic information truly ‘exceptional’?

Competing philosophies

3.41 The literature about public policy making in respect of genetic information, especially in the United States, has tended to feature two polarised schools of thought. Those who favour ‘genetic exceptionalism’ view genetic information as uniquely powerful and posing special threats to privacy and discrimination that mandate dedicated and higher levels of legal protection. By way of contrast, the ‘genetic inclusivists’ argue that genetic information is just one of a number of sources of personal health and medical information, and there is no need for any higher or special protections.

3.42 Professors Annas, Glantz and Roche of the Boston University School of Public Health, the authors of the influential US Model Genetic Privacy Act,³³ have been the main proponents of genetic exceptionalism.

To the extent that we accord special status to our genes and what they reveal, genetic information is uniquely powerful and uniquely personal, and thus merits unique privacy protection.³⁴

3.43 Annas, Glantz and Roche offer three justifications for this view, along the lines discussed above. First, a person’s DNA ‘can predict an individual’s likely medical future for a variety of conditions’. Indeed, they argue that one’s DNA is a

31 Australian Council of Trade Unions, *Submission G037*, 14 January 2002.

32 K Yamagishi, ‘When a 12.86% Mortality is More Dangerous than 24.14%: Implications for Risk Communication’ (1997) 11 *Applied Cognitive Psychology* 495, 495–496, 505.

33 This Model Bill strongly influenced the Genetic Privacy and Non-discrimination Bill 1998, introduced into the Australian Parliament by Australian Democrats Senator Natasha Stott Despoja in March 1998. See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.27]–[2.29] for a discussion of this Bill, and its consideration by a Senate committee. For the committee report, see Senate Legal and Constitutional Legislation Committee, *Provisions of the Genetic Privacy and Non-discrimination Bill 1998*, The Parliament of Australia, <www.aph.gov.au/senate/committee/legcon_ctte/genetic/index.htm>, 21 August 2002, 1. The Bill was restored to the Senate *Notice Paper* on 14 May 2002.

34 G Annas, L Glantz and P Roche, ‘Drafting the Genetic Privacy Act: Science, Policy and Practical Considerations’ (1995) 23 *Journal of Law, Medicine and Ethics* 360, 365.

[c]oded probabilistic future diary because it describes an important part of a person's unique future and, as such, can affect and undermine an individual's view of his/her life's possibilities. Unlike ordinary diaries that are created by the writer, the information contained in one's DNA, which is stable and can be stored for long periods of time, is in code and is largely unknown to the person. Most of the code cannot now be broken, but parts are being deciphered almost daily.³⁵

3.44 Second, genetic information about an individual also 'divulges personal information about one's parents, siblings, and children'.³⁶ Third, there is a legitimate concern about the possibility of genetic discrimination, since there is a history of genetics being used to stigmatise and victimise.

3.45 However, there also are strong arguments that genetic information is not fundamentally different from other sorts of health and medical information, and therefore does not invariably merit special treatment. Dr Thomas Murray³⁷ has suggested that much of the drive behind genetic exceptionalism is based upon a generalised image of genetic information as 'a mysterious, powerful and inexorable force that will dominate and control our futures'.³⁸

3.46 Another leading American expert in this area, Professor Lawrence Gostin,³⁹ once argued that there are 'compelling justifications' for special privacy protection for genetic information, grounded in

the sheer breadth of information discoverable; the potential to unlock secrets that are currently unknown about the person; the unique quality of the information enabling certain identification of the individual; the stability of DNA rendering distant future applications possible; and the generalizability of the data to families, genetically related communities, and ethnic and racial populations.⁴⁰

3.47 However, Gostin has since come to the conclusion that:

Genetic exceptionalism is flawed for two reasons: (1) strict protections of autonomy, privacy and equal treatment of persons with genetic conditions threaten the accomplishment of public goods; and (2) there is no clear demarcation separating genetic data from other health data; other health data deserve protections in a national health information infrastructure.⁴¹

3.48 On this view, genetic information is neither distinctive nor unique in its ability to predict an individual's future health, but indicates only a rough range of probabilities. Information about lifestyle (smoker or non-smoker, skydiver or racing car driver, miner or office worker) and non-genetic test results (for example, for

35 Ibid, 360.

36 Ibid.

37 President of the Hastings Center for Bioethics, in New York; formerly Chair of the Human Genome Project's ELSI Task Force on Genetic Information and Insurance.

38 T Murray, 'Genetic Exceptionalism and "Future Diaries": Is Genetic Information Different From Other Medical Information?' in M Rothstein (ed), *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) Yale University Press, New Haven, 60, 64.

39 Professor of Law, Georgetown University and Professor of Public Health, Johns Hopkins University.

40 L Gostin, 'Genetic Privacy' (1995) 23 *Journal of Law, Medicine and Ethics* 320, 326.

41 L Gostin and J Hodge, Jr, 'Genetic Privacy and the Law: An End to Genetic Exceptionalism' (1999) 40 *Jurimetrics* 21.

hepatitis or HIV/AIDS) also provide important clues to present and future health. Murray notes that many other factors and forms of health information ‘afford equally interesting predictions’ and ‘have implications for future health that are every bit as cogent and sensitive as genetic predispositions’.⁴²

3.49 Similarly, other non-genetic test results will contain very sensitive personal information, with the potential to cause distress, discrimination and stigma—such as a positive result for HIV/AIDS, tuberculosis, Hepatitis or a sexually transmitted disease. Murray notes:

Again, genetics is not alone. Institutions and individuals can and have used all sorts of information, both visible and occult, as the basis for discrimination. ... Perhaps what really frightens and galls us about discrimination on the basis of genetic information is its reliance on information about us over which we have no control and may not even know ourselves. Here again it is the hidden and mysterious nature of genetic information, joined with its aura of power and ubiquity, lurking close beneath the surface of our discomfort.⁴³

3.50 Acceptance of genetic exceptionalism, Murray argues, would in practice require health systems to adopt a ‘two bucket theory of disease’, categorising and tossing every disease and risk factor into either the ‘genetic’ or the ‘non-genetic’ bucket—whereas ‘many diseases and risks don’t fit neatly into either bucket’.⁴⁴

3.51 In rejecting genetic exceptionalism, Murray writes that his Task Force on genetics and insurance ultimately concluded:

There was no good moral justification for treating genetic information, genetic diseases, or genetic risk factors as categorically different from other medical information, diseases or risk factors. ... Our need for health care in most cases will be the product of a complex mix of factors, genetic and non-genetic, both within the scope of our responsibility and outside of that scope. The distinction between genetic and non-genetic factors is not the crucial one.⁴⁵

3.52 As a general matter, consultations and submissions to this Inquiry supported this view. The Commonwealth Department of Health and Ageing understood genetic information as part of a continuum of personal and health information, which would be difficult to ‘quarantine’ and subject to a different set of rules in the health sector.⁴⁶

3.53 Similarly, health law expert Dr Roger Magnusson submitted:

Genetic information is likely to become, in future, an integral part of each patient’s individualised health care program. In my view, it is reasonable to assume that, over time, genetic information will become ‘smeared across’—so to speak—the individual’s clinical record, since it will be a relevant component of health status in

42 T Murray, ‘Genetic Exceptionalism and “Future Diaries”: Is Genetic Information Different From Other Medical Information?’ in M Rothstein (ed), *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) Yale University Press, New Haven, 60, 64.

43 Ibid, 60.

44 Ibid, 67.

45 Ibid, 71.

46 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

many areas. Whether or not this information is or should be regarded as especially sensitive will depend, just like non-genetic information, on what it is.

The future role of genetic information within clinical care provides an important and pragmatic argument against genetic exceptionalism. Ultimately, there will be little point in seeking to compartmentalise and quarantine genetic health information behind additional privacy or security barriers. A generic solution to the privacy challenges of genetic health information is preferable.⁴⁷

3.54 By contrast, some submissions supported the exceptional nature of genetic information. The New South Wales Nurses' Association submitted that genetic information:

is more powerful than other forms of health information. As such it requires special legal protection and other exceptional measures ... Our position is based upon a clear potential for abuse by significant decision makers—employers, insurers and public authorities.⁴⁸

3.55 Ian Turnbull criticised the approach identified by the Inquiry in DP 66:

The Inquiry's acceptance of the anti-exceptionalist position ... is fundamentally flawed in that the concern and interest and need for the inquiry itself deny that position. It denies the untapped resource and probable rapid advances that will flow from the Human Genome Project and, unfortunately, from the reported gestations that are supposed to provide the first human clones. It is the easy option and non-predictive approach and reflects why the law always lags behind advancing technologies. In my view the main reason that genetic information is unique is because every single piece contains to some predictable extent information about third parties which those third parties may not even know. The modern advances go to improving the predictability, turning probabilities into certainties.⁴⁹

Towards a contextual approach

3.56 The University of Michigan's Life Sciences, Values and Society Program has noted that:

Throughout the early 1990s many state legislatures pursued an exceptionalist approach, however as the nation moved into the 21st century state legislatures nationwide seemed to be moving towards the middle ground.⁵⁰

3.57 This trend is also reflected in the Inquiry's approach to these issues. There is little value in choosing between opposing schools of thought for the purpose of defining our own philosophy about the nature of genetic information. The Inquiry accepts enough of the inclusivist or anti-exceptionalist argument to believe that it would be a mistake to deal in isolation with the issues surrounding genetic information through a single piece of dedicated legislation—for example, a Genetic Privacy,

47 R Magnusson, *Submission G039*, 10 January 2002; Queensland Government, *Submission G161*, 16 May 2002.

48 New South Wales Nurses' Association, *Submission G090*, 21 January 2002.

49 I Turnbull, *Submission G277*, 21 December 2002.

50 Life Sciences Values and Society Program (University of Michigan), *Genetic Testing*, <www.lifesciences.umich.edu/pdf/GeneTest.pdf>, 20 February 2003.

Discrimination and Research Act. To do so would unfairly privilege genetic information as against all other forms of relevant health and medical information—so that, for example, a person suffering from a genetically linked cancer is ‘in’, but someone suffering a cancer that is not (currently) known to be genetically linked is ‘out’. Equally importantly, such an approach would divorce genetic information from the principles, processes and institutions that have been developed over time to provide ethical oversight of research, ensure best practice in clinical medicine, protect personal privacy, and prohibit unlawful discrimination.

3.58 The Inquiry certainly does accept that there are some special features and issues attaching to genetic information such that it is necessary to engage in a thorough inspection of the existing legal, ethical and regulatory landscape to ensure that they are adequate to the task.

3.59 Magnusson has suggested that there may be less of an exceptionalist case for dealing with genetic information than for HIV/AIDS information:

Genetic exceptionalism has a prominent precedent in HIV/AIDS, a life-threatening infection that emerged in an environment of widespread ignorance and prejudice. Gradually, over a period of about 10 years, Australian legislatures enacted a body of AIDS-specific laws in most jurisdictions.⁵¹ On the other hand, unlike information relating to HIV/AIDS status, there is nothing discrete or self-contained about genetic information as a form of health information. As clinical genetics continues to develop, any attempt to compartmentalise genetic health data from other forms of health information will likely become unworkable. This is because—as the clinical implications of the genetic determinants of disease come to be better understood—genetic testing, and the volume of clinical genetic information, will both increase. Genetics-specific privacy laws will only contribute yet another layer of legal complexity (and health privacy law is already bewilderingly complex), while constituting a major irritant to health providers themselves.⁵²

3.60 The Centre for Law and Genetics also recommended that the Inquiry

pursue the ‘integrated’ approach for the following reasons:

- the issues raised in the protection of human genetic information cover a wide range of activities from research to establish medical practice;
- regulation in this range of areas has traditionally centred on professional codes of ethical practice;
- many genetic tests produce results that do not differ to any great extent from other categories of private health information; and

51 R Magnusson, ‘Australian HIV/AIDS Legislation: A Review for Doctors’ (1996) 26 *Australian and New Zealand Journal of Medicine* 396; J Godwin and others, *Australian HIV/AIDS Legal Guide* (2nd ed, 1993) Federation Press, Sydney.

52 R Magnusson, *Submission G039*, 10 January 2002.

- in the next decade genetics will become a less unique and more commonplace tool in diagnosis, treatment, and healthcare generally.⁵³

3.61 Privacy NSW submitted that:

On one view the kind of information derived from DNA samples is so utterly unlike other kinds of personal information that it needs to be the subject of separate regulation. Arguments in favour of the separate regulation of genetic information are particularly appealing in the current regulatory environment where information privacy protection varies widely across and within jurisdictions.

On another view, the social implications of genetic information are so broad that a specific regulatory framework, just for genetics, would be unlikely to pick up all of them. Separate genetic regulation may risk engendering an overly deterministic approach to health care and privacy which fails to adapt to changing issues and technology. Integrating genetic safeguards in mainstream privacy legislation may ensure a more consistent and open process of adaptation to new social values and needs. In some specific areas such as insurance and employment, however, the exceptional features of genetic information may justify special treatment.⁵⁴

3.62 The bulk of this Report examines the use of genetic information in a variety of different contexts.⁵⁵ Where existing laws or processes are inadequate or inappropriate, the Inquiry has recommended their reform. Where there are major gaps, the Inquiry has proposed a number of new, targeted laws and processes.

3.63 In recognition of the fact that genetic science and technology are moving very rapidly, the Inquiry has also recommended that a new standing body—the Human Genetics Commission of Australia—be established to provide on-going advice to government.⁵⁶

The dangers of ‘genetic essentialism’

3.64 Aldous Huxley’s *Brave New World*, which was published in 1932, was one of the first cultural responses to the fascination with eugenics as a modernist principle for using science to improve society and social organisation.⁵⁷ In light of recent scientific advances, popular culture is again beginning to consider the chilling vision of a society organised around genetic determinism in films such as ‘GATTACA’.⁵⁸

53 Centre for Law and Genetics, *Submission G048*, 14 January 2002. See also Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; Life Sciences Network, *Submission G129*, 19 March 2002.

54 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

55 The Queensland Government also has urged a contextual approach, noting that ‘it would appear that a single new enactment may not be sufficient to overcome situations where there may be a threat of discrimination, in for eg insurance or employment, or threat to privacy and confidentiality’: Queensland Government, *Submission G161*, 16 May 2002.

56 See Ch 5.

57 A Huxley, *Brave New World* (1932) Penguin Modern Classics, London.

58 See Ch 4.

3.65 ‘Genetic essentialism’ is a reductionist view of human beings as essentially consisting of their genes,⁵⁹ with human worth describable in the language of genetics. It is closely associated with ‘genetic determinism’—the belief that human health and behaviour are basically predetermined by, and co-extensive with, a person’s genetic make-up. That is

personal traits are predictable and permanent, determined at conception, ‘hard-wired’ into the human constitution . [T]his ideology minimizes the importance of social context.⁶⁰

3.66 One of the discoverers of the DNA double helix, Nobel Prize winner Dr James Watson, famously has said that ‘[w]e used to think our future was in the stars. Now we know it is in our genes’. The widespread use of genetic information to identify individuals or groups at risk for disease or harm from a work environment; to guide provision of social benefits or services; or to classify people in any way, may challenge or change the way we think about what it means to be human.

3.67 As a society, will we come to measure the worth of a person by his or her genetic status? Will we come to regard all illness and even behaviour and preferences (political, sexual, cultural, aesthetic) as genetically predetermined? Will we come to view some gene-linked behavioural traits (obesity, smoking, drug and alcohol dependence) with more sympathy, in the same way that we now avoid attributing fault to, and stigmatising, persons with other genetic disorders?⁶¹ If so, will this challenge the fundamental ideas upon which civil society and the legal system are built, which emphasise free will, autonomy, and individual moral, social and criminal responsibility?⁶²

3.68 The Office of the Federal Privacy Commissioner submitted that advances in genetic science

should not override the fact that, as human beings, we have the unique capacity to make rational decisions about our lives. This means that the greatest human right is the freedom to choose. Even if our understanding of the interaction of the determining factors were to improve enormously, except where social considerations require it (for example, the lawful prevention of harms to others), the existence of a free society presumes that the individual is free to choose.

Especially where that choice involves our own bodies and destinies, the voluntary nature of our decision-making excludes reliance on the concept that our lives and well-being are pre-determined genetically. To hold otherwise is to render an exercise of an individual’s free will in a democratic society as a meaningless practice. With that freedom to choose, there comes the social recognition of the need for people to take responsibility for their decisions, which could otherwise be evaded by pleading

59 Or ‘Genes ‘R’ Us’, as one submission put it: I Barns, *Submission G056*, 8 January 2002.

60 R Dreyfuss and D Nelkin, ‘The Jurisprudence of Genetics’ (1992) 45 *Vanderbilt Law Review* 313, 316-321.

61 M Segal, ‘Autonomy and the Family in the Testing of Adolescents for Behavioural Traits’ (Paper presented at Seventh International Conference of the Human Genome Organisation, 17 April 2002).

62 For a discussion of how genetics might impact upon traditional notions of criminal responsibility, see C Wells, ‘I Blame the Parents’: Fitting New Genes in Old Criminal Laws’ (1998) 61 *Modern Law Review* 724.

genetic pre-determinism. The consequences of the latter, for example, in the criminal justice system, are unthinkable.⁶³

3.69 Murray has warned that

genetic risks may come to be seen as *the* explanation for complex multifactorial diseases. They may also be seen as fundamental, defining characteristics of the persons who have such risks, essentially reducing those persons to their genetic propensities. ... we do not have to pretend that genes are unimportant to avoid determinism or reductionism. We should give genes their due, but no more than that.⁶⁴

... there is a vicious circularity in insisting that genetic information is different and must be given special treatment. The more we repeat that genetic information is fundamentally unlike other kinds of medical information, the more support we implicitly provide for genetic determinism, for the notion that genetics exerts special power over our lives.⁶⁵

3.70 Ridley has cautioned against adopting a crude dichotomy that equates genetic influences with ‘determinism’, and environmental influences with ‘freedom’.

There has been a long tradition among a certain kind of science writer to say that the world of biology is divided into people who believe in genetic determinism and people who believe in freedom. Yet these same writers have rejected genetic determinism only by establishing other forms of biological determinism in its place—the determinism of parental influence or social conditioning. It is odd that so many writers who defend human dignity against the tyranny of our genes seem happy to accept the tyranny of our surroundings. ... The crude distinction between genes as implacable programmers of a Calvinist predestination and the environment as the home of liberal free will is a fallacy.⁶⁶

3.71 A number of serious concerns may be raised about the social consequences of adopting or internalising the tenets of genetic essentialism, even where this is borne of optimism for the benefits of medical and scientific advances. For example, an over-concentration of research into the field of genetics could lead to neglect of the effects on human health of other factors, such as the physical, social, spiritual and economic environments in which people live.⁶⁷

3.72 Many communities in Australia have close family and cultural links and are aware of their origins and heritage. Nelkin and Lindee have cautioned against supplanting human identity and relationships with molecular biology.

63 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002. See also S Raeburn, *Submission G033*, 2 January 2002.

64 T Murray, ‘Genetic Exceptionalism and “Future Diaries”: Is Genetic Information Different From Other Medical Information?’ in M Rothstein (ed), *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) Yale University Press, New Haven, 60, 70.

65 Ibid, 71.

66 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 302-303.

67 See R Lewontin, *The Triple Helix: Gene, Organism and Environment* (2000) Harvard University Press, Cambridge. Regarding the spiritual dimension in health, see Christian Science Committee on Publication, *Submission G127*, 19 March 2002.

As the science of genetics has moved from the laboratory to mass culture, from professional journals to the television screen, the gene has been transformed. Instead of a piece of hereditary information, it has become the key to human relationships and the basis of family cohesion. Instead of a string of purines and pyrimidines, it has become the essence of identity and the source of social difference. Instead of an important molecule, it has become the secular equivalent of the human soul.⁶⁸

3.73 In Chapter 36, the Inquiry notes that there have already been disputes in some Aboriginal communities about the use of genetic testing to confirm or deny kinship relations—and thus to determine Aboriginality. A prominent example was the use of genetic testing to assist in determining eligibility to vote in the Aboriginal and Torres Strait Islander Commission regional elections in Tasmania in 2002.

3.74 Current attitudes of social solidarity could be threatened by genetic essentialism. For example, an expectation could develop that those with genetic susceptibilities, or those at risk of having children with a genetic disorder, should take financial responsibility for their own and their affected children's health care.⁶⁹

3.75 As discussed in Chapter 23, there is a clear need for much more public and professional education about genetics, and the sensible application of genetic information within a range of important contexts.

3.76 The submission from Queensland Advocacy Inc summarises very well the social forces and tensions involved:

Genetic determinism is not necessarily a belief in a causal world in which only genes have a determining effect on human health and behaviour. It could rather be the belief that the world is best served through emphasising the role of genes and seeking to order life accordingly. ...

The most important response to the state of affairs in which this does occur, is to create opportunities for greater public involvement in the cycles of construction, deconstruction and reconstruction of genetic information. Human genetic information is ultimately, not about genes; it is about people. The information, because it originates in language and social practices, can be used in two ways. Either it can be regarded as private, which centres power in professions and individuals, or it can be regarded as public, which places power back into the community. The responsibilities created by either response are enormous. On either side lie distinctly undesirable prospects—enormous professional and corporate power or overwhelming public apathy—and the future may be charted through one of these factors ensuring that the other occurs. The task of ethics, including this inquiry, is to work against either occurring.⁷⁰

68 D Nelkin and S Lindee, *The DNA Mystique: The Gene as a Cultural Icon* (1995) W Freeman, New York, 198.

69 Australian Federation of Right to Life Association, *Submission G082*, 17 January 2002.

70 Queensland Advocacy Inc, *Submission G088*, 4 February 2002.

3.77 The challenge for our society is to maintain its moral and ethical compass, supporting those aspects of genetic science that reduce pain and suffering and increase the quality of life, while firmly resisting the perverse use of genetic information in a way that diminishes personal freedom and responsibility, and creates new opportunities for unfair discrimination.

4. Planning for the Future

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A glimpse of the future?

4.1 The film ‘GATTACA’ was released in 1997.¹ It portrays life in a ‘not-so-distant future’ in which genetic engineering permits parents to screen embryos before implantation for the purpose of reproduction—avoiding those that are genetically imperfect and selecting those that offer a genetic guarantee of health, stamina and physical attractiveness. One reviewer described the film in the following terms:

The main focus of *Gattaca* is the struggle of a genetically inferior man, Vincent Freeman, to survive and prosper in a world where his kind is routinely discriminated against.

Shortly after they were married, Vincent's parents decided to start a family the old-fashioned way, without any help from doctors and test tubes. The result was a boy who was diagnosed as 99% likely to have a serious heart defect. That rendered Vincent ineligible for all but the most menial of jobs. But his dream was to one day work at The Gattaca Aerospace Corporation and participate in the first-ever manned flight to the moons of Saturn. For most ‘in-valids’, this would have remained a fantasy, but Vincent possessed the determination and drive to make it real.

With the help of a shady middle-man, Vincent locates Jerome Morrow, a genetically superior individual who was paralysed as the result of an accident. He agrees to sell Vincent his identity (including blood and urine on demand, fingerprints, hair and other body debris, etc). So, equipped with Jerome's genetic resume, which guarantees him work anywhere, Vincent applies for a position at Gattaca. He is accepted and

1 A Niccol, *GATTACA* (1997), Columbia Pictures.

quickly proves his worth to everyone. But, a week before he is to attain his lifelong ambition of making a space flight, he becomes a suspect in a murder investigation and his carefully-guarded secret is in danger of being exposed.²

4.2 With the indulgence of a sympathetic medical officer who chooses not to disclose Vincent's deception, Vincent transcends his 'genetic prophecy'. Despite a life expectancy of only 33 years and 'already 10,000 [heart] beats overdue', in the final scene Vincent is launched into the night sky on his mission into space.

4.3 GATTACA identifies many themes that are central to the present Inquiry: the prospect that genetic science may in time enable a person's genetic destiny to be mapped out at birth, with all their flaws, predispositions and susceptibilities; the prospect that those with better genetic profiles may be favoured over those with weaker profiles, creating a class of 'healthy-ill' or 'worried-well'; and the prospect that those who are genetically disadvantaged may defy scientific predictions and succeed beyond expectations, while those who are genetically advantaged may not fulfil their potential—because, as Vincent Freeman states, 'there is no gene for fate'.

The march of science

4.4 At the time of GATTACA's release, some reviewers described the film as based on a 'chillingly feasible premise'.³ Others, however, thought the science to be suspect, commenting that 'it is highly dubious whether any of the genetic engineering portrayed in the film will ever be possible'.⁴

4.5 The intervening years offer fresh insights into the plausibility of GATTACA's underlying premise. In the six years since the film was released there have been many important developments in genetic science. Those that have received a deal of media coverage include:

- the cloning of 'Dolly the sheep' (announced in 1997) and the subsequent cloning of other animals, including Australia's first cloned merino 'Matilda' (2000);⁵
- sex-selection of embryos through a technique of sperm sorting (1998);
- the completion of the first mapping of the human genome by the international consortium of scientists involved in the Human Genome Project (1999);
- the use of gene therapy by French researchers to treat severe combined immunodeficiency disorder in two infants (2000);⁶

2 J Berardinelli, *Gattaca*, <www.movie-reviews.colossus.net/movies/g/gattaca.html>, 16 February 2003.

3 Ibid.

4 J O'Ehley, *GATTACA*, <www.sciflicks.com/gattaca/review.html>, 16 February 2003.

5 ABC Science Online, *Australia's First Cloned Sheep and Calf*, <www.abc.net.au/science/news/stories/s123089.htm>, 16 February 2003.

6 R Dobson, 'Gene Therapy Saves Immune Deficient Babies in France' (2000) 320 *British Medical Journal* 1225.

- the alleged reproductive cloning of a human being by an Italian doctor, Dr Severino Antinori, and by the Raelian Movement (2002);⁷ and
- the advent of ‘designer babies’, such as the case of a deaf lesbian couple in the United States who consciously chose a deaf sperm donor to increase the chance of conceiving a baby with the same disability (2002).⁸

4.6 In addition to advances in genetic science and technology, there have been rapid developments in the uses to which existing genetic technology has been put. For example, the Australian artist Pro Hart has begun to incorporate his genetic material in his paintings in order to facilitate authentication of his artworks and avoid forgery.⁹ One United Kingdom biotechnology company, Sciona, has begun to market an over-the-counter genetic testing service, linking dietary advice to variations on nine selected genes thought responsible for producing enzymes involved in general ‘body maintenance’.¹⁰ More frivolously, the *New York Times* has reported that DNA parentage tests are being used as a basis for a television game show in which a talk-show host announces the test results on air before the putative parents and a studio audience.¹¹ And one English firm has begun to market ‘the ultimate personalised gift’—a piece of unique jewellery that incorporates the donor’s DNA.¹²

4.7 While it is difficult to predict the course of human progress with any pretence of accuracy, the Inquiry is of the view that a number of nascent developments in genetic medicine, science and technology will place increasing pressure on the central issues confronting this Inquiry. These include:

- The further development of gene chip or micro-array technology, which will enable a large number of genetic tests to be performed on a sample simultaneously.¹³
- The likely reduction in the costs of genetic testing as technology is refined and adopted more widely in the community. In such circumstances there may be increasing pressures on governments, employers, insurers and others to uncover genetic information about individuals with whom they interact.

7 Clonaid, <www.clonaid.com>, 16 February 2003.

8 D Teather, *Lesbian Couple Have Deaf Baby by Choice*, *The Guardian*, 8 April 2002, <www.guardian.co.uk/international/story/0,3604,680616,00.html>, 19 February 2003.

9 O Hill-Douglas, *Cheeky Pro Hart puts DNA Signature on his Paintings*, The Age Company Ltd, <www.theage.com.au/articles/2002/03/06/1015365716233.html>, 20 February 2003.

10 Sciona, describes itself as ‘commercialising research in the areas of nutrigenomics and pharmacogenomics in order to create a range of services that enhance people’s lives and well-being.’ See Sciona Ltd, *About Sciona*, <www.sciona.com/about/index.htm>, 22 July 2002; J Meek, ‘Genetic Testing Rules “Unenforceable”’, *The Guardian*, 4 June 2002.

11 A Stanley, ‘In DNA Tests, TV Finds Elixir to Raise Ratings’, *New York Times*, 19 March 2002.

12 DesignAgifts, <www.designagifts.co.uk>, 16 February 2003.

13 See GenomeWeb Staff Reporter, *Microarray Market Could Top \$5 Billion by 2010 Study Says*, <www.genomeweb.com/articles/view.asp?Article=2003212115041>, 16 February 2003, predicting high growth in the micro-array market over the next decade.

- The likely increase in the speed of genetic testing, enabling testing to be carried out in minutes rather than days. This may open up the possibility of using genetic screening in new situations, such as border control.¹⁴
- The expanding medical understanding of the role of genetics in common diseases and conditions (such as asthma and hypertension), as well as expanding pharmacological understanding of the role of genetics in the metabolism of drugs.
- The expanding understanding of the role of genetics in the expression of a range of non-medical traits, including human behaviours such as aggression and addiction.
- The expansion of genetic screening across broad population groups, facilitating the collection of an unprecedented amount of sensitive health information about individuals and communities. Examples in Australia include school-based screening for Tay-Sachs disease and employment-based screening for haemochromatosis through the HaemScreen program.¹⁵ Other countries are developing broader genetic screening programs.
- The increasing capacity of information technology to provide rapid access to personal information (including genetic information) stored in disparate locations. Bioinformatics may permit the linkage of health data from disparate sources, including newborn screening cards, pathology samples, research studies and clinical records. In doing so, the linkages may create new and powerful information, but also heighten concern about the privacy of such information.¹⁶
- The introduction of personal identification cards that containing biometric information, including genetic information, and which may be used in a variety of contexts such as access to public services and immigration control.¹⁷
- Increasing pressures from commercialisation, whether that be from governments selling population genetic data to researchers, as in Iceland, Estonia and Tonga; biotechnology companies seeking entrance to niche markets; large pharmaceutical companies seeking to profit from advances in pharmacogenetics; or individuals seeking to patent human genes or genetic processes.

14 J Robertson, 'The \$1000 Genome: Ethical and Legal Hurdles' (2003) 4 *Nature Reviews Genetics* 82, discussing the development of faster and cheaper ways of whole-genome genotyping of individuals.

15 HaemScreen is a pilot genetic screening program for haemochromatosis run by the Murdoch Children's Research Institute and Genetic Health Services Victoria at the Royal Children's Hospital, Melbourne.

16 An example is the UK Biobank, which aims to track 500,000 volunteers for a research study aimed at establishing how genes, lifestyle and environmental factors interact to affect people's health. See The UK Biobank, <www.ukbiobank.ac.uk>, 19 February 2003.

17 See Secretary of State for the Home Department, *Entitlement Cards and Identity Fraud: A Consultation Paper* (2002), London, 104, which considers but rejects as 'too invasive' the use of DNA information in an identity card.

Government support for biotechnology

4.8 The Australian Government's strong support for biotechnology suggests that the pressure of change is likely to continue. In common with many developed countries, Australia has a policy, expressed in the Federal Government's *Innovation Statement*,¹⁸ which places great reliance for our economic future on the emerging new technologies, including human genetic technology. Significant steps have been taken to implement this policy:

- The National Statement on Ethical Conduct in Research Involving Humans¹⁹ has set down a comprehensive national ethical regulatory framework for the conduct of research in general and genetic research in particular.²⁰ (See Part D of this Report.)
- Biotechnology Australia is a whole of government initiative²¹ to coordinate efforts to develop biotechnology for the benefit of the Australian community.²²
- The Ralph Report on taxation reform has recommended reforms to income tax arrangements to ensure that the Australian taxation regime for biotechnology companies is consistent with other OECD nations, as a means of encouraging investment in Australian biotechnology.²³
- A major review of health and medical research in Australia has been undertaken. The Wills Report²⁴ refers particularly to the need to take advantage of advances in biotechnology to improve the health of the Australian population, to build the economy and to create valuable jobs.²⁵ It recognises that this window of opportunity would close given the pace of change unless Australia acts promptly.
- In January 2002, the Federal Government announced that, as part of a shift towards setting national priority areas for research, one-third (approximately \$170 million) of the Australian Research Council's funding grants for 2003

18 Commonwealth of Australia, *Backing Australia's Ability: An Innovation Action Plan for the Future*, <www.backingaus.innovation.gov.au/default.htm>, 24 July 2002.

19 Prepared by the Australian Health Ethics Committee under the relevant provisions of the *National Health and Medical Research Council Act 1992* (Cth) and endorsed by the Australian Vice Chancellors' Committee, the Australian Research Council and the various learned Academies in 1999.

20 Ibid, Principles 16.1–16.23 and the section on Human Tissue (Principle 15).

21 Involving the Commonwealth Departments of Industry, Tourism and Resources; Environment and Heritage; Agriculture, Fisheries and Forestry; Health and Ageing; and Education, Science and Training.

22 Commonwealth of Australia, *Australian Biotechnology: Progress and Achievements*, <www.biotechnology.gov.au/Industry_Research/National_Strategy/prop?biotech_prog_achiev.pdf>, 16 July 2002.

23 Review of Business Taxation, *A Tax System Redesigned: More Certain, Equitable and Durable: Report* (1999), Commonwealth of Australia, Canberra (Ralph Report).

24 Health and Medical Research Strategic Review, *The Virtuous Cycle, Working Together for Health and Medical Research* (1999), Commonwealth of Australia, Canberra (Wills Report).

25 Ibid, 1.

would be reserved for four designated key areas of scientific research, one of which is genome/phenome research.²⁶

4.9 These initiatives recognise that the preconditions for economic growth in the genetic technology sector include reasonable access to research tools (including human biological material), security of investment, and effective and appropriate regulation.

4.10 A central tenet of Biotechnology Australia is to ensure that ‘consistent with safeguarding human health and ensuring environment protection, that Australia captures the benefits of biotechnology for the Australian community, industry and environment’.²⁷ The former federal Minister for Health and Aged Care, the Hon Dr Michael Wooldridge MP, emphasised

the driving imperative of identifying and managing any risks associated with the technology before all other matters, only then can we be truly confident about reaping the broader benefits.²⁸

4.11 Genetic science and technology is developing apace, and will provide the basis for other applied sub-branches of medical and scientific research, including bio-informatics,²⁹ proteomics,³⁰ and pharmacogenetics.³¹ It is of equal importance that these rapid developments are accompanied by informed consideration of the ethical, legal and social implications of the science, as well as the development of secure and appropriate ethical and regulatory frameworks.³²

Social reactions to rapid scientific change

4.12 The pace of scientific advancement in biotechnology and in other related fields creates a degree of social ambivalence about the potential benefits and potential dangers of change. On the one hand, there is very strong public support for breakthroughs promising better medical diagnosis and treatments, and for assisting with law enforcement; on the other, there is some general fear about uncontrolled or ‘mad science’, the spectre of eugenics, threats of biological warfare, reports of

26 See Dr Brendan Nelson, Minister for Education, Science and Training, Minister for Education Science and Training, *Backing Australia's Ability — Funding for Research Priorities Announced*, <www.dest.gov.au/ministers/nelson/jan02/n06_290102.htm>, 20 February 2003. See also D Illing, ‘Nelson says Research Quality Won’t Suffer’, *The Australian*, 6 February 2002. The other designated areas are nano- and bio-materials; complex/intelligent systems; and photon science and technology.

27 See Commonwealth of Australia, *Australian Biotechnology: Progress and Achievements*, <www.biotechnology.gov.au/Industry_Research/National_Strategy/prop?biotech_prog_achiev.pdf>, 16 July 2002.

28 In the Second Reading Speech for the Gene Technology Bill 2000 (Cth). See Commonwealth of Australia, *Parliamentary Debates*, House of Representatives, 22 June 2000, 18104 (The Hon Dr Michael Wooldridge (Minister for Health and Aged Care)).

29 Harnessing the power of new information technology to advances in genetic science and technology.

30 Studying the genetic influences on protein production.

31 Tailoring drug remedies to an individual or group’s particular genetic characteristics.

32 See Commonwealth of Australia, *Australian Biotechnology: Progress and Achievements*, <www.biotechnology.gov.au/Industry_Research/National_Strategy/prop?biotech_prog_achiev.pdf>, 16 July 2002.

xenotransplantation (transplants from one species to another), the loss of privacy, and the increased possibilities for genetic discrimination.³³

4.13 It is an important lesson for Australians that Europeans appear to be losing faith in the ability of public authorities to regulate biotechnology adequately in the public interest. In part, this is because of perceived inadequate government and corporate responses to crises in Europe over foot and mouth disease, Creutzfeldt-Jakob Disease, genetically modified foods, and human cloning.³⁴ European surveys indicate a deepening suspicion of public authorities, technical experts and commercial organisations operating in this area, as well as a high (and growing) degree of scepticism about international institutions, farmers' associations and religious organisations as sources of information about biotechnology.³⁵

4.14 Similarly, a recent survey conducted in the United States by the Genetics and Public Policy Center of Johns Hopkins University found that 'Americans are both hopeful and fearful about the rapidly advancing power of scientists to manipulate human reproduction'. There was strong opposition to human reproductive cloning, but general support for health-related applications of the new genetic technologies. There also was strong support for government regulation in this area, across all political party lines. The Center expressed concern that the survey results for the eight 'knowledge questions' asked of respondents indicated that the 'public's knowledge about these technologies is not keeping pace with the steep growth in genetic science'.³⁶

4.15 By way of contrast to the position in Europe, recent Australian surveys commissioned by Biotechnology Australia have found an increased level of trust in Australian government agencies as both a source of factual information and as regulators. The CSIRO was regarded as a credible source of information by 85% of respondents; Food Standards Australia New Zealand (formerly the Australia New Zealand Food Authority) and the Office of Gene Technology Regulator both scored 73%; and Biotechnology Australia was rated as credible by 58% of respondents.³⁷

4.16 However, this survey also found a high level of anxiety about the pace of biotechnological change and society's capacity to regulate it effectively:

Most respondents felt that biotechnology is changing at such a rapid pace that developments cannot possibly be anticipated and legislated against. In addition, it was generally felt that Australian society and government are powerless compared to the

33 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.7].

34 Only 45% of Europeans agreed with the statement that their governments regulate biotechnology well enough, compared with 29% who disagree, and 26% who are not sure: Eurobarometer 52.1, *The Europeans and Biotechnology*, <www.europa.eu.int/comm/research/quality-of-life/eurobarometer.html>, 19 February 2003.

35 Consumer organisations, the medical profession and environmental protection organisations fared best, while universities, animal protection organisations and the media (20%) had modest levels of support. See *Ibid.*

36 Genetics and Public Policy Center, *Americans Deeply Divided about use of Genetic Technologies in Reproduction*, <www.dnapolicy.org/research/index.jhtml>, 24 February 2003.

37 Millward Brown Australia, *Biotechnology Public Awareness Survey Final Report*, Biotechnology Australia, <www.biotechnology.gov.au/MBsurveyresults.pdf>, 21 August 2002.

international financial and political power of the large multinational companies driving biotechnological innovations. A key component of concern was the perception that there are no or inadequate controls over the process, motivations and outcomes of the development and application [of] biotechnology and gene technology in Australia. This was particularly a concern for those applications which were seen to raise complex and disturbing questions about human life.³⁸

4.17 The major challenge for the Inquiry, then, is to steer a sensible path through these concerns in order to make recommendations that meet the public desire for further advances in genetic science in aid of human health, while providing sufficient reassurance to the Australian community that any such work will be subject to searching ethical scrutiny and effective legal controls.

Law reform in times of rapid scientific change

4.18 A critical issue for this Inquiry is how to make today's law reform relevant to the scientific developments of tomorrow. The importance and the difficulty of achieving this was a common theme in submissions made to the Inquiry. For example, Louise Martinovic submitted:

There is widespread community concern that as developments in [human genetics] are occurring rapidly, the law will 'fall behind' and not be able to adequately protect the privacy of the individual against inappropriate discriminatory use of their genetic information.³⁹

4.19 Graham Whittaker identified the need for sound prediction and flexible solutions:

We are informed that the results of the current Human Genome Project and subsequent research, also building on existing knowledge, are likely to produce more rapid advances in human genetic technology than have hitherto been achieved. A number of experts have attempted to speculate about future developments, but the future is impossible to predict and so changes to legislation etc need to be a best effort to suit what is most likely to occur, and to allow dynamic modifications.⁴⁰

4.20 Similarly, the Office of the Federal Privacy Commissioner (OFPC) stated that:

It is vital for Australia as a competitive and forward-thinking nation that the institutional measures, which are proposed for the protection of genetic privacy, are capable of accommodating genetic and technological developments and their pace of change.⁴¹

4.21 The Inquiry is conscious of the need for law reform to be sensitive to the dynamic environment in which medical, scientific and technological developments are taking place in the field of human genetics. That environment includes extensive

38 Ibid, 29.

39 L Martinovic, *Submission G006*, 30 May 2001. See also Australian Medical Association, *Submission G091*, 29 January 2002.

40 G Whittaker, *Submission G085*, 21 January 2002.

41 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

debate about the impact of developing knowledge of genetic information on how we see ourselves as individuals, as family members and as citizens. The values that inform and found the ethics of our society are likely to be challenged by such new developments. Remaining open to these challenges is an essential part of this dynamic environment.

4.22 Below the Inquiry identifies seven attributes of the reform process, which are aimed at ensuring that the recommendations in this Report meet the needs that are likely to arise in the short to medium term. Prognostication may be more art than science. However, as a leading futurist has suggested, the purpose of thinking ahead in this fashion is not to provide an accurate picture of tomorrow but to improve decision making in the present.⁴²

4.23 In summary, it is the Inquiry's view that governments and other public institutions should be sensitive to the dynamic environment in which medical, scientific and technological developments are taking place in the field of human genetics, when they consider proposals to reform laws, codes, guidelines and practices relating to human genetic information. In particular, and as discussed further below, governments and other public institutions should:

- Promote widespread community participation in the formulation of relevant rules and principles;
- Find appropriate balances between competing interests;
- Adopt processes that facilitate contributions from all relevant disciplines;
- Consider the cross-border implications of the issues, whether they be federal or international in character;
- Consider forms of regulation that are flexible and quick to adapt to changing circumstances;
- Seek simple and effective regulation through greater harmonisation of the regulatory regimes in different jurisdictions; and
- Establish and maintain such institutions as are appropriate to address, on an on-going basis, issues relating to the use and protection of human genetic information.

Widespread community participation

4.24 In IP 26, the Inquiry noted that public optimism about the potential benefits flowing from advances in human genetics is tempered by concerns arising from rapid change. The Inquiry stated that:

42 P Schwartz, *The Art of the Long View: Planning for the Future in an Uncertain World* (1995) Currency Doubleday, New York.

On the one hand, there is very strong public support for breakthroughs promising better medical diagnosis and treatments, and for assisting with law enforcement (including identification of missing persons); on the other, there is some general fear about uncontrolled or 'mad science', the spectre of eugenics, threats of biological warfare, reports of xenotransplantation (transplants from one species to another), the loss of privacy, and the increased possibilities for genetic discrimination.⁴³

4.25 The Inquiry also noted that a federal government agency, Biotechnology Australia, had commissioned a study of public attitudes to biotechnology, which reported as follows:

Most respondents felt that biotechnology is changing at such a rapid pace that developments cannot possibly be anticipated and legislated against. In addition, it was generally felt that Australian society and government are powerless compared to the international financial and political power of the large multinational companies driving biotechnological innovations. A key component of concern was the perception that there are no or inadequate controls over the process, motivations and outcomes of the development and application [of] biotechnology and gene technology in Australia. This was particularly a concern for those applications which were seen to raise complex, and disturbing questions about human life.⁴⁴

4.26 The Inquiry considers it necessary to ensure broad public consultation and community participation in any reform process if public concerns about human genetics are to be adequately addressed. Community engagement has been fundamental to the methodology of the Australian Law Reform Commission since its inception, just as public consultation is integral to the work of the Australian Health Ethics Committee (AHEC) and the National Health and Medical Research Council (NHMRC). As indicated in Chapter 1, that methodology has been followed in the present Inquiry, with the holding of a large number of public meetings; widespread dissemination of IP 26, DP 66 and genetics brochures; participation in numerous public forums on genetics; and receipt and consideration of over 310 public submissions. Similar principles should also be applied to any processes of on-going review that are established as a result of this Report.

4.27 The Inquiry believes that open and accountable processes that engage with the public are essential for several reasons.⁴⁵ They are beneficial:

- for those consulted, because they facilitate civic participation and have a valuable educative function in an area of science that requires lifelong learning;
- for the process of law reform, because those with day-to-day experience of the law can indicate how law and practice actually affect them; and

43 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.7].

44 Millward Brown Australia, *Biotechnology Public Awareness Survey Final Report*, Biotechnology Australia, <www.biotechnology.gov.au/MBsurveyresults.pdf>, 21 August 2002, 29.

45 See further, B Opeskin, 'Engaging the Public: Community Participation in the Genetic Information Inquiry' (2002) 80 *Reform* 53.

- for reform outcomes, because laws, codes and guidelines are more effective when the people who will be regulated by them participate in their formulation.

4.28 Widespread community participation in the process of reform or review is thus seen by the Inquiry as essential to maintaining public trust.

Balance and compromise

4.29 A second attribute to consider when seeking longevity of reform proposals is the need for balance and compromise. At the outset, the Inquiry identified a major challenge as finding a ‘sensible path’ through conflicting public concerns with respect to genetic information.⁴⁶ The balance is difficult to achieve in practice because relevant interests often compete or clash. This is evident in the submissions received on many issues considered in this Report.

4.30 One example is the conflict between the interests of genetic researchers, who need to secure the participation of many data subjects, and the interests of potential volunteers, who may fear that their participation will generate personal information that subsequently may have to be disclosed to insurers, employers or others. Another example is the conflict between the interests of employers, who must fulfil their common law and statutory duties to provide a healthy and safe work environment for their employees, and the interests of employees, who may fear adverse discrimination because of a genetic susceptibility to a disease triggered, for example, by exposure to hazardous substances in the workplace.

4.31 The need to balance interests has been recognised in the approach of similar bodies overseas. In the Foreword to a report of the United Kingdom Human Genetics Commission, Baroness Helena Kennedy remarked:

In the subtitle of this report we talk of balancing interests. This is an important aspect of our work. I am very much aware of the fact that people approach this issue of personal genetic information from varying perspectives. We have tried here to take account of a wide spectrum of views and have attempted to reach conclusions which are morally defensible and sensitive to the different interests involved.⁴⁷

4.32 In similar vein, the NSW Health Department stated that:

It is important that there is a mechanism to ensure that law reform, codes of practice and guidelines are sensitive and flexible enough to be effective in the dynamic environment of human genetics; and that there is balance and compromise between individual and societal rights, concerning genetic information in the community, medical profession, researchers, businesses and governments.⁴⁸

4.33 The Inquiry considers that black and white solutions to the complex problems thrown up by human genetics will rarely be appropriate. However, the

46 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.15]–[2.23].

47 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 5.

48 NSW Health Department, *Submission G303*, 13 January 2003.

precise balance to be struck between competing interests need not be the same for all time, nor for all societies. Moreover, a finely-tailored solution need not subjugate one group's interests to advance those of another: an appropriate balance may be mutually beneficial. The Anti-Discrimination Board of NSW made this point in its submission when addressing the potentially competing interests of medical researchers and data subjects in the regulation of human research.

Fear of genetic discrimination is also likely to impact upon people's willingness to participate in research. ... Rather than acting as an impediment to the development and application of genetic technology, effective anti-discrimination and privacy legislation are critical to realising the public health benefits of genetic information. Conversely, if we fail to provide such protection, discrimination and privacy concerns will act as disincentives to testing and research participation and have negative consequences for individual and public health outcomes.⁴⁹

4.34 There is a wealth of policies and ethical pronouncements from international agencies and NGOs that readily use general terms like 'fairness', 'equity', 'reciprocity', 'justice' and 'non-discrimination'. No one could possibly argue with such principles nor the sentiments underlying them. However, the real difficulty comes in making the choices involved in balancing legitimate but competing interests in practice. These difficulties include taking into account the reality of the high costs of medical and scientific research and of clinical trials; the current general preference of governments in industrialised countries (including Australia) for market solutions; the trends toward and pressures of privatisation, corporatisation, and commercialisation in such countries; and the limited resources and pools of expertise available for formal regulatory authorities.

Interdisciplinary approach

4.35 A third attribute of successful reform for the future is maintaining an interdisciplinary approach to the issues. The subject matter of this Inquiry is a unique fusion of law, health ethics, and medical and related sciences. This is reflected in the identity of the partners to the joint inquiry (the ALRC and AHEC); the expertise of their respective members and staffs; the portfolios of the Ministers to whom the Inquiry has reported (the Attorney-General and the Minister for Health and Ageing); the composition of the Advisory Committee;⁵⁰ the diversity of individuals and organisations with whom consultations have been conducted; and the range of submissions.

4.36 The importance of contributions from people with differing interests and perspectives was echoed in a number of submissions. For example, in the course of rejecting the suggestion that the disclosure of genetic information to a patient's relatives should be left in the hands of professional medical opinion, the OFPC commented that this approach fails to consider that:

49 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

50 See the List of Participants in this Report for the full composition of the Advisory Committee.

the need to accommodate a range of competing interests calls for input from a number of disciplines, such as the law, science, ethics as well as health care.⁵¹

4.37 The Office of the Victorian Privacy Commissioner stated that it

strongly supports the efforts of the Inquiry to engage the interest of a wide range of disciplines and to deepen community engagement in the issues raised by developments in genetics. These issues are profound. They will require the participation of specialists from a range of varied disciplines and the views of non-specialists if the laws and policies adopted are to be perceived as legitimate.⁵²

4.38 In the Inquiry's view, successful law reform now and in the future requires that an interdisciplinary approach to these issues be maintained.

Global perspective

4.39 Medical science and health ethics are international disciplines and do not conform to state or national boundaries. For this reason, policy makers in the field of human genetics need to be mindful of the implications of proposals in an increasingly borderless world. In practice, the capacity for transborder transactions may limit the capacity to regulate certain kinds of conduct.

4.40 For example, in Chapters 11 and 35 the Inquiry addresses the regulation of human genetic testing, including parentage testing. At present, parentage testing is available from Australian laboratories, which may or may not be accredited according to Australian standards, and from overseas laboratories, which may or may not be accredited in accordance with the standards prevailing in those foreign jurisdictions. If parentage testing is heavily regulated in Australia, one consequence may be to encourage people to use the less regulated facilities of overseas laboratories, whose services are often marketed over the Internet. Similarly, if the conditions for medical research in Australia are excessively stringent, research facilities may move offshore to sites that are more conducive to their particular brand of research, or Australian researchers may be denied participation in international multi-centre research studies. Reflecting the latter concern, the Queensland Government commented that:

With the prevalence now of global sharing of research information, it is essential that the privacy of individuals' genetic information and tissues samples be protected. However, provisions limiting the transborder flow of human genetic samples and information must be balanced against the possibility of excluding Australian researchers from the benefits of international collaboration and information sharing.⁵³

4.41 This is not to suggest that Australia should adopt the standards of the lowest common denominator by participating in a 'race to the bottom'. Rather, it is to call attention to the global environment in which information, people, goods and services may move across borders, whether interstate or international. It is also to call attention

51 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

52 Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002.

53 Queensland Government, *Submission G274*, 18 December 2002.

to the fact that maintaining high legal and ethical standards in Australia with respect to human genetic information cannot prevent other jurisdictions from developing standards that may not comport with those at home.

4.42 In some contexts international rules are being developed to promote the observance of minimum standards in the protection of human genetic information. In 1997 the United Nations Educational Scientific and Cultural Organisation (UNESCO) adopted the Universal Declaration on the Human Genome and Human Rights. The Declaration seeks to establish high-order principles and is not binding on member States. An example of a binding instrument (though of no direct relevance to Australia) is the Council of Europe's Convention on Human Rights and Biomedicine, which seeks to

protect the dignity and identity of all human beings and guarantee everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to the application of biology and medicine.⁵⁴

4.43 The Inquiry has investigated the approaches taken in a number of overseas jurisdictions to the protection of human genetic information. The Inquiry has held consultations in Australia with visiting foreign officials, including the Chair of UNESCO's International Bioethics Committee. The Inquiry has also held consultations in a number of European countries with organisations relevant to medical ethics, insurance, employees, employers, privacy and data protection, and these have been supplemented by consultations in the United Kingdom, Canada and the United States.

4.44 The Inquiry considers that future regulation of human genetic information should be sensitive to this global environment. An examination of relevant developments in other jurisdictions enables informed choices to be made for Australia based on international best practice in the field. One submission suggested that Australia could itself promote world best practice by taking

a lead role in engendering the international harmonization of innovative genetic research practice through developing policy and regulatory frameworks under the umbrella of 'Good Genetic Research Practice Guidelines'.⁵⁵

Flexible regulation

4.45 Many submissions to the Inquiry identified a critical ingredient in the regulation of human genetic information—the need for flexible mechanisms that are capable of adapting with appropriate speed to on-going developments in genetic science and medicine.

4.46 The following quotations are representative of a widespread concern among those who made submissions to the Inquiry:

54 *Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine*, opened for signature 4 April 1997, ETS No 164, (entered into force on 1 December 1999). As at 16 February 2003 there were 15 states parties.

55 J Fleming, *Submission G241*, 20 December 2002.

We must avoid placing unrealistic expectations on legislative solutions as they are by no means a universal panacea. We need to be mindful that there are also some drawbacks to a fixed legislative approach, particularly in an area which is undergoing rapid change.⁵⁶

Any legislation is likely to be inadequate to deal with the rapid changes in the area of genetic information. Therefore there needs to be constant surveillance of the ethical, privacy and discrimination issues.⁵⁷

[The Australian Academy of Science] also notes that this area of research is moving very quickly, and that laws can be difficult to amend if they no longer reflect the state of scientific knowledge and medical practice.⁵⁸

The NHMRC Guidelines do not have the same legal effect as legislation; they are not binding. The guidelines do however, have the advantage of being capable of rapid amendment and are therefore considered by some to be a better option for regulation than the enactment of 'genetic specific' legislation.⁵⁹

[The Investment and Financial Services Association] recommends that the industry be regulated by a mandatory Industry Standard, rather than legislation, as a Standard can be rapidly updated in line with scientific advances and changes in community attitudes.⁶⁰

4.47 The Inquiry considers that there will be circumstances in which legislative solutions are entirely appropriate in setting benchmarks that are binding, enforceable and visible in protecting human genetic information. Accordingly, many recommendations in this Report suggest amendments to existing legislative regimes in order to afford better protection to human genetic information. However, the Inquiry generally endorses the view expressed in the above submissions in so far as they call for an awareness of the drawbacks of legislation in responding to the need for reform in circumstances of rapid advances in science and technology. Although it is not possible to endorse a particular regulatory solution in advance for all circumstances, the Inquiry's recommendations recognise the need for a range of flexible solutions, including guidelines, codes of practice and better education. In these situations, the ethical foundations of that regulation will be clear because they reflect an accepted balance of values and principles.

4.48 It may also be appropriate in some circumstances to adopt co-regulatory approaches, where legislation establishes a basic framework supplemented by more detailed industry or professional codes. The *Privacy Act 1988* (Cth) provides one such model, with its National Privacy Principles (NPPs) promulgated by the OFPC; its approved (organisation-based) privacy codes; and the possibility of exemptions pursuant to the OFPC's public interest determinations.⁶¹

56 Centre for Law and Genetics, *Submission G048*, 14 January 2002. See also M Otlowski, *Submission G159*, 24 April 2002.

57 Aboriginal and Torres Strait Islander Social Justice Commissioner — Human Rights and Equal Opportunity Commission, *Submission G160*, 13 May 2002.

58 Australian Academy of Science, *Submission G097*, 21 January 2002.

59 L Martinovic, *Submission G006*, 30 May 2001.

60 Investment and Financial Services Association, *Submission G049*, 14 January 2002.

61 *Privacy Act 1988* (Cth) Pt IIIAA, Pt VI.

4.49 The importance of flexibility also suggests that it is desirable to undertake periodic review of this area to ensure that laws and practices meet society's needs as human genetics develops.

Simplified and harmonised regulation

4.50 During the course of the Inquiry, the ALRC and AHEC have investigated a number of regulatory regimes that are based on state or territory law, either alone or in combination with federal law. In some cases these regimes seek to achieve similar goals by similar means; in others the goals or the means differ from one jurisdiction to another. Some regulatory regimes have been formulated as the result of a deliberate policy of intergovernmental co-operation; in others the regimes have arisen in a piecemeal fashion as one jurisdiction models the practices of another, or pursues its own innovations. Examples of the legislative schemes considered in this Report include laws on:

- Information and health privacy;
- Anti-discrimination;
- Human tissue used for transplantation;
- Occupational health and safety;
- Workers' compensation; and
- Forensic procedures.

4.51 Many consultations and submissions voiced considerable concern, bordering sometimes on dismay, about the diversity between jurisdictions in the regulation of similar activity. For example, in relation to information and health privacy legislation, the Commonwealth Attorney-General's Department remarked on the practical difficulties that arise when organisations are required to comply with a number of related but conflicting laws:

It leads to greater expense when they have to seek professional advice regarding their legal obligations and implement different procedures for compliance. Where relevant it can also lead to forum shopping by consumers in relation to complaint-handling. This is an unsatisfactory situation and should be avoided by having national standards where possible.⁶²

4.52 In relation to forensic procedures legislation, the Office of the Victorian Privacy Commissioner noted that

At a minimum, the development of statutory models around the country reveals that uniformity is lacking. This raises obvious concerns, particularly in relation to the national pooling of DNA data. The quality and security of the stored data will only be as good as the weakest safeguards among the various participants.⁶³

62 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

63 Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002.

4.53 The divergence between federal, state and territory regimes is an inevitable consequence of a federal system of government. Such a system has compensating advantages: it facilitates innovation and experimentation, and it avoids an undue concentration of power in one level of government. However, the real costs of divergence can be seen in the duplication, gaps and inconsistencies in the regulation of a variety of conduct relevant to the protection of human genetic information. This often leads to confusion amongst those to whom the laws are directed and it may ultimately undermine the willingness of individuals to observe the law.

4.54 With these considerations in mind, the Inquiry believes that effective protection of human genetic information will require the worst disparities between jurisdictions to be eliminated over time. Processes will also have to be put in place to seek greater inter-governmental cooperation in relevant areas. Subsequent chapters of this Report address the need for harmonisation in particular contexts such as information and health privacy legislation (Chapter 7), anti-discrimination law (Chapter 9), and forensic procedures legislation (Chapter 40).

Appropriate institutions

4.55 The Inquiry believes that in order for reform to be successful in meeting the challenges of the future, it is essential to establish and maintain appropriate institutions. Bodies like the ALRC and AHEC have an important function to perform in developing a policy framework for protecting human genetic information. However, the present Inquiry is limited by its Terms of Reference: the completion of the present Report signals the end of the Inquiry's formal function in relation to this project. The ALRC, in particular, is limited by its constituting Act to reporting on matters referred to it by the Attorney-General.⁶⁴

4.56 However, advances in scientific knowledge will continue apace, requiring social and political responses. In Chapter 5 the Inquiry proposes the establishment of a standing body, which would have a continuing role in advising government on all aspects of human genetic science, as developments occur. Such a body has been established already in the United Kingdom, under the name of the Human Genetics Commission,⁶⁵ and in the Canadian province of Ontario, under the name of the Advisory Committee on Genetics. As Chapter 5 indicates, there has been broad support in submissions to the Inquiry for the establishment of such a body in Australia, with many of the submissions linking the need for a standing body to the challenges posed by rapidly changing science.

64 *Australian Law Reform Commission Act 1996* (Cth) ss 20–26. The ALRC has, however, received new Terms of Reference for a follow-on inquiry to examine intellectual property rights over genetic materials and related technologies. See The Hon Daryl Williams AM QC MP and Senator the Hon Kay Patterson, *News Release: Inquiry into Human Genetic Property Issues*, 17 December 2002.

65 Human Genetics Commission, *Topics*, <www.hgc.gov.uk/topics.htm>, 20 February 2003.

The scale of reform

4.57 This chapter has suggested a forward-looking approach to law reform, which acknowledges the dynamic nature of the medicine and science of human genetics. However, this does not imply that reform of the present regulatory framework is necessary on a grand scale. Reform may be effective even though the recommended changes in some areas are small.

4.58 Many chapters of this Report advance recommendations for reform that represent modest amendments to existing laws and practices—to extend coverage to genetic information, or clarify or modify the current position. Whenever it has been feasible to do so, the Inquiry has sought to build on existing institutions, laws and practices, rather than seek to erect a new edifice to deal with the challenges posed by human genetic information. In this way, the Inquiry seeks to recommend targeted reform that is efficient, cost-effective and capable of being achieved.

5. A Human Genetics Commission of Australia

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Introduction

5.1 The Terms of Reference for this Inquiry ask whether, and to what extent, a *regulatory framework* is required to protect the privacy, and protection from inappropriate discriminatory use, of human genetic samples and information. As noted in IP 26:

successfully fulfilling this brief not only involves providing adequate protections against the *unlawful* use of genetic information, but also putting into place measures aimed at ensuring that where such information may be used lawfully, it is used properly, fairly and intelligently.¹

The trend towards a national approach

In areas of relevance to this Inquiry

5.2 The current methods of regulation and conflict resolution involve a patchwork of federal, state and territory laws; official guidelines; personal and professional ethics; institutional restraints; peer review and pressure; oversight by public funding authorities and professional associations; supervision by public regulatory and complaints-handling authorities; private interests; and market pressures.

5.3 One fundamental question is the extent to which a national approach to the regulation of human genetics may be required, rather than relying upon the traditional mix of federal, state and territory laws as well as other formal and informal mechanisms.

5.4 The location of regulatory authority in a federal system is always a matter of some contention. Uniformity has obvious advantages in terms of clarity and certainty. However, in a rapidly developing area of science and technology, there also may be something to be said for allowing innovation and experimentation on a state-by-state basis. Given the wide array of activities covered by this Report, and the constitutional limitations on federal legislative power,² only a cooperative approach involving the Commonwealth, States and Territories would assure the successful establishment of a comprehensive national scheme.³

5.5 To a significant extent, the shift towards a national approach is already taking place (see Part B). For example, the extension of privacy protections to cover the private sector was achieved through federal law, regulations and processes, and is overseen by the Federal Privacy Commissioner (see Chapter 7). Aspects of federal anti-discrimination law and industrial law already cover the field (see Chapters 8 and

1 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.20].

2 Section 51 of the Constitution specifies the principal areas in which the Commonwealth Parliament may legislate, such as with respect to interstate and international trade and commerce, taxation, defence, immigration, insurance, and intellectual property rights.

3 There are a number of ways in which this could be achieved, ranging from a referral of powers to the adoption of uniform laws by each jurisdiction. The recent difficulties of achieving a national approach to corporate regulation that also survives constitutional scrutiny by the High Court indicates the traps in this area. See *Re Wakim; Ex parte McNally* (1999) 198 CLR 511.

30). Intellectual property rights in relation to genetics are determined according to federal laws and international agreements.⁴

5.6 The regulation of ‘therapeutic devices’ now has a national framework through the Therapeutic Goods Administration (TGA) (see Chapter 11). The regulation of plant and animal genetics in Australia is now the responsibility of the federal Office of the Gene Technology Regulator (OGTR).⁵

5.7 In June 2001, the Council of Australian Governments (COAG)—representing the federal, state and territory governments—agreed that it was strongly in the public interest to adopt a national approach to human cloning, stem cell research and related matters. In September 2001, a report of the House of Representatives Standing Committee on Legal and Constitutional Affairs recommended a uniform, national approach to legislation and the establishment of a national licensing body to regulate research involving embryonic stem cells and prohibit human reproductive cloning.⁶ On 5 April 2002, COAG formally agreed to adopt a nationally consistent approach to legislation dealing with human cloning, stem cell research and related matters, with a national regulatory framework for the licensing and monitoring of embryonic stem cell research (under specified conditions).⁷

5.8 On 27 June 2002, the Prime Minister, the Hon John Howard MP, introduced the Research Involving Embryos and Prohibition of Human Cloning Bill 2002. The Bill subsequently was split into two and after considerable debate enacted into law as the *Research Involving Human Embryos Act 2002* (Cth) and the *Prohibition of Human Cloning Act 2002* (Cth). The latter creates a number of serious criminal offences, carrying maximum penalties of 10 to 15 years. The former Act also creates a number of criminal offences,⁸ covering unauthorised use of an ‘excess ART embryo’.⁹ However, for these purposes, the most important feature of that Act is that it establishes a new Principal Committee of the National Health and Medical Research Council (NHMRC)—the Embryo Research Licensing Committee—to regulate all research in Australia involving human embryos (including embryonic stem cells),

4 Especially the *Patents Act 1990* (Cth) and the *World Trade Organization Agreement on Trade-Related Aspects of Intellectual Property Rights Agreement (TRIPS) annexed to the Marrakesh Agreement Establishing the World Trade Organization*, 15 April 1994, ATS 8, (entered into force on 1 January 1995), which Australia incorporated into domestic law with the *Patents (World Trade Organization Amendments) Act 1994* (Cth). See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [1.65]–[1.78]. As discussed in Ch 1, the ALRC has received separate terms of reference to inquire into Intellectual Property Rights over Genetic Materials and Genetic Related Technologies, with special reference to human health.

5 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [3.13]–[3.29] for a description of the role, structure and powers of the OGTR.

6 House of Representatives Standing Committee on Legal and Constitutional Affairs, *Human Cloning: Scientific, Ethical and Regulatory Aspects of Human Cloning and Stem Cell Research* (2001), Parliament of the Commonwealth of Australia, Canberra.

7 M Metherell and D Smith, ‘Stem-cell Go-ahead as PM Gives Way’, *The Sydney Morning Herald*, 6 April 2002, 1; J Kerin, ‘States Agree to PM’s Stem-cell Limits’, *The Australian* (Sydney), 6 April 2002, 4.

8 See ss 10–12.

9 As defined in s 9.

through a national system of licensing (ss 20–28), monitoring and sanctions for breach (ss 33–41).

5.9 According to the Regulation Impact Statement tabled with the original Bill, this new national regulatory system has been necessitated by:

- the current lack of consistency in regulatory coverage of human cloning and other unacceptable practices across the jurisdictions;
- the absence of a comprehensive, nationally consistent system for the regulation of research involving human embryos;
- the fundamental ethical issues posed by destruction of embryos for research and other uses;
- the ‘uneven playing field’ for researchers created by the inconsistent regulation, which may reduce the competitiveness of some researchers relative to their counterparts in other jurisdictions; and
- the impact that the current lack of certainty or national consistency in the regulatory environment may have on Australia’s international competitiveness.

5.10 Under the Act,¹⁰ the Embryo Research Licensing Committee has responsibilities for administering the licensing system, maintaining a publicly available database containing information about licensed embryo research, and monitoring compliance with the legislation.¹¹ The Financial Impact Statement contained in the Explanatory Memorandum to the original Bill estimated that:

Following the passage of the legislation, costs are realistically expected to be approximately \$3m per annum, with an upper maximum of \$6m. This involves a fixed cost to support the NHMRC Licensing Committee and provide for ongoing compliance monitoring related to the prohibited practices.

Emerging Canadian approach

5.11 In Canada, which has a federal system similar to Australia’s, there also have been strong moves towards a national approach to biotechnology regulation. The Premiers of all provinces and territories have agreed to make genetics one of five priority areas for national work, and at the August 2001 Premiers Conference, Ontario committed to producing a report on genetics and human health. The resulting report, *Genetics, Testing and Gene Patenting: Charting New Territory in Healthcare*, was accepted unanimously at a special Premiers Conference in Vancouver in January 2002. The Report calls for concerted national action aimed at

the development of a shared vision across jurisdictions and for the development of shared resources. In short, it is a call for a comprehensive, patient-centred framework to assist jurisdictions in maximizing the benefits offered by new technologies and to

10 See ss 14–15 regarding the functions and powers of the Embryo Research Licensing Committee.

11 See ss 33–41.

set paths for collaborative work to better understand and address the risks. A comprehensive framework, if developed, could move Canada and all provinces and territories into the forefront of preparing for the impact of genetics. This preparation will need to take several forms. There is a strong need for greater public engagement, for increased capacity in our health system to incorporate change, and for examining new ways in which we regulate and protect.¹²

5.12 The Report recommended the establishment of an ‘interjurisdictional coordinating body’¹³ or human genetics commission, along the lines of the UK Human Genetics Commission, to provide national leadership, oversight and regulation in this area:

The report urges governments to work together to ensure appropriate and comparable quality standards are in place across all jurisdictions providing genetic testing including: appropriate criteria for deciding when to test, monitoring processes for lab quality, protocols for ensuring appropriate counselling and support, and processes regarding test reviews for accuracy and reliability.

The report notes the need for appropriate capacity to monitor trends in medical genetics and assist all jurisdictions in addressing the ethical, legal and service delivery issues they will face. Stressing the need for a coordinated approach, the report suggests the possible creation of a human genetics commission to assist all jurisdictions.

The report also notes the importance of ensuring comparable quality assurance regimes and standards are in place and urges jurisdictions to cooperate in developing common approaches. In terms of federal review and approval processes, the report stresses the need for vigilance in the review and approval of new kit-based forms of genetic tests.¹⁴

5.13 The Canadian Government has accepted the Report as the basis for national (federal and provincial) work in genetics, and a working group (the Coordinating Committee on Genetics and Health) has been established to look at a range of implementation issues, including whether to establish the ‘interjurisdictional coordinating body’.

Adding a regional or international dimension?

5.14 The Inquiry’s Terms of Reference are focussed on determining the best regulatory framework for Australia, and the recommendations in this Report are geared towards that end. However, if at some time in the future the Federal Government saw an advantage in taking a regional or international approach to the protection of human genetic information, there is nothing in the pattern of recommendations that should

12 Province of Ontario, *Report to Premiers — Genetics, Testing and Gene Patenting: Charting New Territory in Healthcare*, <www.gov.on.ca/health/english/pub/ministry/geneticsrep02/report_e.pdf>, 20 February 2003.

13 Ibid, Recommendations 14–15, xxi.

14 Ibid, iv.

restrict this. Some submissions to this Inquiry noted that they increasingly operate internationally, at least in terms of sharing information, expertise and experiences.¹⁵

5.15 For example, the Human Genetics Commission of Australia (proposed below) could be ‘regionalised’, in the same way that Food Standards Australia New Zealand¹⁶ is a statutory authority that spans the Tasman, with responsibility for developing food standards based on scientific and technical criteria.¹⁷ The Australian and New Zealand governments have agreed in principle to establish a single trans-Tasman body to take over the combined roles of the Australian TGA and the equivalent New Zealand body (Medsafe) in regulating therapeutic goods.¹⁸

5.16 Similarly, if the Australian Government considered that it was a pacesetter in this field, it could move in time to share this expertise with other countries in the region, or internationally, along the lines of the work of the Australian Centre for International Agricultural Research—a statutory authority established with a mandate to mobilise Australia’s research capacity to help solve agricultural research problems in developing countries.¹⁹

The international trend towards standing advisory bodies

5.17 The Inquiry noted in DP 66²⁰—and it was remarked upon in many public meetings, consultations and written submissions—that it is somewhat ironic that the regulation of gene technology with respect to plants and animals has been formalised, providing the government with a source of high level advice in this area, but the same is not yet true with respect to human genetics.

5.18 In DP 66, the Inquiry referred to the clear international trend towards the establishment of standing bodies to advise governments on human genetics and biotechnology.²¹ These developments provide a number of different models for consideration—although care is needed in making direct comparisons, since each body has its own particular set of functions and priorities, relationships with other public authorities, pattern of membership, level of resources, and so on.

15 See, for example, Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.
 16 Previously known as the Australia New Zealand Food Authority, established under the *Australia New Zealand Food Authority Act 1991* (Cth).
 17 Parliamentary Secretary to the Federal Minister for Health and Ageing, *New Food Standards Australia New Zealand Launched*, <www.health.gov.au/mediarel/yr2002/tw/tw02020.htm>, 20 February 2003.
 18 See Therapeutic Goods Administration and Medsafe, *A Proposal for a Trans Tasman Agency to Regulate Therapeutic Goods*, <www.health.gov.au/tga/docs/pdf/tgprop.pdf>, 20 February 2003.
 19 *Australian Centre for International Agricultural Research Act 1982* (Cth). See also Australian Centre for International Agricultural Research, *About ACIAR*, <www.aciar.gov.au/about/aciar.htm>, 20 February 2003.
 20 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [3.48].
 21 *Ibid* [3.48]–[3.78].

5.19 In DP 66, the Inquiry described the nature and functions of a number of overseas standing advisory bodies operating in the area of bioethics and biotechnology. These included the European Union's Life Sciences High Level Group,²² the US President's Council on Bioethics,²³ and the Canadian Biotechnology Advisory Committee.²⁴ Having considered the various models, the Inquiry believes that the two most relevant ones for Australian purposes are the UK Human Genetics Commission and the Ontario (Canada) Advisory Committee on Genetics, which are described more fully below.

UK Human Genetics Commission

5.20 The United Kingdom Human Genetics Commission (HGC) was established following a comprehensive review by the British government in May 1999 of the regulatory and advisory framework for biotechnology, and replaces an earlier advisory committee. While that review indicated that the then existing system for regulating individual products and processes was operated satisfactorily, it was concluded that changes were needed to make the advisory framework:

- more transparent, in order to gain public and professional confidence;
- more streamlined, in order to avoid gaps, overlaps and fragmentation; and
- capable of dealing with rapid developments, and to be able take broad social and ethical issues fully into account.

5.21 The HGC now plays a key role in the UK's advisory and regulatory framework, including:

- assisting in the identification of gaps, overlaps, fragmentation or other problems, and ways of addressing them, by promoting coordination between bodies in the advisory and regulatory framework for human genetics;
- developing an overview of the regulatory and advisory framework, enabling the HGC to advise Ministers as needed on issues relevant to the framework as a whole;
- managing change, by providing information that will inform Ministers' decisions on the practical implications of advances, by identifying current and potential developments in human genetics and their implications for the National Health Service (NHS) and providing guidance as needed on general issues about the introduction and use of novel technologies, including in the NHS; and

22 Ibid [3.57].

23 Ibid [3.58]–[3.64].

24 Ibid [3.65]–[3.68].

- providing advice to Ministers, to inform decisions on broad social and ethical issues, in particular information on the current situation, likely developments and the views, wishes and concerns of the public and other stakeholders.

5.22 The HGC already has published a number of useful research and information documents, commissioned a survey of public opinion, and engaged in a significant level of public consultation.²⁵

5.23 The 22 part-time members of the HGC cover a wide range of expertise, interests and experience, including clinical genetics, genetic research, general medical practice, law, bioethics, theology, disability advocacy, nursing, pharmaceutical research, consumer protection, journalism, and family studies. The members include the chair of the Human Fertilisation and Embryology Authority and the nominees of the Chief Medical Officers from each of the four home counties. Appointments are usually for three years and may be renewed. The current Chair of the HGC is Baroness Helena Kennedy, a leading barrister, and the Deputy Chair is Professor Alexander McCall Smith, a leading authority on health and medical law.

5.24 In order to develop its projects, the HGC has operated through a number of Working Groups and Sub-Groups (comprised of HGC members and co-opted experts). These include the Working Group on the Storage, Protection and Use of Genetic Information (six HGC members plus one co-opted); the Working Group on Genetic Testing Services Supplied Direct to the Public (ten HGC members); the Public Involvement in Genetics Sub-Group (six HGC members plus three co-opted); and the Horizon-Scanning Sub-Group (four HGC members plus three co-opted).

5.25 In a particularly interesting initiative, the HGC also has set up a Community Consultative Panel of 106 people to provide the direct insights and experiences of people with a genetic disorder and to act as a sounding board for the HGC's reports and recommendations. The Panel includes people who have experience of single gene, chromosomal and multifactorial disorders, and of early and late onset disorders. Some Panel members are themselves affected, others are carriers or have experience as a parent of a child affected by a genetic disorder or as a carer for someone in the family who is affected.²⁶

5.26 The Secretariat for the HGC is provided by officials of the Department of Health and the Office of Science and Technology, and is based (and physically housed) within that Department.²⁷

25 Human Genetics Commission, *The Human Genetics Commission Website*, <www.hgc.gov.uk/>, 19 February 2003.

26 See the Human Genetics Commission website: Human Genetics Commission, *HGC's Consultative Panel of People Affected by a Genetic Disorder*, Department of Health (UK), <www.hgc.gov.uk/cpanel/index.htm>, 20 February 2003.

27 The budget for its operations in the financial year 2000/2001—not including the staff costs of the Secretariat—was approximately £250,000, plus specific project funding of £102,500. See Human Genetics Commission, *First Annual Report 2001*, <www.hgc.gov.uk/business_publications_annualreport_first.pdf>, 20 February 2003, 56.

Ontario Advisory Committee on Genetics

5.27 In April 2000, the Ministry of Health and Long-Term Care of the Canadian Province of Ontario established an Advisory Committee on New Predictive Genetic Technologies in order 'to help Ontario navigate the new frontier of human genetic medicine and science'.²⁸ Its mandate is to

develop a policy framework for introducing new genetic predictive testing and services into Ontario's health care system. This framework would help ensure that the provincial health system promotes wellness and improves health outcomes in advance of the appearance of disease. The Committee would also develop guidelines, principles, broad criteria and advice to guide decisions on how new genetic services should be incorporated into the province's health care system.²⁹

5.28 The multidisciplinary Advisory Committee included geneticists; a genetic counsellor; family physicians; genetic researchers; laboratory directors; academics in law, ethics and medicine; educators; a clinical epidemiologist; an expert in psychosocial issues; and representatives from the Canadian Cancer Society, the Heart and Stroke Foundation, the Huntington Society of Canada, the Ontario Association of Medical Laboratories, the Ontario College of Family Physicians, the Ontario Hospital Association, and officers of the Ministry of Health and Long-Term Care.

5.29 The work of the Committee was divided among six sub-committees, which were able to co-opt additional expertise, as needed: education; evaluation; clinical practice; psychosocial issues; laboratory practice; and legal and ethical issues.

5.30 The Report has a number of interesting things to say about the impacts of the new genetic technology on health systems, clinical practice, patients and the general community. However, the central recommendation was that:

the need for further consultation and implementation be recognized through an ongoing process. With this in mind, it is recommended that Ontario establish a Provincial Genetics Advisory Committee.³⁰

5.31 The suggested areas of coverage for this proposed permanent advisory committee include:

- new developments in the genetic sciences;
- evaluation of existing genetic services;
- recommendations on the timely provision of new genetic tests and services following formal evaluation of proposed genetic testing by the committee;
- legal and social issues;

28 *Report of the Provincial Advisory Committee on New Predictive Technologies (2001)*, Canada, <www.gov.on.ca/health/english/pub/ministry/geneticsrep01/genetic_report.pdf>, 20 February 2003, 1.

29 *Ibid.*

30 *Ibid.*, 70.

- human and infrastructure resource requirements for genetic services;
- educational needs for Ontarians, including professions involved in all aspects of the provision of genetic services;
- a process for the implementation of new genetic services that includes both public and private laboratories; and
- any other areas as requested by the provincial government.³¹

5.32 The Report also recommended that the permanent advisory committee's membership should be broadly constituted:

reflective of the broad based expertise necessary to achieve its mandate including geneticists, genetic counsellors, health economists, legal/ethical experts, epidemiologists, laboratory scientists, mental health professionals, and community representatives, including members of health related voluntary organizations.³²

5.33 Another issue considered was to whom the permanent advisory committee should report. Although it was acknowledged that many of the issues that will arise for the committee would fall within the jurisdiction of other departments, such as Education, Finance, and Consumer and Commercial Relations, it was decided that it would be best for the committee to be situated in the Ministry for Health and Long-Term Care, since its core responsibilities would be connected to patient interests and the improvement of the publicly funded health care system. The Report also sensibly proposed that the permanent advisory committee maintain close liaison with other relevant committees, such as those that deal with the nature and quality of laboratory services.³³

5.34 On 24 July 2002, the Minister for Health and Long-Term Care announced the establishment of the Ontario Advisory Committee on Genetics, with a brief to

provide advice to the government about the newest developments in genetic sciences and new genetic tests to benefit the people of Ontario. ... The committee's mandate will also include clinical evaluation of existing genetic services and recommendations on the timeliness of new genetic tests and services following formal evaluation of proposed genetic testing by the committee.³⁴

5.35 The foundation Chair of the Advisory Committee on Genetics is Dr Ronald Carter, President of the Canadian College of Medical Geneticists, and Head of Cancer Genetics for the Regional Genetic Services of South Central Ontario.

31 Ibid.

32 Ibid.

33 Ibid.

34 Ontario Ministry of Health and Long-Term Care, *Clement Names Chair of the Genetics Advisory Committee*, <www.gov.on.ca/health>, 27 July 2002.

A Human Genetics Commission of Australia

Is there a need?

5.36 It may be that there once was a time when the suggested solution to every significant social issue was the establishment of a specialised agency or tribunal to deal with that issue. However, as a general matter, it is now the ALRC's strong preference to utilise or build upon existing institutions wherever possible, and the Commission

is reluctant to recommend the establishment of a new body if the functions envisaged for [it] could be as efficiently and effectively carried out by existing agencies.³⁵

5.37 However, it is sometimes the case that the establishment of a new federal body is necessary and desirable,³⁶ such as where:

- there are no plausible institutional alternatives, even with a re-arrangement of functions or an addition of budget and staff for an existing body;
- such action represents an efficient and effective use of public funds; and
- the federal government has, or wants to have, a stake in the area and can contribute positively by providing leadership or coordination at the national level.³⁷

5.38 Everything written and said about emerging genetic science and technology—including in the public meetings of, and submissions to, the Inquiry—emphasises the rapid speed at which developments are taking place.

5.39 In IP 26, the Inquiry noted that:

The pace of biotechnological change equally affects Australia, and Australian governments arguably have the same need for ready access to the best possible advice about current and potential developments in human genetics and their implications for health care and human rights. ... [T]here is a continuing need for our laws, and the fundamental concepts underlying those laws, to be reviewed and revised to address advances made by human genetic science and technology. However, no such standing body on human genetics currently exists in Australia.³⁸

5.40 In the case of human genetics, the pace of change challenges the capacity of governments to develop sound policy and put in place effective regulatory strategies—although the same agreement could be made, for example, in relation to new information and communications technologies, the growth of the internet and related

35 Australian Law Reform Commission, *Managing Justice: A Review of the Federal Civil Justice System*, Report 89 (2000), ALRC, Sydney [4.78]. The discussion about 'federal agency non-proliferation' occurred in the course of concluding not to recommend the establishment of a new Federal Legal Services Forum, as had been mooted in the preceding Discussion Paper.

36 For example, within the Attorney-General's portfolio, the Government recently has funded the establishment of a National Judicial College of Australia and a National Pro Bono Resource Centre.

37 And only where, of course, the Commonwealth has constitutional authority to act.

38 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.158].

e-commerce applications. However, the ‘new genetics’ also contains special and further challenges for the way in which our society defines the ‘normal’, and the fundamental way in which people think about themselves and their own humanity.

5.41 In 2002, the American political philosopher Francis Fukuyama³⁹ wrote that:

Some new technologies are frightening from the start, and the need to establish political controls over their development and use is obvious to all. When the first atomic bomb was detonated at Alamogordo, New Mexico, in the summer of 1945, not one of the witnesses to this event failed to understand that a terrible new potential for destruction had been created. Nuclear weapons were thus from the very beginning ringed with political controls. Individuals could not freely develop nuclear technology on their own or traffic in the parts necessary to create atomic bombs, and in time, nations that became signatories to the 1968 non-proliferation treaty agreed to control international trade in nuclear technology.

Other new technologies appear to be much more benign, and are consequently subject to little or no regulation. Personal computers and the Internet, for example, promised to create wealth, increase access to information, and foster community among their users. ...

Biotechnology falls somewhere between these extremes. Transgenic crops and human genetic engineering make people far more uneasy than do personal computers or the Internet. But biotechnology also promises important benefits for human health and well-being. When presented with an advance like the ability to cure cystic fibrosis or diabetes, it is hard for people to articulate reasons why their unease with the technology should stand in the way of progress. It is easiest to object to a new biotechnology if its development leads to a botched clinical trial or to a deadly allergic reaction to a genetically modified food. But the real threat of biotechnology is far more subtle and therefore harder to weigh in any utilitarian calculus. It lies in the possibilities of human cloning, ‘designer babies’—eugenic selection for intelligence, sex, and personality—and eventually, the end of the human species as such.

In the face of the challenge from a technology like this, where good and bad are intimately connected, there can be only one possible response: We must regulate its development—and set up institutions that will discriminate between those technological advances that further human flourishing, and those that pose a threat to human dignity and well-being. These regulatory institutions must have the power to enforce these discriminations on a national and, ultimately, an international level.⁴⁰

5.42 Fukuyama—an unlikely champion of heavy-handed regulation, given his previous writings—went on to suggest that the new biotechnology raises so many challenges to the survival of society as we know it that formal regulation is required.

It has been a long time since anyone has proposed that what the world needs is more regulation. Regulation—and particularly international regulation—is not something that should be called for lightly. Before the Reagan-Thatcher revolutions of the 1980s,

39 The Bernard L Schwartz Professor of International Political Economy at the School of Advanced International Affairs, Johns Hopkins University. Professor Fukuyama is also a member of the President’s Council on Bioethics.

40 F Fukuyama, *Our Posthuman Future: Consequences of the Biotechnology Revolution* (2002) Profile Books, London, 181–182.

many sectors of the economies of North America, Europe, and Japan were vastly overregulated, and many continue to be so today. Regulation brings with it many inefficiencies and even pathologies. ... But in the end, there are certain types of social problems that can only be addressed through formal government control. ... Many people today believe that biotechnology should not and cannot, as a practical matter, be controlled. Both of these conclusions are wrong ...⁴¹

5.43 As noted above, Australian governments have chosen to take direct, concerted action to prohibit various activities, such as human reproductive cloning, and have proposed giving the new regulator, the Embryo Research Licensing Committee of the NHMRC, strong powers, with serious criminal penalties attaching to any intentional breach.

5.44 However, this Inquiry is concerned primarily with protecting individuals against the adverse effects of the misuse of genetic information (loss of privacy, unfair discrimination) and of unethical practices (loss of autonomy), rather than the potential direct threat to the future of the human genome that may be occasioned by certain types of experimentation. The balanced strategy the Inquiry is proposing relies more on providing high level advice to government policy-makers, and putting in place measures to ensure more careful and intelligent handling of genetic information and samples, with a number of targeted interventions in certain cases where the potential harm from the misuse or misunderstanding of genetic information is great, and the impositions are minor by comparison (for example, in most employment circumstances).

5.45 There is thus a critical need to ensure that the general community is much better informed about genetics and human health, and disabused of unbalanced notions of genetic determinism. While there is currently strong public support for genetic research, there is always the danger of a backlash if the promise of such work is over-sold or poorly understood. An editorial in the *British Medical Journal* in 2002 put the case well:

Over time, the tendency has been to expand diagnostic and treatment boundaries, and to include in the 'disease' category people with milder manifestations of pathology and lower levels of risk. Genetic tests for markers that may not result in symptoms for half a century or more could be new examples of a process of premature medicalisation of attaching the 'disease' label before it has been established that prevention or treatment is clearly beneficial. Treating the presence of a genetic marker as though it were the clinical disease can be very unhelpful. ...

Unless it is established that a genetic variant is a pointer to beneficial action, there is a potential for inappropriate medicalisation through the spread of poorly understood tests. The perceptions of risk resulting from such tests may bear little relation to the scientific facts and uncertainties. Inflated ideas about risks could result in people carrying such polymorphisms being treated unfairly in many areas, including employment or insurance. ...

41 Ibid, 183–184.

The antidote to genetics as a driver of medicalisation lies in remaining sceptical and level headed. Genetic claims, tests, and products should be treated in the same way that other medical markers and interventions are increasingly treated: with rigorous evaluation. The successful management of genetic medicalisation will depend on clinical evaluation, integrity, and transparency and on providing accurate information to consumers and patients. Public education about interventions based on genetic science will also be needed to prevent inappropriate social responses that may either lead to discrimination or, conversely, prohibit the adoption of tests and treatments that can reduce or prevent disability. Genetic technologies have the potential to be of major benefit to society, but their introduction must be measured, attentive to the social and ethical considerations of the day, and, most importantly, based on best evidence.⁴²

5.46 Later in this chapter, the enhancement of public and professional education about human genetics is assigned to the proposed new Human Genetics Commission of Australia as one of its areas of shared responsibility.

Submissions following IP 26

5.47 In IP 26 the Inquiry asked whether we need ‘our own version’ of the UK’s HGC.⁴³ The submissions strongly favoured such an approach on its own merits, as well as by way of contrast with the alternative of trying to develop legislative or regulatory prescriptions in a timely fashion. Common key points included the rapid pace of change in genetic science; the need for high level, independent advice in this area for governments, health care professionals, industry and individuals; and the need for an agency that would promote a consistent, national approach to the many important issues.

5.48 The Commonwealth Department of Health and Ageing submitted that:

A national mechanism or ‘standing body’ may be helpful in ensuring the provision of integrated advice to government on a range of human genetic matters. ...

All jurisdictions are grappling with the issues relating to the protection of health information generally. Given the potential to create a ‘railway gauge’ scenario of differing legislative regimes across jurisdictions, the Department strongly supports the adoption of a *nationally consistent* approach to the protection of health (including genetic) information. Notwithstanding the moves in this direction with federal privacy legislation, there is a need for national coordination and leadership in developing standards, guidelines, and codes of conduct, particularly in relation to the commercial uses of genetic information.

The NHMRC, through its Research Committee, Health Advisory Committee and AHEC, has important responsibilities in advising government in relation to public health standards, medical and public health research, and ethics. The AHMAC [Australian Health Ministers Advisory Council] has established a number of important mechanisms for developing national approaches to health care issues. Notwithstanding the valuable contributions of these existing advisory mechanisms, the Department notes that a national mechanism or ‘standing body’ may be helpful to

42 D Melzer and R Zimmern, ‘Genetics and Medicalisation’ (2002) 324 *British Medical Journal* 863.

43 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [2.159].

coordinate government advice and policy development on the complex issues raised by the rapid developments in human genetic information and technologies.

The recently established UK Human Genetics Commission and the breadth of matters within its purview provides one possible model that could be examined. Such a body could support a coordinated national effort to ensure that the potential benefits of emerging technological advances, particularly in relation to health, are realised for both individuals and the public while minimising any potential risks.⁴⁴

5.49 The Human Genetics Society of Australasia (HGSA), whose membership comprises clinical geneticists and those practising in laboratory-based human genetics in Australasia, wrote that:

The HGSA believes an independent Genetic Advisory Committee should be established to advise the Government in relation to the use of genetic testing in insurance and employment. There may also be a role for such a committee to provide ongoing advice to the Government on the complex issues, which will continue to arise from the rapidly evolving field of human genetics. It is recognised that existing government committees and working groups are considering various aspects of the impact of genetics in the health care system, and may make similar recommendations in the future. This committee might be envisaged to have a similar role to the UK Human Genetics Commission (HGC). The HGC was established following a comprehensive review of the regulatory and advisory framework of biotechnology. Its aim is to be transparent, in order to gain public and professional confidence; be streamlined, in order to avoid gaps, overlaps and fragmentation; ensure capacity to deal with rapid developments, and to take broad social and ethical issues fully into account within the field of human genetics.⁴⁵

5.50 The Australian Academy of Science noted that

this area of research is moving very quickly, and that laws can be difficult to amend if they no longer reflect the state of scientific knowledge and medical practice. Therefore, we urge the creation of an Australian Standing Advisory Body with both public and professional representatives to regulate issues in this area, similar to the UK Human Genetics Commission.⁴⁶

5.51 The Genetics Advisory Committee of the Victorian Department of Human Services supported the establishment of a standing body

to advise government on all aspects of human genetics, not just regulation/privacy. These issues could be viewed as a subset of the need to have national uniformity in population screening tests, indications and availability of other genetic tests, indications and availability of other genetic tests (eg whether through Medicare subsidy or through specific programs which receive block funding, eg the Family Cancer Genetics Service), a national approach to better information and training for health professionals as well as information to the public.⁴⁷

5.52 The Association of Genetic Support of Australasia wrote that:

44 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

45 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

46 Australian Academy of Science, *Submission G097*, 21 January 2002.

47 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002.

We believe a standing body to advise the government on issues related to human genetics should be established. It is important that its representation includes those with skills to address the breadth of issues generated by the new developments in technology. The UK and Canada have embraced this policy, recognising the need for continued assessment of the impact of the new genetics and the need for development of new policies and the revision of old to meet new challenges.⁴⁸

5.53 The Australian Medical Association submitted that:

It is important that a more uniform, national approach to the protection of human genetic information is developed. This is to ensure that all Australians are afforded the same rights and protection.

The AMA believes that gene technology has the potential to revolutionise both preventative medicine and the management of a vast range of medical conditions. We are keen to see the development in this country of a framework that provides for an acceptable medical use of genetic information based not only on the best available scientific evidence but also relevant social, ethical and legal considerations. ...

A standing body should be established to advise the government on issues related to human genetics. This body should have wide community representation to ensure that a diversity of opinions is considered.⁴⁹

5.54 The Institute of Actuaries of Australia, which has taken a strong interest in this area because of the potential effects of emerging genetic science on insurance,⁵⁰ submitted that:

Given the very rapid progress taking place in genetics it is likely that community views will also change rapidly. New issues are likely to emerge requiring fresh thinking on suitable controls or on the continued applicability of controls that are already in place. In times of rapid change, there would be benefit to the Government in having a standing body available to alert it to matters of interest and to provide advice, this body should include consumers and experts from all interested disciplines.⁵¹

5.55 Privacy NSW submitted that:

The establishment of such a body is critical, particularly whilst genetic privacy regulation in Australia remains undeveloped and as the efficacy of any new legislation is monitored. The model provided by the UK's Human Genetics Commission includes appropriate representation in terms of expert and lay membership.⁵²

5.56 The Office of the Federal Privacy Commissioner noted that:

there is a need for a body of sufficient authority and status to serve as a focus and as a paradigm for the work of our social institutions. A multi-disciplinary body would be capable of identifying and responding to the influences of, and the interplay between,

48 Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002.

49 Australian Medical Association, *Submission G091*, 29 January 2002.

50 See A Doble and others, *Genetics in Society 2001* (2001) Institute of Actuaries of Australia.

51 Institute of Actuaries of Australia, *Submission G105*, 7 March 2002.

52 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

science, the health and legal professions, research and commerce. With these capacities, it would be an invaluable source of advice for government.⁵³

5.57 The Australian Privacy Charter Council wrote that:

These issues are too important to only be the subject of a one-off inquiry, however thorough. We are attracted to the idea of a standing committee or commission, properly resourced and genuinely independent, to monitor developments, and to play an adjudicative role in relation to exceptional uses, retention, data linkage etc.⁵⁴

5.58 Support in similar terms came from the Neurofibromatosis Association of Australia; the Australian Society for Medical Research; the New South Wales Nurses' Association; the New South Wales Genetics Service Advisory Committee; Women's Health Victoria; the Australian Huntington's Disease Association (NSW); the Androgen Insensitivity Syndrome Support Group Australia; the Disability Discrimination Legal Service; the Queensland University of Technology's Human Research Ethics Committee; Sydney IVF Limited; the National Council of Jewish Women; and a number of individuals and support groups with particular interests and experience in this area.⁵⁵

Submissions following DP 66

5.59 In DP 66, the Inquiry made a specific proposal that 'a Human Genetics Commission of Australia (HGCA) should be established under federal legislation as an independent, stand-alone, statutory authority with sufficient resources to fulfil its mission'.⁵⁶

5.60 About 40 written submissions received in response to DP 66 directly addressed this matter, with almost all of them expressing support⁵⁷—often strong

53 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

54 Australian Privacy Charter Council, *Submission G120*, 18 March 2002.

55 Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; Australian Society for Medical Research, *Submission G124*, 18 March 2002; New South Wales Nurses' Association, *Submission G090*, 21 January 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Womens Health Victoria, *Submission G076*, 3 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002; Disability Discrimination Legal Service, *Submission G146*, 28 March 2002; Queensland University of Technology, *Submission G109*, 14 March 2002; Sydney IVF Limited, *Submission G062*, 14 January 2002; National Council of Jewish Women of Australia, *Submission G008*, 21 May 2001; H Wellmore, *Submission G064*, 5 January 2002; L Martinovic, *Submission G006*, 30 May 2001; J Torrens, *Submission G029*, 11 January 2002; *Confidential Submission G040CON*, 14 January 2002; F Richards, *Submission G044*, 14 January 2002; *Confidential Submission G051CON*, 14 January 2002; *Confidential Submission G058CON*, 13 January 2002; J Boyle, *Submission G057*, 14 January 2002; *Confidential Submission G142CON*, 25 March 2002.

56 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 3–1.

57 The NSW Police Service expressed concern that this 'would add another layer of bureaucracy to DNA testing in the law enforcement context'. See NSW Police Service, *Submission G306*, 22 January 2003. However, it is not proposed that the HGCA play a significant role in this area. See Part J of this Report.

support—for the proposal.⁵⁸ Other submissions *assumed* the establishment of an HGCA, referring to the role and importance of such a body in dealing with specific issues, such as the identification of ‘sensitive’ genetic tests, the approval of certain tests for use by the insurance industry or employers, and the enhancement of community and professional education in the area of human genetics.

5.61 The NSW Health Department submitted that:

Subject to agreement about funding arrangements the Department supports the proposal for a standing advisory body on human genetics as has been done in the UK, Canada and elsewhere. An overarching body such as the proposed Human Genetics Commission of Australia would achieve a coordinated approach to all issues. The Department notes that the regulation of gene technology with respect to plants and animals has been formalised through the Office of the Gene Technology regulator, but the same has not yet occurred with respect to human genetics.

The Department notes that the content of most of the ... proposals has been, or is currently, the subject of AHMAC and NPHP Working Groups concerned with genetic testing. An overarching body such as the proposed HGCA would be preferable in achieving a coordinated approach to all issues.⁵⁹

5.62 The Royal College of Pathologists of Australasia submitted that:

The College wishes to lend its support to the proposal to establish the HGCA. The rapidly advancing knowledge base of human genetics will continue to pose new questions and challenges for Australian society in the years to come. The proposed HGCA will enable the work of the Commission to continue in the future, so that

58 See generally, Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Australian Medical Association, *Submission G212*, 29 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Investment and Financial Services Association, *Submission G244*, 19 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; J Guameri, *Submission G210*, 28 November 2002; J Fleming, *Submission G241*, 20 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002; Eli Lilly Australia Pty Ltd, *Submission G247*, 19 December 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Law Society of New South Wales, *Submission G285*, 18 December 2002; Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Department of Health and Human Services Tasmania, *Submission G225*, 4 December 2002; Australian Huntington's Disease Association (NSW), *Submission G268*, 20 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002; Department of Human Services Victoria - Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Genetic Support Council WA, *Submission G243*, 19 December 2002.

59 NSW Health Department, *Submission G303*, 13 January 2003.

legislative frameworks and ethical safeguards can adapt to the social needs of the Australian community in response to these challenging issues.⁶⁰

5.63 Haemophilia Foundation Victoria wrote that:

As medicine and technology develop at a swift pace, it is essential that the government be advised as appropriate by a group of suitably qualified individuals who are fluent in human genetics, as well as in sociological, ethical and legal issues. HFV believes that a body such as the proposed Human Genetics Commission of Australia (HGCA) is essential to keeping governments informed regarding this dynamic, fast paced branch of science.⁶¹

5.64 In submissions and consultations the Inquiry also heard strong and consistent views that there is an urgent need for a more coordinated, national approach to harmonised laws and procedures to replace the existing, confusing ‘patchwork’; for better compliance and effective regulation,⁶² and for a coordinated national approach to related bioethical issues.

5.65 The Genetic Support Network of Victoria submitted that:

It is clearly evident that in a population of small numbers separated by vast distances such as we have here in Australia, people affected by rare genetic conditions will benefit from a national organisation.⁶³

5.66 Similarly, the Department of Health and Human Services Tasmania argued that:

the key reason for having a national body from the point of view of Tasmania, and we presume other smaller States and Territories, is that we do not have the capacity to develop, assess and provide genetic health services in isolation.⁶⁴

5.67 The Commonwealth Department of Health and Ageing affirmed their earlier support, stating that:

Given the rapid advances in genetic technology and the wide range of social, ethical and legal issues raised by new technologies, there is a need for a robust approach to responding to new issues to ensure better health outcomes. In the view of the Department a holistic approach to dealing with new developments has the value of ensuring that emerging issues are identified and responded to in a timely manner and that overlapping issues are dealt with in a consistent manner. It is the view of the Department that it is necessary to strive for a *nationally consistent* approach to

60 Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002.

61 Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002.

62 See eg Centre for Law and Genetics, *Submission G255*, 21 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002; N Stott Despoja, *Submission G198*, 27 November 2002.

63 Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

64 Department of Health and Human Services Tasmania, *Submission G225*, 4 December 2002.

privacy, ethical and discrimination issues. ... [T]he Department supports the establishment of a high-level advisory body to assist in these endeavours.⁶⁵

Establishment as a statutory authority

5.68 In the light of the consultations and submissions, the Inquiry believes that there are convincing grounds for establishing a standing body to deal with issues affecting human genetics in Australia. The Inquiry maintains its strong preference for such a body to be established as an independent, federal statutory authority, under the name of the Human Genetics Commission of Australia. Establishing the body in this form will:

- make clear its independence, and thus preserve public confidence in its findings and advice;
- encourage the public to communicate with the HGCA, even where the material is personal and sensitive and people might feel uncomfortable about communicating in this way with ‘the Government’;
- demonstrate its status as a permanent agency;
- demonstrate its status as a multidisciplinary body, whose work extends beyond a single government department;
- develop and maintain both in-house expertise and networks of experts and stakeholders;
- ensure sufficient budget resources to fulfil its mission; and
- provide it with sufficient operational flexibility to deploy its resources most effectively.

5.69 There is ample constitutional foundation for the legislation necessary to establish such a body. In common with many offices or bodies established by federal legislation (such as the OGTR and the Office of the Privacy Commissioner), the Commonwealth is able to rely on a suite of legislative powers for this purpose.⁶⁶ For example, the HGCA’s role in relation to insurance is supported by s 51(xiv) of the Constitution (‘insurance’), and its role in immigration by s 51(xxvii) (‘immigration and emigration’). In addition, there are federal powers in relation to trade and commerce (s 51(i)), corporations (s 51(xx)) and external affairs (s 51(xxix)), to name a few.

5.70 Even in the absence of specific legislative powers, the High Court has held that the Commonwealth has an implied nationhood power, by which it may ‘engage in enterprise and activities peculiarly adapted to the government of a nation and which cannot otherwise be carried on for the benefit of the nation’.⁶⁷ This power has been

⁶⁵ Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

⁶⁶ See *Gene Technology Act 2000* (Cth) s 13; *Privacy Act 1988* (Cth) Pt IV.

⁶⁷ *Victoria v Commonwealth* (1975) 134 CLR 338, 397 (Mason J).

held to be a possible basis for legislation establishing the Commonwealth Scientific and Industrial Research Organisation (CSIRO) and the Australian Bicentennial Authority,⁶⁸ and is particularly adapted to an organisation like the HGCA, which has educational and advisory functions in relation to matters of genetic science and human health. Moreover, under s 81 of the Constitution, moneys may be appropriated from Consolidated Revenue ‘for the purposes of the Commonwealth’, and this has been held to authorise the Commonwealth expending moneys ‘on inquiries, investigation and advocacy in relation to matters affecting public health, notwithstanding the absence of specific legislative power’.⁶⁹

5.71 Given the broad subject matter covered, the HGCA should have strong links with a number of relevant federal ministries, including the Department of Health and Ageing; the Attorney-General’s Department; Prime Minister and Cabinet; the Department of Education, Science and Training; and the Department of Employment and Workplace Relations.

5.72 However, given that statutory authorities normally are required to have a direct relationship with a particular department for budget and accountability purposes, the Inquiry agrees with the view of the Canadian authorities that this logically should be the Department of Health and Ageing. The central concerns of the HGCA will mainly relate to human health—and this is often true even where the policy advice is being applied in another context, such as employment.

5.73 As noted above, there was overwhelming support in the submissions and consultations for the proposition that the HGCA be established as an independent, statutory authority.⁷⁰

5.74 An alternative approach put to the Inquiry involved the establishment of the genetics advisory body as another Principal Committee of the NHMRC. In consultations, the CEO of the NHMRC, Professor Alan Pettigrew, was strongly of this view. The submission of the South Australian Department of Human Services also

68 Ibid, 397 (Mason J); *Davis v Commonwealth* (1988) 166 CLR 79.

69 *Victoria v Commonwealth* (1975) 134 CLR 338, 397 (Mason J). See also *Attorney-General of Victoria v Commonwealth* (1945) 71 CLR 237, 257.

70 See Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Australian Medical Association, *Submission G212*, 29 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Investment and Financial Services Association, *Submission G244*, 19 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Department of Health and Human Services Tasmania, *Submission G225*, 4 December 2002; Australian Huntington’s Disease Association (NSW), *Submission G268*, 20 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Children’s Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Genetic Support Council WA, *Submission G243*, 19 December 2002.

suggested this, noting that there are advantages in placing under the one umbrella such bodies as AHEC, the new Embryo Research Licensing Committee, and the HGCA.⁷¹

5.75 Earlier in this chapter, the Inquiry noted its strong preference not to establish a new government agency where an existing one could be expanded or adapted for the purpose, unless there were compelling reasons to the contrary. However, after considerable debate, the Inquiry has firmed in its view that establishing the HGCA as a committee of the NHMRC would be less satisfactory for a number of reasons. These include the fact that:

- the need for real *and perceived* independence is critical to winning public confidence—in an area in which there is significant anxiety about whether scientific and medical research is being properly regulated in the public interest;⁷²
- the role envisaged for the HGCA goes well beyond traditional ‘health and medical’ concerns, and extends to a very wide array of matters, such as insurance, employment, immigration, parentage, access to services and so on;
- establishment as an NHMRC Principal Committee would mean that the HGCA would be significantly less likely to gain the necessary resources to operate full-time and establish a high profile; and
- establishment as an NHMRC Principal Committee would mean that the HGCA would be significantly less likely to maintain continuity of programs and preserve long-term corporate memory—and, similarly, would be less likely to develop and retain expertise in-house amongst its continuing staff.

5.76 The submission from the Commonwealth Department of Health and Ageing strongly supported the establishment of a high-level advisory body focussing on the rapid developments in human genetic technologies. However, while the Department agreed that such a body needs to be independent and well-resourced,

it considers that establishment as a statutory body is not appropriate. Where the advisory body recommends that new regulations are required, these could be developed in cooperation with one of the existing regulatory authorities, professional bodies or industry associations and then administered by the relevant existing authority.⁷³

5.77 The Department appears to be equating ‘statutory authority’ with ‘regulatory authority’ in the submission, which is not the intention of the Inquiry. As noted above, the Inquiry accepts the view that the HGCA should be an advisory body, leaving any regulatory functions in this area to others. However, the Inquiry remains of the view that the best way to guarantee the standing and independence of the HGCA would be through establishment under its own Act of Parliament—in much the same way that

71 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

72 See Ch 4.

73 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

the ALRC is a federal statutory authority (but not a regulatory authority) constituted under the *Australian Law Reform Commission Act 1996* (Cth).

5.78 However, the Inquiry wishes to note that if our favoured approach (see Recommendation 5–1, below) is rejected by Government, the alternative approach of establishing a body with similar functions as a new Principal Committee of the NHMRC would be far preferable to leaving Australia without any expert advisory committee at this critical time in the development of genetic science and the consequent challenges and opportunities raised for our society.

Proposed functions

High-level advice on human genetics

5.79 Many submissions emphasised the need for governments and other policy-makers to receive cutting-edge advice about a range of complex issues raised by the rapidly developing field of human genetics, which integrates a broad range of expertise, experiences and perspectives.

5.80 The University of Tasmania’s Centre for Law and Genetics wrote that:

It is important that the final report recognises that the current rate of development of human genetics is accelerating and following new pathways. Just as an individual’s genetic profile produces a *future diary* (see Lori Andrews’ *Future Perfect*) future developments should be monitored and reasoned advice given. The proposed HGCA should develop the capacity to preview, monitor and respond to emerging issues in a strategic and planned fashion rather than react to issues. The research phase of human genetics will develop into the application phase. New issues will emerge and the proposed HGCA will have a pivotal responsibility for presenting advice that is coherent, comprehensive and reasoned.

5.81 A number of submissions and consultations pointed to the tension in proposals in DP 66, which primarily spoke of the body having an advisory role, but also appeared to suggest that the body might have some direct, regulatory functions⁷⁴—such as ‘approving specific genetic tests for use by the insurance industry for risk-rating purposes, or by employers for compelling occupational health and safety reasons’.⁷⁵

74 See eg Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Investment and Financial Services Association, *Submission G244*, 19 December 2002; Children’s Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

75 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 3–3.

5.82 The Inquiry agrees that this ambiguity needs clarification, and the Recommendations in this chapter have been refined to focus on the establishment of the HGCA as advisory body. To the extent that monitoring or enforcement is required in areas in which the HGCA provides advice, this should be left to other bodies, such as the TGA and the Human Rights and Equal Opportunity Commission, and to the courts and other complaints-handling mechanisms. Similarly, compliance might be achieved by the particular use that other bodies make of the HGCA's advice or recommendations.⁷⁶

5.83 In this respect, the standing body envisaged by the Inquiry would have a role along the lines of the UK's HGC and the Ontario Advisory Committee on Genetics. Accordingly, the Inquiry recommends that the role of the HGCA should be to provide:

- on-going, high-level, technical and strategic advice to Australian governments about current and emerging issues in human genetics;
- similar high-level advice on the ethical, legal and social implications arising from these developments, including consideration of any impact on human rights, and analysis of cost-benefit issues;
- national leadership in managing the process of change, including engagement of the public on these issues;
- relevant expertise and a consultative mechanism for the development of policy statements and national guidelines in this area, where appropriate in association with other governmental agencies or the relevant industries and organisations;
- assistance with the development and coordination of community, school, university and professional education about human genetics;
- advice and a consultative mechanism to assist the relevant bodies in identifying strategic priorities for research in human genetics; and
- a focus for the coordination and integration of various national—and perhaps regional and international—programs and initiatives.

5.84 The NHMRC has a range of statutory functions under the NHMRC Act, including funding health and medical research, issuing advice and guidelines on health issues, and developing ethical guidelines in regard to medical research and other ethical issues related to health. Part D of this Report notes the particular role of AHEC, as one of the NHMRC's Principal Committees, in supporting and overseeing human research ethics committees and in issuing guidelines that are binding on those committees and on researchers and institutions that receive NHMRC funding. The Inquiry wishes to make clear that there is no intention to develop a role for the HGCA

76 For example, in Ch 27 the Inquiry recommends that peak insurance bodies should require their members to comply with the recommendations of the HGCA regarding the use of particular genetic tests in underwriting.

which might conflict with the statutory functions of the NHMRC and its Principal Committees. As emphasised below, there will be a need for the HGCA to be alert to these matters and to consult widely as it develops its work plans.

Developing testing policy, especially in sensitive areas

5.85 Not all genetic tests raise the same ethical, privacy and discrimination concerns. Some genetic tests for diagnostic purposes may simply be more accurate and less invasive alternatives to discovering information that could be obtained by other means. For example, a buccal swab submitted for DNA analysis would be preferable in most respects to a liver biopsy performed to determine whether a person has haemochromatosis, or a ‘sweat test’ conducted on an infant to diagnose whether the child has cystic fibrosis. Many other current and future genetic tests, however, will fall into a ‘grey zone’, without clear indications whether or not they should be characterised as ‘diagnostic’ rather than ‘predictive’ or ‘research-related’ procedures.⁷⁷

5.86 Chapter 11 of this Report deals with issues relating to the regulation of access to genetic testing. Some of the Inquiry’s recommendations in this area turn on the existence of an independent, expert advisory body that can provide leadership in, among other things, identifying genetic tests that have particular concerns or sensitivities attached to them, and thus may require special treatment.

5.87 The existing framework for regulating therapeutic goods operated by the TGA provides an example of regulation that is carefully tailored to the sensitivities raised by particular products. Among other things, the TGA operates a classification system for medical devices by which the level of regulatory control is proportional to the degree of risk associated with the product. The system takes into account the benefits offered by use of the device, its intended purpose, and the effectiveness of the risk management techniques applied during design, manufacture and use.⁷⁸ By and large, this rating is done on technical, and increasingly internationally harmonised, ‘rules based’ standards. However, it is still open to the TGA to increase the risk classification for public policy reasons. For example, the Inquiry understands that silicon gel breast implants should be in ‘Class 2’ according to the technical rules, but lingering concerns about the safety experience with breast implants in Australia has led to the imposition of ‘Class 3’ status, with a higher level of scrutiny by the regulator.⁷⁹

5.88 Similarly, following advice from the Inter-Governmental Committee on AIDS, Hepatitis and Related Diseases, medical practitioners and dentists do not have access to new ‘rapid tests’ for HIV/AIDS or Hepatitis C because of the view that, in general, they do not have the expertise to read the test results accurately or to provide the proper counselling. Instead, test kits for HIV/AIDS and Hepatitis C are supplied only to laboratories that participate in the National Reference Laboratory’s Quality Assurance program for those tests.

77 See Ch 3 and Ch 10 on genetic testing and information.

78 See Therapeutic Goods Administration, *Australian Medical Devices Guidelines: An Overview of the New Medical Devices Regulatory System* (2002) Department of Health and Ageing.

79 Therapeutic Goods Administration, *Consultation*, Canberra, 28 May 2002.

5.89 It is possible that some genetic tests, including those provided directly to the public, could be seen to require special rules and processes.⁸⁰ The TGA has its own expert committees and advisory panels, but discussions with senior officials has indicated that the TGA would welcome the recommendations of an independent, national advisory body on human genetics to assist it with setting the policy parameters for regulation in this area.

5.90 Official reports in the United States⁸¹ and Canada⁸² have recognised that the approval process for genetic testing technology should go beyond a simple technical assessment of the product and include a second compulsory stage, as such tests are ‘likely to raise many clinical, ethical or legal issues [that] would require a rigorous and formal evaluation by a multidisciplinary team’.⁸³ The Canadian report also notes that special concerns are raised by the possible availability in future of kit and home-based testing through direct-to-consumer marketing and over the Internet. The report suggests that these issues would be appropriate for policy advice, education and oversight by a national coordinating body, such as a human genetics commission.⁸⁴

5.91 The HGSA stated that an HGCA could play an important role in

identifying genetic tests that have particular concerns or sensitivities attached to them, and may thus require special treatment. In carrying out this responsibility, HGCA would work closely with existing regulatory bodies [such as the TGA and NPAAC] to develop recommendations for clinical and laboratory practice, including mechanisms such as restricted clinical request pathways, to ensure that genetic tests are delivered with appropriate counselling, consent and attention to privacy.⁸⁵

5.92 The Genetic Support Network of Victoria submitted that an HGCA would be well-placed to fulfil this role if it incorporated the views and experiences of genetic support organisations, through membership and consultation:

We see a role for genetic support organisations in assisting to identify genetic tests that have particular concerns or sensitivities attached to them, as some concerns and sensitivities may not be evident to people who have not had a particular test themselves. While professionals may well identify the various clinical, scientific, economic and psychological issues which could arise, there may be other personal

80 See Ch 11.

81 N Holtzman and M Watson, *Promoting Safe and Effective Genetic Testing in the United States: Final Report of the Task Force on Genetic Testing* (1997), National Institutes of Health — Department of Energy Working Group on Ethical, Legal and Social Implications of Human Genome Research.

82 Province of Ontario, *Report to Premiers — Genetics, Testing and Gene Patenting: Charting New Territory in Healthcare*, <www.gov.on.ca/health/english/pub/ministry/geneticsrep02/report_e.pdf>, 20 February 2003.

83 Ibid, 79. See also I Blancquaert and others, ‘Oversight Mechanisms for Technology Transfer in Molecular Genetics’ (2001) 2 *ISUMA Canadian Journal of Policy Research* 103.

84 Province of Ontario, *Report to Premiers — Genetics, Testing and Gene Patenting: Charting New Territory in Healthcare*, <www.gov.on.ca/health/english/pub/ministry/geneticsrep02/report_e.pdf>, 20 February 2003, iii and viii.

85 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002. See also New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

and social issues which are more complex when placed in the combined context of family, health service provision, insurance options, education and other factors.⁸⁶

5.93 The Centre for Law and Genetics also called for the HGCA to assess genetics tests with specific reference to cost benefit analysis, human rights impact, and strategic priorities in the delivery of genetic services.⁸⁷ The South Australian Department of Human Services also noted that the strategic decision to utilise certain genetic tests should be well informed and ‘not influenced by commercial or other interests’.⁸⁸

5.94 Senator Natasha Stott Despoja submitted that the HGCA should subject tests used in population genetic screening programs to particular scrutiny, writing that:

Tests used in population genetic screening programs must meet an agreed standard for reliability, sensitivity and utility. All genetic screening tests should comply with strict standards set by one peak body such as the proposed HGCA. Compromise has the potential to undermine the entire legislative package. The potential costs to the health care system, the potential harm caused by diagnosis where necessary counselling is not available, the potential for wrong test results, and the potential for test results to be used negatively for insurance and employment purposes make regulation governing the reliability, sensitivity and use of population genetic screening programs an absolute necessity.⁸⁹

Advice in other key areas

5.95 The Inquiry sees a key role for the HGCA in providing expert policy advice to governments, industry, service providers and others about the sensible uses of genetic information and testing technology across a range of subject areas, such as insurance and employment, both from the technical standpoint as well as in consideration of the ethical, legal and social implications.

Insurance

5.96 As discussed in Chapter 27 of this Report, in the United Kingdom, the Genetics and Insurance Committee (GAIC) has the role of determining applications for the approval of specific genetic tests for use by the insurance industry, having regard to the scientific reliability and actuarial relevance of the test. GAIC comprises representatives from the insurance industry, actuaries, scientific community, and general community. Under the British insurance industry’s voluntary code, insurers will only take account of genetic tests that have been authorised for this purpose by GAIC.⁹⁰ Such authorisation has been granted for a number of genetic tests.⁹¹

86 Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

87 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

88 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

89 N Stott Despoja, *Submission G198*, 27 November 2002.

90 Association of British Insurers, *Genetic Testing: ABI Code of Practice* (1999), Association of British Insurers, London, Principle 33.

91 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [11.163]–[11.168].

5.97 No such body currently exists in Australia, although many submissions stressed the need for this sort of function to be performed here, including those from the Australian Academy of Science; the HGSA; the Australian Medical Association; the Genetic Support Network of Victoria; NSW Legal Aid; and Senator Stott Despoja.⁹²

5.98 The Centre for Law and Genetics submitted that:

The prospects for ensuring that accurate and reliable information is uniformly available to agents and brokers would be greatly enhanced if this responsibility was shared between the insurance industry and government, through the work of an expert committee established for the specific purpose of evaluating the scientific and actuarial relevance of genetic tests proposed for use by the insurance industry in setting insurance premiums, along the lines of the Genetics and Insurance Committee (GAIC) established in the United Kingdom.⁹³

5.99 The Australian Huntington's Disease Association (NSW) agreed that there is a need for an 'independent advisory committee appropriate for use/interpretation of genetic tests and information for insurance purposes'.⁹⁴

5.100 The Anglican Diocese Sydney, submitted that it was

concerned about possible discrimination in use of genetic information, particularly in view of the current understanding of the actuarial significance of some information. We believe that those tests to be used for insurance and the way they are used should be approved by scientists and not the industry, ie by the proposed HGCA.⁹⁵

5.101 Privacy NSW also suggested that the advisory committee should review the use of genetic test and family history information by insurance companies in light of advances in genetics,⁹⁶ and recommended that:

the use and disclosure of genetic information should be prohibited in general and health insurance until specific tests are approved by an independent review body, such as the proposed Genetics Advisory Committee.⁹⁷

5.102 As noted above, a number of submissions emphasised that the HGCA should not be both an advisory body and a regulator. In this particular context, the Queensland Government wrote that:

The need to develop detailed policy advice regarding which genetic tests should be approved for use by the insurance industry for risk-rating purposes, or by employers for compelling occupational health and safety reasons is acknowledged. However, the

92 Australian Academy of Science, *Submission G097*, 21 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Australian Medical Association, *Submission G212*, 29 November 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; N Stott Despoja, *Submission G198*, 27 November 2002.

93 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

94 Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002.

95 Anglican Diocese of Sydney, *Submission G256*, 20 December 2002.

96 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

97 *Ibid.*

proposal that the HGCA have responsibility for approving such tests appears to be inconsistent with its proposed advisory role. It may be more appropriate for the proposed HGCA to provide advice to policy-makers.⁹⁸

5.103 The peak association representing life insurers and other financial advisers in Australia, the Investment and Financial Services Association (IFSA), accepted the value of an independent, expert body providing advice about the intelligent use of genetic testing and information in the insurance industry, but rejected the need for the HGCA to play a formal regulatory role. Responding to the reference in DP 66 to the HGCA playing a ‘monitoring role’ with respect to the industry, IFSA submitted that the HGCA instead should be

undertaking a ‘research’ rather than a ‘monitoring’ role to keep abreast of developments in the industry with respect to the use of genetic information; and a role in reviewing and, where appropriate, disallowing existing or approving new genetic tests on appropriate grounds. It is acknowledged that the insurance industry would only seek disclosure of the results of relevant and appropriately approved genetic tests.⁹⁹

5.104 In Chapter 27 the Inquiry supports the view that the HGCA should assess the use of genetic tests in underwriting and that it should make recommendations regarding their use for that purpose. Rather than ‘approving’ or ‘disapproving’ tests, as DP 66 had suggested, the Inquiry believes that the HGCA’s recommendations can be given effect through industry codes of practice.

Employment

5.105 A number of submissions drew together the need for expert advice in the area of insurance with emerging concerns about the potential for the use and misuse of genetic testing by employers. For example, the Centre for Law and Genetics argued that:

the legitimacy of requiring genetic testing in a given situation should itself be the subject of independent review to ensure that the criteria for justifying genetic have been established. Further, it is essential that the testing undertaken is reliable and accurate and that an objective, scientifically well founded assessment is made of that test result. Responsibility for regulation and oversight of the use of genetic testing should be vested in an independent body with multi-disciplinary expertise specifically set up for this purpose.¹⁰⁰

5.106 The Haemophilia Foundation Victoria wrote that it was

particularly pleased that the proposed HGCA would have responsibility for approving specific genetic tests that may be used by the insurance industry and employers. This is an area of particular concern to the Foundation and as such the Foundation is

98 Queensland Government, *Submission G274*, 18 December 2002. See also Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

99 Investment and Financial Services Association, *Submission G244*, 19 December 2002.

100 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

delighted that such a body will oversee the appropriate and responsible use of this information in insurance and employment settings.¹⁰¹

5.107 The Anti-Discrimination Board of NSW submitted that:

In our view the HGCA's role in relation to policy statements and national guidelines must include development of a national testing policy in broader terms than outlined in proposal 3-3. Such a policy should include employment related testing, pre and post test discussions to enable informed consent to testing and issues in relation to insurance companies access to or use of genetic tests. In addition to testing issues in insurance and employment, we strongly agree that the HGCA should have a policy development role in critical areas such as insurance and employment generally.¹⁰²

5.108 In Parts G and H of this Report, the Inquiry considers in much greater detail the use of genetic testing and information in the contexts of insurance and employment, and a number of specific recommendations for reform are built around the establishment of an HGCA which can provide this kind of technical and policy advice.

Public and professional education

5.109 There is little doubt that, for the benefit of the entire community, the revolutionary progress in genetic science needs to be matched with a major effort aimed at significantly raising public and professional awareness and understanding of these advances, and their ramifications for policy and practice.

5.110 There has been some criticism of the Human Genome Project for spending billions of dollars on scientific research, and some considerable amount on bioethics,¹⁰³ but very little on public education. The Director of Roche Genetics, Klaus Lindpainter, has argued that public education

is as much a key challenge to researchers as linking genes to function. The lack of public understanding will slow the introduction of breakthroughs on the scientific side and slow the drive to put data and patient protection mechanisms in place as genomic research moves from the lab to the clinic. All of this will rely on all of us engaging in a dialogue with the public.¹⁰⁴

5.111 The Ontario Advisory Committee on New Predictive Genetic Technologies recommended developing and promoting a genetics educational program for everyone in that Province, including health professionals and policy makers, to meet public and professional needs. The Committee also recommended developing a specific education program around each new predictive genetic test approved as an 'insured service.'¹⁰⁵

101 Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002.

102 Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002.

103 About 3–5% of the budget of the public consortium involved in the Human Genome Project was dedicated to exploring the ethical, legal and social implications (ELSI) of the medical and scientific research. See Human Genome Project, *Ethical, Legal, and Social Issues Research*, National Institutes of Health—Department of Energy Working Group on Ethical, Legal and Social Implications of Human Genome Research, <www.ornl.gov/TechResources/Human_Genome/research/elsi.html>, 10 March 2003.

104 K. Howard, *Human Genome Project "Flat on its Face", Must Educate the Public*, <www.genomeweb.com/articles/view.asp?Article=20026418210>, 20 February 2003.

105 That is, one for which a rebate may be claimed under the provincial public health insurance scheme.

Until recently, much of the practice of genetics involved diagnosing rare inherited disorders, estimating risk for family members, and providing prenatal diagnosis. There was little need for most health care providers to have any more than a rudimentary knowledge of genetics. Now, there is an urgent need for the Ministries of Education and of Colleges and Universities to review the curricula of secondary and post-secondary schools and incorporate core genetic issues.¹⁰⁶

5.112 Many submissions to the Inquiry also stressed the need for a concerted effort at greater public and professional education in this area, and a number linked this to the establishment of an HGCA. For example, the Genetics Advisory Committee of the Victorian Department of Human Services wrote that a standing advisory commission should lead ‘a national approach to better information and training for health professionals as well as information to the public’.¹⁰⁷ Women’s Health Victoria submitted that:

There is definitely a need to educate health professionals better about practical and ethical principles involved in genetic testing and information. This could be one of the functions of the standing body on human genetic information.¹⁰⁸

5.113 The Department of Health and Human Services Tasmania also wrote to

note and support the proposals concerning public and professional education on human genetics. We expect that this education will result in increased requirements for services such as genetic testing and counselling. We have seen this in a small way with local education initiatives for professionals in the area of cancer genetics. The informed professionals are more likely to request counselling and testing services for their patients.¹⁰⁹

5.114 The Office of the Federal Privacy Commissioner wrote that:

To achieve the necessary social acceptance of genetic and technological advances, it will be essential for our institutions to operate within the following ‘learning framework’, which should encourage them to:

- keep abreast of advances in genetic knowledge, learning from their own experience and the experience of the scientific and medical applications of advances in genetic knowledge;
- in particular, ensure that health professionals and medical researchers learn from the community support groups representing those with genetic disorders in developing policies and strategies;
- incorporate that knowledge and experience in their decision-making processes;
- ensure that their decisions are transparent and accountable;

106 *Report of the Provincial Advisory Committee on New Predictive Technologies (2001)*, Canada, <www.gov.on.ca/health/english/pub/ministry/geneticsrep01/genetic_report.pdf>, 20 February 2003.

107 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002.

108 Womens Health Victoria, *Submission G076*, 3 January 2002.

109 Department of Health and Human Services Tasmania, *Submission G225*, 4 December 2002.

- provide the community with best possible means of understanding developments in genetic knowledge and of participating fully in the decisions which may dramatically affect their lives;
- be receptive and responsive to community perceptions, concerns and to the promotion of their legitimate interests; and
- regard as paramount an ethical approach to all their activities and outcomes.¹¹⁰

5.115 The Androgen Insensitivity Syndrome Support Group Australia wrote that the HGCA should ‘also be responsible for considering potential avenues of education of the wider community about the uses of genetic information’.¹¹¹ The Neurofibromatosis Association of Australia submitted that:

We see the role of such a body as not just participating in the regulatory framework, but also as being a focus for public education on issues related to human genetics. We see the creation of such a body as being an important outcome of this current inquiry.¹¹²

5.116 The Association of Genetic Support of Australasia wrote that it was

pleased to note the importance placed in this proposal and we would like to emphasise how important it is that the HGCA liaise with the public, especially community support groups and umbrella organizations on managing the process of change and in developing policies. This will ensure that directives e.g. policies education etc. match what is needed in practice at the grass roots level. We agree with the development of community, school, university and professional education about human genetics and agree the focus must be first on a national level which in turn is linked internationally.¹¹³

5.117 Although the HGCA will be in a good position to promote, advise and assist educational initiatives, this will be more at the ‘big picture’ level: it is not anticipated that this body necessarily will be a direct provider of education. As the submissions noted, there is already substantial activity in genetics education in Australia at many levels. This will be another area of shared responsibility in which the HGCA will need to work closely with other groups and institutions, such as departments of health and education; the HGSA; state and territory school curriculum authorities; university medical schools; the Royal Colleges; the Divisions of General Practice, and so on. Professional education on genetics is dealt with in more detail in Chapter 23.

Strategic priorities for research

5.118 One of the terms of reference for the UK HGC is ‘to advise on strategic priorities for research’. The Centre for Law and Genetics noted that:

110 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

111 Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002.

112 Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002.

113 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

The NHMRC is the appropriate body for settings strategic projects for health and medical research. The Australian Research Council does have a strategic function in ELSI research but it may be advisable to include [in] the terms of reference of the HGCA something along the lines of: to advise, after consultation with relevant research funding organisations, on strategic priorities for research in human genetics.¹¹⁴

5.119 The Inquiry agrees with both aspects of the Centre's submission. It is certainly *not* envisaged that the HGCA would seek directly to set research priorities—this is quintessentially the role of the NHMRC and Australian Research Council (ARC) (and, of course, the Government, where it provides funds to support research in specified areas of national importance, as it does from time to time).

5.120 However, in the course of its work as a body dedicated to the study of a wide range of issues surrounding human genetics, the HGCA no doubt will be in an excellent position to assess emerging issues in human genetics and observe the state of existing research in Australia in these areas—including the gaps. The Inquiry believes that the transmission of this experience and awareness to the NHMRC and the ARC would be valuable in assisting those bodies to make better informed decisions about research needs and priorities in human genetics.

Own motion inquiries and ministerial references

5.121 As a standing expert body, the HGCA should be available as a national resource to be utilised for inquiry into, or the provision of expert advice on, matters relating to human genetics—whether on its own motion or upon the formal reference of the responsible minister or ministers. In a fast-moving area of scientific development there will no doubt be matters arising over time which will require sensible, expert commentary and advice, both to inform the community and to promote sound public policy-making.

5.122 For example, this Report does not deal with matters relating to 'behavioural genetics'—an undeveloped and unproven area of research, which suggests that certain behavioural traits, such as sexual orientation, political orientation, alcohol and drug addiction, or propensity to violence, are genetically determined to a significant degree. Of course, any labelling of 'abnormal' traits would have to proceed from a clear demarcation of what is regarded as the 'normal' range of human behaviour. The Nuffield Council on Bioethics in the United Kingdom aptly has described this as

a complicated area of research in genetics, often controversial, occasionally explosive and with the capacity to ignite dangerous passions.¹¹⁵

5.123 This form of genetic determinism (see Chapter 3) does not presently figure in legal or social policy-making, and the Inquiry was not presented with any related complaints or practical problems. However, it is conceivable that in the future employers might seek to discriminate based upon notions of behavioural genetics ('we

114 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

115 Nuffield Council on Bioethics, *Genetics and Human Behaviour: The Ethical Context* (2001), Nuffield Council on Bioethics, London, 5.

can't hire people with a propensity towards violence in our industry'), or an accused person might seek to deflect or diminish criminal responsibility because of genetic factors ('my genes made me do it'), and so on (see Chapter 44). In that event, an independent, authoritative, national body like the HGCA would be perfectly placed to lead community education and debate on the issue.

Monitoring the implementation of this Report

5.124 Although the HGCA will develop its own initiatives and priorities over time, the Inquiry believes that it will be a useful starting point for the HGCA to undertake an active program of monitoring the implementation of the broad strategies and specific recommendations made in this Report. In order to assist this process, the Inquiry has included an 'Implementation Schedule' in this Report, indicating the body or bodies responsible for implementing each recommendation.

Structure of the HGCA

5.125 Given the wide-ranging brief of the HGCA and the probable substantial size of the potential membership group (see below), an effective structure might resemble that of the NHMRC, which involves a central Council with substantial diversity,¹¹⁶ and a number of principal committees,¹¹⁷ as well as other committees and working groups established from time to time for specific purposes.

5.126 In DP 66, the Inquiry suggested that:

Without wishing to be overly prescriptive at this stage, it appears to the Inquiry that the HGCA would need at least two principal committees of its own, including:

- a Technical Committee—to provide the required technical, scientific and medical advice needed; and
- an Ethical, Legal and Social Implications Committee—to ensure that such matters and a broad range of social perspectives are always given full consideration.

Most, if not all, of the policy advice sought from the HGCA will involve a mix of these issues—for example, the identification of 'sensitive' genetic tests that require restricted access or additional counselling and support; advice to the TGA about risk classification; and advice to insurers and employers about permissible uses and interpretations of genetic testing—so that the Committees often will need to sit together, or at least contain some degree of cross-membership.¹¹⁸

116 *National Health and Medical Research Council Act 1992* (Cth) s 20 stipulates that the membership must contain persons with a background, knowledge or expertise in indigenous health needs, health care training, medicine, nursing, science, the trade union movement, business, consumer issues, social welfare services, environmental issues and public health issues. Under s 21, the Minister must seek nominations from the relevant bodies in each category before making the appointment.

117 Chairs of the principal committees are automatically members of the Council: *Ibid* s 20(c). The number of principal committees has increased to five with the establishment of the Embryo Research Licensing Committee, as discussed above.

118 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, [3.152]–[3.153]. See also Proposal 3–4.

5.127 The submissions strongly supported this general approach.¹¹⁹ For example, the Centre for Law and Genetics wrote that:

The Proposal to establish at least a Technical and ELSI Committee follows the pattern of the *National Health and Medical Research Council Act 1992* (Cth) and the *Gene Technology Act 2000* (Cth). Both Acts include provision for the establishment of principal committees. A Technical and an ELSI Committee are good candidates. We presume that the legislation would also include provision to appoint other principal committees, in the manner of the NHMRC legislation.¹²⁰

5.128 The Commonwealth Department of Health and Ageing agreed that:

The function proposed for a high-level advisory body and the range of issues which need to be addressed would warrant the establishment of a Technical Committee and an Ethical, Legal and Social Implications Committee. A broad range of expertise and a process of community consultation would assist in fostering community confidence in the proposed body. Experience in other areas of biotechnology suggests that community confidence in expertise and independence of the advisory body would be critical to public acceptance of action based on its recommendations. Strict conflict of interest guidelines would have to be developed for each committee.¹²¹

5.129 A number of submissions also proposed the addition of another standing committee or two. Several noted the strong emphasis on education throughout DP 66, and suggested this should be reflected in the committee structure of the HGCA. For example, the HGSA wrote that:

Throughout the discussion paper, education has been highlighted as a major function of the HGCA. An Education Committee should be added as a principal committee.¹²²

5.130 Similarly, the Centre for Genetics Education submitted that:

Given the focus that education has been given in the Discussion Paper, the HGCA would benefit from having an Education Committee as a separate principal committee in addition to those proposed. Education will need to address technical, ethical, legal and social issues.¹²³

119 See eg Australian Medical Association, *Submission G212*, 29 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

120 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

121 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

122 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

123 Centre for Genetics Education, *Submission G232*, 18 December 2002. See also Department of Health Western Australia, *Submission G271*, 23 December 2002.

5.131 Several submissions also called for the HGCA to have a separate, formal, community consultative committee, along the lines of that of the OGTR in Australia. For example, Privacy NSW submitted that:

The proposed structure for the HGCA should include a mechanism to provide for due weight to be given to the views of community groups including consumer and minority groups. For this reason, we favour the structure of the Office of the Gene Technology Regulator which includes a broad-based community committee, the Gene Technology Community Consultative Committee (GTCCC). A similar approach should be adopted in relation to the regulation of human genetics technology.¹²⁴

5.132 The NSW Health Department suggested that the HGCA should also encompass a committee on clinical genetic services:

Principal committees should also include clinical services. Medical practitioners and genetic counsellors are at the forefront of patient access to genetics information and services.¹²⁵

5.133 The Inquiry agrees that education will be an important aspect of the work of the HGCA, and that body will have important things to say about the effective and efficient delivery of clinical genetic services. The Inquiry also agrees that engagement of the general public, and involvement of the genetic support community in particular, will be vital to the success of the HGCA.

5.134 However, the Inquiry prefers to leave such detailed organisational arrangements for the HGCA itself. For example, rather than waiting for ministerial appointments to a principal committee on education, the HGCA may prefer the flexibility of making its own staff appointments to an internal Education Unit, or it may prefer to establish an educational working group or advisory committee, or commission particular educational projects from other providers.

5.135 Similarly, the HGCA may wish to follow the UK HGC's approach of establishing, through its own initiative, a large, voluntary Community Consultative Panel, rather than having a statutorily-prescribed Community Consultative Committee along the lines of the OGTR. Although no one disputed the importance of community consultation, the Inquiry heard differing views about the effectiveness of the OGTR model during its own consultations. For example, some people suggested that the functions of the Community Consultative Committee and the Ethical, Legal and Social Issues (ELSI) Committee were not clearly differentiated, especially where the latter contained community and consumer representatives. Others commented that the three committees needed to interact more frequently, and that the ELSI and technical committees needed to hear community perspectives more often, but that this was made more difficult by the organisational structure.

124 Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002. See also Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

125 NSW Health Department, *Submission G303*, 13 January 2003.

5.136 The Inquiry also is mindful of the efforts by the NHMRC to improve communication and consultation with Aboriginal and Torres Strait Islander communities. After some years of experimentation with different models, the NHMRC recently has opted against having a separate committee dealing with indigenous health issues, and instead adopted the approach of promoting coordination among indigenous representatives across all of the NHMRC committees, through the new Indigenous Health Forum.

5.137 No doubt other issues will arise from time to time in which the HGCA may believe that the best way to inform itself—or to promote community education, consultation and debate—will be through the establishment of an ad hoc committee or working group. The Inquiry believes that the formal structure of the HGCA should be open enough to enable it to be responsive to emerging issues and to operate flexibly in this manner.

Membership of the HGCA

5.138 Overwhelmingly, submissions to the Inquiry have strongly emphasised the need for balanced and broad-based membership, with both expert and community representation.¹²⁶ Similarly, a public opinion survey conducted by the University of Western Australia in 2000 found strong support for an advisory group that comprised members of the general public as well as scientists, medical professionals and others.¹²⁷

5.139 The Commonwealth Department of Health and Ageing submitted that:

Should such a body be established, its composition should be broadly based and might include medical practitioners, ethicists, researchers, geneticists, privacy and anti-discrimination expertise, insurance and actuarial expertise, genetic counsellors and educators, lawyers, consumer representatives, disability advocates and media representatives.¹²⁸

126 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Australian Medical Association, *Submission G212*, 29 November 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; *Confidential Submission G040CON*, 14 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; New South Wales Nurses' Association, *Submission G090*, 21 January 2002; Australian Medical Association, *Submission G091*, 29 January 2002; Australian Academy of Science, *Submission G097*, 21 January 2002; Institute of Actuaries of Australia, *Submission G105*, 7 March 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002; Queensland University of Technology, *Submission G109*, 14 March 2002; Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; Australian Society for Medical Research, *Submission G124*, 18 March 2002; Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002; *Confidential Submission G142CON*, 25 March 2002; Disability Discrimination Legal Service, *Submission G146*, 28 March 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

127 University of Western Australia Survey Research Centre, *Attitudes Towards Human Genome Epidemiology*, University of Western Australia, <www.gshh.uwa.edu.au/survey.html>, 19 February 2003.

128 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

5.140 The Disability Discrimination Legal Service (DDLS) wrote that:

The DDLS considers that such a standing body should be established and that it includes researchers with specific expertise in genetic counselling and ethical concerns, representation that reflects the special and direct ways in which people with disabilities are affected by human genetic information, as well as broad community representation. Essential with this representation is the need to have consumer representation by groups directly affected by human genetic information—the ‘nothing about us without us’ principle. Great care should be applied to balancing representation of such a body to ensure that insurance and industry representatives do not have undue influence, particularly in consideration of issues relating to discrimination based on the use of human genetic information.¹²⁹

5.141 The Genetic Support Network of Victoria added that:

We believe that people affected by genetic conditions are the most important stakeholders in a standing advisory body on human genetics (because if these people did not have genetic conditions, there would be little need for such an advisory body). Therefore we would expect to see people affected by genetic conditions well represented on the HGCA, with more than just ‘token’ representation. A national alliance of support organisations should certainly provide representatives for the Ethical, Legal and Social Implications [Committee].¹³⁰

5.142 Other submissions highlighted the need to include:

- clinical geneticists and medical practitioners;¹³¹
- genetic counsellors;¹³²
- health and medical researchers;¹³³

129 Disability Discrimination Legal Service, *Submission G146*, 28 March 2002.

130 Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

131 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Australian Medical Association, *Submission G212*, 29 November 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; *Confidential Submission G051CON*, 14 January 2002; Australian Medical Association, *Submission G091*, 29 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

132 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Centre for Genetics Education, *Submission G232*, 18 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; *Confidential Submission G051CON*, 14 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

133 Sydney IVF Limited, *Submission G246*, 19 December 2002; F Richards, *Submission G044*, 14 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; *Confidential Submission G051CON*, 14 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Sydney IVF Limited, *Submission G062*, 14 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

- representatives of genetic support groups and disability advocacy groups;¹³⁴
- ethicists;¹³⁵
- health consumer representatives;¹³⁶
- lawyers (to cover privacy, discrimination and health law matters);¹³⁷
- insurance industry representatives;¹³⁸
- actuaries;¹³⁹
- employers and trade union representatives;¹⁴⁰
- science communicators;¹⁴¹

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- 134 Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002; Australian Huntington's Disease Association (NSW), *Submission G268*, 20 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; *Confidential Submission G051CON*, 14 January 2002; Sydney IVF Limited, *Submission G062*, 14 January 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002; Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002, Disability Discrimination Legal Service, *Submission G146*, 28 March 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.
- 135 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.
- 136 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Australian Huntington's Disease Association (NSW), *Submission G268*, 20 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; F Richards, *Submission G044*, 14 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Institute of Actuaries of Australia, *Submission G105*, 7 March 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.
- 137 NSW Health Department, *Submission G303*, 13 January 2003; F Richards, *Submission G044*, 14 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; *Confidential Submission G051CON*, 14 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.
- 138 Sydney IVF Limited, *Submission G246*, 19 December 2002; F Richards, *Submission G044*, 14 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Sydney IVF Limited, *Submission G062*, 14 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.
- 139 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.
- 140 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; F Richards, *Submission G044*, 14 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002.
- 141 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

- medical records staff, health informaticists, information security experts, administrators and policy makers;¹⁴²
- the current president of the HGSA;¹⁴³
- pharmaceutical companies;¹⁴⁴ and
- the Australian Biospecimen Network.¹⁴⁵

5.143 The Inquiry agrees that the HGCA will need a wide spectrum of expertise, including both expert and community-based representation.¹⁴⁶ To the list above, the Inquiry would add representation from indigenous communities, given the importance and sensitivity of many of the issues in this area to Aboriginal and Torres Strait Islander communities.

5.144 As discussed above, the Inquiry believes that a national approach to laws, practices and systems is needed to protect human genetic information most effectively. Given the fact that many of the areas under review, including health care, are shared federal-state responsibilities, HGCA membership also should contain some jurisdictional balance, perhaps involving some positions dedicated to representatives from state and territory health departments.

5.145 Another strategy would be to consult the States and Territories with respect to other appointments. For example, the Chairs of the NHMRC, AHEC and the Embryo Research Licensing Committee only may be appointed by the federal Minister for Health and Ageing after consultation with State and Territory counterparts,¹⁴⁷ as well as other relevant Ministers and organisations.

5.146 The Victorian Department of Human Services noted that:

A body such as a Human Genetics Commission of Australia (HGCA) would provide an important focus for both technical and broader policy discussions and decisions concerning genetic information and testing. In providing expertise and advice to *all* Australian governments, and providing them with the opportunity to participate in its discussions and the development of its proposals, an HGCA would be particularly important, in view of the concentration of genetic health information held in the State public sector, governed by a variety of State laws.

Models of other national bodies often include membership of two State and Territory representatives, nominated by Government (and, in some instances, representing the smaller and larger States).

142 NSW Health Department, *Submission G303*, 13 January 2003.

143 Department of Health Western Australia, *Submission G271*, 23 December 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002.

144 Eli Lilly Australia Pty Ltd, *Submission G247*, 19 December 2002.

145 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

146 See *National Health and Medical Research Council Act 1992* (Cth) s 20.

147 *Ibid* ss 21(2), 36(4). The NHMRC also contains representatives from each state and territory health authority: s 20(d). See also *Research Involving Human Embryos Act 2002* (Cth) s 16. This position was supported in relation to the HGCA by Queensland Government, *Submission G274*, 18 December 2002.

The role of such State/Territory nominees would be to provide a source of information on matters within the purview of the States and Territories to other members of the Commission, especially in relation to health and privacy legislation and policy. Such representation would be particularly relevant, in view of the Inquiry's emphasis on a national/ uniform approach to legal responses to particular issues.¹⁴⁸

5.147 The Inquiry suggests that membership of HGCA committees be on a part-time basis, in keeping with NHMRC practice: the sort of people who have the qualifications and experience to serve in this way already will have busy lives. In order to provide the drive and continuity for the organisation, however, the Inquiry suggests that the Chair/CEO position be a full-time one. The Deputy Chair position and such other Commissioners as are appointed could be full-time or part-time, depending upon workload, resources, and individual availability.

5.148 As is the case with appointments to the NHMRC,¹⁴⁹ including the new Embryo Research Licensing Committee,¹⁵⁰ appointments to the HGCA should be for a term of up to three years (as specified in each case in the instrument of appointment), with the possibility of re-appointment. In order to ensure continuity of operations, initial appointees to the HGCA should have staggered terms, so that only a third of the appointments can expire in any year. As noted above, the HGCA itself should have the power to appoint consultants and to co-opt persons to serve on ad hoc working groups and panels.¹⁵¹

Resources

5.149 The HGCA should be given sufficient budget resources to fulfil its mission. Apart from the need to provide funds for appointees, the budget should allow for a secretariat, the development of significant in-house expertise, the ability to commission research and expert consultants where the need arises, and the need to engage the public through a website, publications, public meetings and other means.¹⁵²

148 Department of Human Services Victoria - Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002.

149 *National Health and Medical Research Council Act 1992* (Cth) s 21(6).

150 *Research Involving Human Embryos Act 2002* (Cth) s 17(2).

151 See Centre for Genetics Education, *Submission G232*, 18 December 2002.

152 As a rough guide, the OGTR has an annual budget of \$8 million (but also has 'policing' responsibilities), and the proposed new Embryo Research Licensing Committee has a projected budget of \$3–6 million per annum. See Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Queensland Government, *Submission G274*, 18 December 2002.

Openness and accountability

Open, transparent and inclusive processes

5.150 In DP 66, the Inquiry proposed that ‘As a general rule, meetings of the HGCA and its committees should be open to the public’.¹⁵³ In support of this proposal, the Inquiry wrote that:

as a general rule, meetings of the HGCA and its sub-committees should be open to the public (including media representatives), in order to ensure a high degree of transparency and accountability, maintain public confidence in the integrity of its operations, and promote public engagement.¹⁵⁴

5.151 The submission from the Australian Academy of Science also emphasised the importance of openness:

It is important that discussions of matters of principle by this body should be conducted in public, so that the public is aware of the issues and the way in which differences of opinion are resolved.¹⁵⁵

5.152 Proposal 3–6 received support from a number of quarters,¹⁵⁶ and especially from genetic support groups. For example, the Genetic Support Network of Victoria wrote that:

In fact we would take it a step further and say that not only should the meetings be open to the public as a gesture, but that the public should be openly invited to meetings and encouraged to participate.¹⁵⁷

5.153 The Haemophilia Foundation Victoria submitted that:

This is an emotive issue with far reaching social and economic implications. As such, it is imperative that HGCA meetings be open to the public wherever reasonably possible. The public must be afforded the opportunity to comment upon recommendations in an informed and timely manner to ensure that the HGCA is ‘grounded’ by community concerns, issues and implications.¹⁵⁸

5.154 Similarly, the Androgen Insensitivity Syndrome Support Group Australia stressed that:

153 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 3–6.

154 *Ibid* [3.155].

155 Australian Academy of Science, *Submission G097*, 21 January 2002.

156 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Australian Medical Association, *Submission G212*, 29 November 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002.

157 Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

158 Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002. See also Australian Huntington’s Disease Association (NSW), *Submission G268*, 20 December 2002.

Ensuring that meetings of both the Technical and Ethical/Social committees are open to the public, will ensure not only transparency of the proceedings but the potential for experts that may not form part of a committee to have a say about issues being discussed. Making known in advance subjects and issues for discussion at the Commission, would ensure widest possible consultation amongst genetics support and information groups and medical/scientific experts alike.¹⁵⁹

5.155 No one argued *against* the proposition that the operations of the HGCA should be open, transparent and accountable. However, a number of submissions qualified their support for the proposal by pointing to practical concerns. For example, the Queensland Government wrote that:

It is recognised that it is important to ensure transparency and accountability and the development of public confidence in the proposed HGCA. However, open committee meetings may not always be a practical means of achieving the desired outcome and may encumber the proposed HGCA unnecessarily.¹⁶⁰

5.156 The Department of Health of Western Australia wrote that it

agrees that the HGCA and its committees should be accountable and its operations transparent for the reasons stated in [DP 66, para] 3.154. However, meetings that are open to the public, including media representatives, have the potential to hinder frank and open discussion. Public/media attendance at meetings may stifle the expression of individual opinion or comments that may otherwise be expressed in a closed meeting. That is not to say that meeting agendas, resolutions, communiqués etc should not be made available to the public in a timely fashion. The operation of the gene technology committees established under the *Gene Technology Act 2000* might provide a useful model.¹⁶¹

5.157 The Institute of Actuaries of Australia wrote that:

IAAust supports the proposal, noting particularly the inclusion of the words ‘as a general rule’. The Inquiry has recognised the sensitive nature of human genetic information, and the commercial nature of some industries where the use of genetic information may be important. It is imperative that committees of the HGCA must be allowed to meet in camera where appropriate, eg where sensitive personal information or commercial in confidence information is being discussed.¹⁶²

5.158 The Centre for Law and Genetics noted that:

We agree with this Proposal [but] suggest that the proposed HGCA could follow the procedure adopted by the Council of the NHMRC that holds parts of its meetings as closed sessions but opens up a number of topics to the public and the press.¹⁶³

5.159 The Centre for Genetics Education submitted that, while it agreed with the general thrust of the Proposal, ‘it may not always be practicable. In those cases, the minutes of the meeting should be accessible to the public’.¹⁶⁴ This approach is taken,

159 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

160 Queensland Government, *Submission G274*, 18 December 2002.

161 Department of Health Western Australia, *Submission G271*, 23 December 2002.

162 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

163 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

164 Centre for Genetics Education, *Submission G232*, 18 December 2002.

for example, by the OGTR, which publishes a public communiqué after its meetings, in which it identifies core issues and decisions of that meeting.¹⁶⁵

5.160 The Inquiry maintains its view that the culture of the new HGCA should be one that places a high value on promoting open, transparent, and inclusive processes. However, the Inquiry also recognises that, as a practical matter, it would be neither practical nor desirable for every moment of every meeting of the HGCA or its committees and working groups to be open to the public. For example, a portion of those who made submissions to this Inquiry requested that these be kept confidential, owing to the sensitive, personal information disclosed. Similarly, the HGCA will have occasion to solicit and consider personally sensitive, or perhaps commercially sensitive, information. The HGCA and its committees also will need some space to reflect and to develop ideas, draft proposals and documents before these are made available for public comment and consultation. The original proposal was not intended to inhibit such routine institutional activities and the recommendations below have been re-worded to make this clearer.

Accountability and reporting requirements

5.161 As with all federal statutory authorities, the HGCA should be accountable to the Commonwealth Parliament for its activities and its use of public funds, through an Annual Report¹⁶⁶ and through the Senate Budget Estimates Committee process.

5.162 In keeping with its primary role of providing high level policy advice on human genetics to the Australian community, the enabling legislation should also specify that the HGCA may present such other reports to Parliament as it wishes to make from time to time.¹⁶⁷ As the HGCA will be an advisory body, it is not anticipated that its policy advice would be subject to administrative merits review.

Need for effective liaison

5.163 In DP 66, the Inquiry proposed that the HGCA ‘should liaise closely with relevant government departments, authorities and entities to promote a national approach to the protection of human genetic information’.¹⁶⁸ The Centre for Law and Genetics noted that:

This is perhaps the most important Proposal in relation to the HGCA. The Inquiry demonstrated that there is no uniform national coordination of the myriad issues in human genetics. There is a mosaic of organizations, public and private ... that are involved in consideration of issues involving human genetics.¹⁶⁹

165 Office of the Gene Technology Regulator, *Consultation*, Canberra, 20 November 2002.

166 See eg *Research Involving Human Embryos Act 2002* (Cth) s 18.

167 See eg *Ibid* s 19.

168 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 3–6.

169 Centre for Law and Genetics, *Submission G255*, 21 December 2002. See also Centre for Genetics Education, *Submission G232*, 18 December 2002.

5.164 The Queensland Government also commented on the importance of this aspect, submitting that:

A national approach to the protection of human genetic information can only be achieved through close working relationships with government and non-government bodies. There is clearly potential for an overlap in roles between the proposed HGCA and other established bodies. Therefore, in order for the proposed HGCA to function effectively as an advisory body, close liaison with government and non-government entities would be essential. Close liaison with business and community stakeholders, especially health and insurance industry and consumer bodies would be essential, particularly if open public hearings are not conducted.¹⁷⁰

5.165 While noting the obvious importance of effective liaison, the submission from Senator Natasha Stott Despoja also warned against the dangers of becoming pre-occupied with consultation to the detriment of effective policy development and service delivery:

I agree that the proposed HGCA should liaise closely with other relevant governmental departments, authorities and entities to promote a national and coordinated approach to the protection of human genetic information. However, legislative requirements for wide-ranging consultation should not be so onerous that the entire process is allowed to delay the introduction of federal laws to protect human genetic information. Such a problem exists currently in the United States of America where vested interests, party politics, and other purportedly more urgent legislative matters have conspired to delay the passing of numerous genetic privacy bills through the Congress. Indeed, the gridlock that exists has meant that many hundreds of individuals have already been, and hundreds of thousands more will soon be, seriously hurt by the absence of satisfactory, all-encompassing genetic privacy legislation.¹⁷¹

5.166 The Inquiry affirms its view that it will be necessary for the HGCA to maintain close liaison with relevant government departments and authorities, to ensure that there is no duplication of efforts, nor any gaps in coverage with respect to the effective protection of human genetic information. As the length and scope of this Report amply demonstrate, human genetic information is already used across a wide array of contexts, and this is likely to increase over time. Responsibility for most of these activities will be shared across departments and authorities, and at both the federal, state and territory levels. Apart from government instrumentalities, there are many other key stakeholders—interested individuals, genetic support groups, researchers, professional associations, industry bodies, trade unions, and so on.

5.167 One of the functions of effective liaison will be to ensure that the wide variety of bodies with an interest in human genetics will be able to coordinate their efforts, rather than duplicate them.

170 Queensland Government, *Submission G274*, 18 December 2002. See also Department of Human Services South Australia, *Submission G288*, 23 December 2002.

171 N Stott Despoja, *Submission G198*, 27 November 2002; on this point, the submission refers to a position paper by the Council for Responsible Genetics: Council for Responsible Genetics, *Genetic Discrimination: A Position Paper Presented by the Council for Responsible Genetics*, <www.genewatch.org/programs/privacy/genetic-disc-position.html>, 5 March 2003.

5.168 As an indication of the types of liaison, the Inquiry suggests that there be regular communication and consultation with, among others:

- the NHMRC and its Principal Committees (including AHEC), the Embryo Research Licensing Committee, and the Gene Therapy Research Advisory Panel;¹⁷²
- the Australian Health Ministers Advisory Council and its working parties;¹⁷³
- the Commonwealth Department of Health and Ageing, and state and territory counterparts;
- the Medical Services Advisory Committee, the Pharmaceutical Benefits Advisory Committee, Public Health Partnerships, and professional associations such as the Human Genetics Society Australasia;¹⁷⁴
- the Commonwealth Department of Education, Science and Training, as well as other bodies responsible for research funding and management (such as the Australian Research Council and the NHMRC's Research Committee), as well as for the design and delivery of educational programs;¹⁷⁵
- the Office of the Gene Technology Regulator;¹⁷⁶
- the Therapeutic Goods Administration;¹⁷⁷
- the National Pathology Accreditation Advisory Council;¹⁷⁸
- the Office of the Federal Privacy Commissioner, state privacy commissioners, and those who hold similar positions in the health privacy field in the States and Territories;¹⁷⁹
- Biotechnology Australia;
- the Australian Law Reform Commission;
- the Human Rights and Equal Opportunity Commission, and state and territory counterparts; and

172 See Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

173 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

174 Queensland Government, *Submission G274*, 18 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

175 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

176 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

177 *Ibid.*

178 NSW Health Department, *Submission G303*, 13 January 2003.

179 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

- other relevant stakeholders in the public and private sectors, including genetic support organisations, professional associations, and industry bodies.¹⁸⁰

Periodic review

5.169 In common with standing practice, the Inquiry recommends that the HGCA should be subject to a basic review two years after establishment, and then a more thorough independent review after five years of operation. This review schedule should be provided for in the HGCA's constitutive legislation.¹⁸¹

5.170 Finally, although 'sunset clauses' are a feature of the enabling legislation of some new statutory authorities,¹⁸² the Inquiry does not favour using this device in the present case. Complex issues about how to deal with advances in genetic science and technology will not cease to exist in three or five or ten years—indeed, such concerns are likely to continue, if not accelerate, for some years to come. If it happens that the HGCA comes to outlive its usefulness, it will not be a difficult matter for the Parliament to pass legislation abolishing it.

Recommendation 5–1. The Commonwealth should establish a Human Genetics Commission of Australia (HGCA) under federal legislation as an independent statutory authority with sufficient resources to fulfil its mission.

Recommendation 5–2. As a general matter, the role of the HGCA should be to provide:

- on-going, high-level, technical and strategic advice to Australian governments about current and emerging issues in human genetics;
- similar high-level advice on the ethical, legal and social implications arising from these developments, including consideration of any impact on human rights, and analysis of cost-benefit issues;
- national leadership in managing the process of change in relation to human genetics, including engagement of the public on these issues;
- relevant expertise and a consultative mechanism for the development of policy statements and national guidelines in this area, where appropriate in association with other government agencies or the relevant industries and organisations;
- assistance with the development and coordination of community, school, university and professional education about human genetics;

180 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

181 See eg *Research Involving Human Embryos Act 2002* (Cth) s 47.

182 For example, the Embryo Research Licensing Committee: *Ibid* s 46.

- advice and a consultative mechanism to assist relevant bodies in identifying strategic priorities for research in human genetics; and
- a focus for the coordination and integration of various national—and perhaps regional and international—programs and initiatives.

Recommendation 5–3. The HGCA also should have specific role in:

- identifying genetic tests that have particular concerns or sensitivities attached to them, and thus may require special treatment;
- making recommendations about the suitability of specific genetic tests (and the appropriate analysis and treatment of results) for use by the insurance industry (for example for risk-rating purposes), and by employers (for example for occupational health and safety reasons);
- performing any similar function or providing expert advice on any other matters relating to human genetics, whether on its own motion or in response to a formal reference from the responsible minister or ministers; and
- monitoring the implementation of the broad strategies and specific recommendations identified in this Report.

Recommendation 5–4. The HGCA structure should involve at least two principal committees: (a) a Technical Committee, and (b) an Ethical, Legal and Social Implications Committee. However, this should not preclude the HGCA from establishing other committees or working groups (for example, on education, or community consultation) from time to time, as it sees fit.

Recommendation 5–5. Appointments to the HGCA should ensure a balanced and broad-based range of expertise, experience and perspectives relevant to the evaluation and delivery of genetic health services, and the use and protection of human genetic information and genetic samples. The appointments process should involve consultation with state and territory governments, relevant communities and other stakeholders.

Recommendation 5–6. The HGCA should operate in an open and transparent manner, to the greatest extent practicable, in order to promote public confidence and engage the wider community in uses of human genetic information.

Recommendation 5–7. The HGCA should be required to present an annual report to Parliament and also should be empowered to make such other reports to Parliament from time to time as it sees fit.

Recommendation 5–8. The HGCA should liaise closely with relevant government departments and authorities, as well as other key stakeholders, in order to promote a national approach to the protection of human genetic information.

Recommendation 5–9. The HGCA should be subject to a basic review two years after establishment, and then a more thorough, independent review after five years of operation.

Part B. Regulatory Framework

6. Ethical Considerations

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Introduction

6.1 The Terms of Reference require the Inquiry to report on whether, and to what extent, a regulatory framework is required to reflect the balance of ethical considerations relevant to the collection and uses of human genetic samples and information in Australia. The Inquiry is also directed to have regard to the range of Australian ethical opinion as to which, if any, uses and applications of human genetic information are ethically acceptable.

6.2 This chapter describes the nature of ethical considerations, the features of genetic information that raise ethical considerations and the ways that decisions involving ethical considerations can be justified. It describes a range of the justifications that are used in Australia, recognising that any regulatory framework for genetic information will need to find a balance among them. Finally, the chapter describes the ongoing and important functions that ethics can continue to perform in a regulatory framework.

What are ethical considerations?

6.3 As IP 26 noted, ethics are

an accumulation of values and principles that address questions of what is good or bad in human affairs. Ethics searches for reasons for acting or refraining from acting; for approving or not approving conduct; for believing or denying something about virtuous or vicious conduct or good or evil rules.¹

6.4 Ethical considerations can be addressed at individual and at societal levels. The way that individuals are affected by the conduct of others merits ethical consideration. The effects on a person of being informed that his father died of Huntington's disease (and that, therefore, there is a fifty percent chance that he has inherited the genetic mutation) can be personally and profoundly harmful. The risk of harm to that person becomes an essential ethical consideration in deciding what information to disclose and how to disclose it. That risk will need to be balanced against the ethical interests in respecting the autonomy of the person affected, and their choice about whether to know or not.

6.5 Revealing genetic information has important ethical implications for individuals as family members. They are vulnerable to the effects of the information on their self-perception and disclosure of information on familial relationships and sense of privacy. A grandfather's discovery that he carries the genetic mutation that impairs his grandson may change and harm his perception of himself and his relationships with his descendants. He may also be concerned about how the privacy of this information will be protected and that the information not lead to differential treatment of himself or his descendants. In these ways, individual interests are related to family and societal interests.

6.6 The way that a society governs the disclosure of such information and the extent to which its laws or other regulatory frameworks control what can be disclosed, express the way that a society balances personal risks and interests against other family, community or societal risks and interests. To prohibit disclosure of genetic information, in order to prevent the kind of harm that a person at risk of Huntington's disease might suffer, may not adequately reflect the needs of others. From balancing ethical considerations, flexible solutions may be derived that accommodate the interests of individuals and the needs of families and society.

6.7 In this way, ethical considerations reflect the kind of society in which we live or would choose to live. As DP 66 explained:

While the term 'ethics' is used in a wide variety of senses, its meaning consistently relates to an 'ethos' or 'way of life.'²

1 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, 106.

2 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, 291.

6.8 The way of life of a society or community can be reflected in the laws it makes.³ The basis for those laws can be described as the ethos of the society so that they express that society's ethics. Indeed, the answers to some of the questions posed in this Inquiry are already provided by existing laws. For example, privacy laws prohibit the collection, use or disclosure of genetic information without consent, except in limited circumstances. Similarly, anti-discrimination laws prohibit the reliance on genetic information in ways that are unfair.

6.9 It can be argued that ethics expresses the fundamental considerations that inform any societal decisions. Ethics brings together and integrates relevant interests, individual, familial, community and societal. Ethics can have an integrative function in the context of biotechnology:

Ethical judgements are not stand-alone judgements, rather they are integrative, holistic, or 'all things considered' judgements. The Canadian moral theorist Thomas Hurka put this point well in a book on the ethics of global warming:

An ethical judgement about climate policy is not just one judgement among many, to be weighed against economic, political, and other judgements in deciding how, all things considered, to act. It is itself an all-things-considered judgement, which takes account of economic and other factors. If a climate policy is right, it is simply right; if it is ethically wrong, it is wrong, period.

That is, in making an ethical judgement about global warming or biotechnology, 'ethics' is not one factor to be considered alongside other factors, like legal, scientific, or economic factors. Rather a sound ethical judgement involves an integration of all the relevant factors. Since expert judgement is relevant in the recognition and understanding of relevant factors and their interplay, combined expertise is essential. In this joint endeavour, what ethicists can contribute on the basis of the ethical theory and work in applied ethics is help in understanding the complex ways in which integrative judgements can be made, criticised and justified.⁴

6.10 The language in which ethics is expressed includes two distinct types of statements. Ethics contains statement about what is good or bad, what ought or ought not be done and the grounds for those assertions. For instance, researchers ought, ethically, to seek consent from people to use their genetic information in research because doing so respects their autonomy and freedom to choose. Or, on the other hand, researchers should be free to use coded genetic information in research without consent because that will enable more information to be used and better research to be conducted. As a result, all members of society, including those whose information is used, will benefit. These statements are often called normative statements—they are statements about how, and why, people should behave.

6.11 Ethics also contains statements about the kinds of justifications that are used in normative statements. For instance, respecting a person's autonomy is a principle of 'principlist ethics'. Acting to achieve the best outcome is, on the other hand, a

3 K Liddell, *Submission G141*, 23 March 2002.

4 M McDonald, *Biotechnology, Ethics and Government: A Synthesis*, Canadian Biotechnology Advisory Committee, <www.cbac-cccb.ca/>, 7 February 2003, 6.

justification based on consequences and not on principles. These justifications are referred to as ‘consequentialist ethics’. Much of the content of this chapter uses normative statements. The chapter describes the range of ethical considerations that are likely to be drawn on in making and justifying decisions about genetic information. The regulatory responses recommended in this Report to protect genetic information reflect a balance among these considerations.

Genetic information and ethics

6.12 As the Inquiry has progressed, the links suggested in IP 26 and DP 66 between ethics and genetic information have been confirmed and extended. In IP 26, it was suggested that these included the following aspects of genetic information:

- how it is obtained;
- what it reveals;
- about whom that information is revealed;
- the degrees of certainty and uncertainty of that information;
- who among those affected should know, and who should not know, some or all of that information;
- which third parties (such as employers, insurers or others) should know or not know;
- what constraints, if any, should limit what people can do with genetic information about others;
- what restrictions, if any, should apply to people because of the genetic information about them;
- what effect, if any, does self-awareness of genetic knowledge have on people; and
- what effect, if any, will widespread knowledge and use of genetic information have on the broader society?⁵

6.13 This list could now be reduced to a simpler but more inclusive one. Who should be permitted to collect, use or disclose genetic information about a person or persons?

- from whom and to whom?
- for what purposes?

⁵ Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, 114.

- with whose consent?
- in what manner?
- on what conditions?

6.14 These questions are, generally speaking, relevant whether the information is being collected in the form of family medical histories or genetic tests, and whether the purposes are related to research, health care, insurance, employment, parentage testing or law enforcement. They are questions of ethics because, as outlined above, the answers we would give are likely to express our views about what collection and use of genetic information would be acceptable to individuals and to Australian communities.

6.15 The characteristics of genetic information that give rise to difficulty in answering these questions were identified in Chapter 3. Genetic information is at once readily available and virtually ubiquitous. It is unique to an individual yet enables familial inferences to be made. From individuals, genetic inferences can be made about family members and from family histories, inferences can be made about individuals. Genetic information is often predictive, concerning probabilities rather than certainties.

6.16 These special characteristics have important consequences for ethics because they challenge established methods of structuring ethical justifications and compel re-examination of those methods. DP 66 explained this in the following way:

It is because genetic information has dramatic potential to change the 'way of life' of persons affected by it, and arguably even to alter our conception of the human 'person', that it raises ethical concerns. These concerns extend beyond the kind of moral dilemmas that may quickly be resolved by reference to pre-existing values, for several reasons.

First, some situations created by advances in genetic science and technology are unprecedented, except perhaps in the works of science fiction writers. As a community, we have yet to determine settled moral values and rules of conduct in relation to the novel possibilities opened up by the rapid development of this field of science. Consequently, the questions that arise in the course of this development call not merely for moral reflection, but for reflection upon morals. ...

Second, human genetic information does not operate within discrete and stable parameters. It tends to spill out from the scientific and medical domains to affect a growing range of human activities and interests. It is not sufficient that this type of information be evaluated solely in terms of its scientific validity. Rather, it must be judged with respect to its impact on our shared 'way of life'. Ethical inquiry aims to allow, and indeed foster, this kind of evaluation. It is centrally concerned with the kind of procedures or discussions that allow all relevant sources of information and viewpoints on a disputed matter to be taken into account in coming to a decision.

In this sense ethics is a rational and impartial activity, concerned to inform and justify decisions and actions. However, this does not imply that an ethical judgment will be a conclusive one. On the contrary, ethics is necessarily an ongoing activity, since our 'way of life' is continually developing. Nor does this emphasis on reason imply that ethical procedures seek to exclude or devalue emotion, but rather that they aim to

place emotional responses (which are often based on strong moral commitments) in a framework in which they can be rationally assessed and balanced against all other relevant perspectives on an issue.⁶

Balancing ethical considerations

6.17 The Inquiry is therefore centrally engaged in ethical issues. The challenge is to articulate, in an inclusive way, the reasons and arguments that could be used to resolve dilemmas and to provide justifications for answers to the questions raised by the Inquiry. The challenge is also to identify values or principles that reflect our way of life. What makes this task problematic is the recognition that our way of life will evolve over time, and may be changed by the use of genetic information itself. Faced with this situation, IP 26 and DP 66 referred to some established sources of values and principles.

6.18 These values and principles are revisited in this chapter and are referred to, where relevant, throughout this Report. The Inquiry's responsibility is to make recommendations that reflect an appropriate balance of ethical considerations. To justify regulatory reform, a balance needs to be reached among the ethical considerations described in this chapter.

6.19 The Inquiry recognises that reaching a balance will often be difficult. Indeed, the concept of a balance suggests that considerations can be weighed against one another, or together, and that one consideration will always have the same weight relative to others. Even if this is so, contexts for decisions will attract different clusters of ethical considerations, so that different relative balancing will be required. Some participants may always give a greater weight to one consideration, for instance, respect for autonomy, than do other participants. Accordingly, when speaking of a balance of ethical considerations, the Inquiry recognises that it is a balance that needs to be considered for each regulatory issue.

6.20 In discussing the ethical contribution to the development of biotechnology policy in Canada, Susan Sherwin noted that a strategy such as that outlined in this chapter will not always offer a definitive solution:

Thus, when approaching complex policy matters, we should actively seek out moral perspectives that help to identify and explore as many moral dimensions of the problem as possible. This requires us to pursue deliberative strategies that will promote sensitivity to the multiple moral considerations that are relevant to the issues before us. Nevertheless, the identification of morally relevant factors does not always produce a unique moral solution. Hence, even if we were able to identify all the morally relevant aspects of a situation, we might still not know how to resolve the specific moral dilemma before us. Although these substantive moral tools (theories) will exclude certain options, they sometimes fail to select a single best solution. In such cases, we must rely on effective strategies that can produce an ethically

⁶ Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, 291–292.

acceptable decision. Only an ethically adequate procedure can resolve these various types of ambiguity.⁷

Aristotle: prudence and uncertainty

6.21 Among the many philosophical and theological concepts and theories that have contributed to the contemporary understanding of ethics, the Ancient Greek concept of prudence stands out as a principle of particular relevance to the area of genetics. According to Aristotle, the prudent person is one who engages in well-conducted deliberation which is timely, measured and takes into account the particular problems and circumstances of the case in order to foresee ‘even the unforeseeable’. The fact that genetic information gives rise to unforeseen, and even unforeseeable, situations involving both benefit and risk, makes the concept of prudence and the standards of deliberation it entails particularly applicable in this area.

6.22 The ‘principle of caution’ has emerged as a modern version of this concept. Hans Jonas, devising an ‘ethic of responsibility’, argues that the capacity of modern technology to transform our lives has altered the primary task of ethics.⁸ Traditional (Platonic) ethics is based on an understanding of the human condition as given once and for all, so that the good to be attained is readily determinable and eternally valid. Modern technological developments challenge this vision, shifting the focus of ethical inquiry from the perspective of eternity to that of temporality ‘in its ever-new, always unprecedented productions, which no knowledge of essence can predict’.⁹

6.23 Given this, the epistemological confidence that underpins traditional systems of morality gives way to a potent sense of uncertainty about the future, as the rate of technological change increases. Jonas makes this uncertainty a central tool in his approach to ethics and argues that the question of responsibility for the changeable and perishable, rather than concern for fidelity to eternally fixed norms, ought to be the dominant concern of modern ethics.

6.24 Iain Robertson, in a submission to the Inquiry,¹⁰ echoed these concerns in advancing ‘uncertainty’ and ‘ambiguity’ as relevant ethical principles. He identified qualitative and quantitative uncertainty—the former illustrated by the uncertainty as to whether accounts of personal illness were true and the latter by the uncertainty as to the causal effects of illness. He also identified general and specific uncertainty—the former illustrated by the uncertainty of knowledge of health across a population and the latter by the uncertainty as to whether an individual’s ill health corresponds to what is known at a population level. He argued that the recognition of these uncertainties was important because certainty of behaviour, for instance by a health professional, can inspire confidence but may not be a reliable guide to the efficacy of treatment.

7 S Sherwin, *Towards an Adequate Ethical Framework for Setting Biotechnology Policy* (2001), Canadian Biotechnology Advisory Committee, Ottawa, 11.

8 H Jonas, *The Imperative of Responsibility: In Search of an Ethics for the Technological Age* (1984) University of Chicago Press, Chicago.

9 *Ibid.*, 126.

10 I Robertson, *Submission G209*, 29 November 2002.

Principlist ethics

6.25 The school of principlist ethics has dominated the field of health research ethics since it first gained momentum in the late 1960s and was given formal articulation in the Belmont Report.¹¹ It is characterised by an assumption that scientific progress is essential for the good of humanity, coupled with a concern to protect individual and group rights that may be endangered in the course of scientific research. It seeks to establish principles that must be respected in carrying out this work, building upon traditional principles of medical practice and ethics. These include the principles set out in the ‘Georgetown mantra’, which are respect for autonomy, beneficence, non-maleficence, and justice.¹² These and related principles are already enforced in legal and regulatory frameworks dealing with human rights and research protocols. Their application to the field of genetics is seen as an extension of existing concepts to cover a field of research and medical practice in which, although the science is new, the ethical issues to which it gives rise are not. For instance, disclosing a genetic diagnosis requires attention to ethical considerations of beneficence and non-maleficence in the same way as disclosure of a non-genetic diagnosis. What is different is that the diagnosis will often have important meaning for the patient’s relatives, which the consideration of harm and benefit must include.

6.26 For example, informing family members of the diagnosis of illness in their father without his consent would, in traditional principlist ethics, be unethical because it would offend the principle of respect for (the father’s) autonomy. Principlist ethics might argue that the same principle should apply to the disclosure of a genetic diagnosis. However, what may be different is that a genetic diagnosis may mean that siblings and children are at risk of a familial cancer, perhaps one that is treatable if identified early and monitored. Reliance on the principle of respect for autonomy alone would ignore the ethical interest of siblings and children in having information to make decisions about their own health.

6.27 The use of genetic information in insurance and employment reflects an application of principlist ethics. In insurance, respecting individuals’ decisions not to undertake genetic testing (their autonomy) so as to obtain insurance on the most beneficial terms can compete with the injustice to other insureds who may over time have to bear a higher premium as a result. The perceived risk of being denied insurance may act as a disincentive to testing, possibly precluding early diagnosis of treatable conditions. This could be said to offend the principle of beneficence. Respecting the autonomy of employees, by allowing them to work where they want, can offend the principle of non-maleficence by exposing them, and sometimes other workers, to avoidable risks of harm. Requiring genetic tests in insurance or employment may pit individual autonomy against protection of, and justice to, others.

11 National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research, *The Belmont Report: Ethical Principles and Guidelines for the Protection of Human Subjects of Research* (1978), US Department of Health, Education and Welfare, Washington.

12 The ‘Georgetown mantra’ was formulated by James Childress and Thomas Beauchamp in 1979. See now T Beauchamp and J Childress, *Principles of Biomedical Ethics* (5th ed, 1999) Oxford University Press, New York.

6.28 Principlist ethical guidance was endorsed in several early submissions to the Inquiry. The Australian Medical Association (AMA) emphasised the importance of the principles of the ‘Georgetown mantra’ in ensuring that patients’ interests are protected.¹³ The Australian Academy of Science also favoured reliance on established principles of health care and research ethics, arguing that genetic information should not be regarded as inherently different from any other form of biological or medical information. The Academy expressed concern that excessive restrictions on the use of clinical data and samples, because of their genetic information, might hinder the conduct of clinical pathology and epidemiology, to the detriment of the future of health care in our community and the international standing of Australian research in these areas.¹⁴ As these submissions recognise, the fact that principlist ethics already informs the regulation of health care and research means that it needs to be included among ethical considerations arising from the use of genetic information.

6.29 However, other submissions questioned the suitability or adequacy of this source of ethics. ACROD Limited doubted that these principles were sufficiently robust and claimed that they need to be supplemented by a more specifically focused code of ethics to address risks of genetic discrimination.¹⁵ Cormac Nagle of the Mercy Hospital for Women preferred a broader approach than one confined to these principles, recognising that, in the specific arena of research ethics, varied membership of review committees would have this effect.¹⁶ Iain Robertson argued

1) that health ethics are highly complex; 2) that the simple ‘algebraic’ approach suggested by the ‘Georgetown Mantra’ is inappropriate, because rational balancing of the different principles cannot possibly cope with the interactions and relative valuing of the principles that would be required; and 3) that an empirical approach to the values placed on each principle by the different real people and groups in our community is required.¹⁷

Consequentialist ethics

6.30 Principlist ethics measures the goodness or badness of conduct by whether or not it accords with a principle. This kind of justification can be, and often is, contrasted with those that determine the goodness or badness of conduct by reference to its consequences. Commonly called consequentialist or utilitarian ethics, this approach to justification, and that of principlist ethics, are two commonly used moral arguments. They are frequently used to support opposing views on conduct. The example of using coded genetic information in research without individual consent can serve as a simple illustration. Principlist ethics might argue that such an act is unethical because it does not respect participants’ autonomy. Consequentialist ethics might argue that the conduct is ethically sound because it will maximise the benefit for society.

13 Australian Medical Association, *Submission G091*, 29 January 2002.

14 Australian Academy of Science, *Submission G097*, 21 January 2002.

15 ACROD Limited, *Submission G239*, 19 December 2002.

16 C Nagle, *Submission G196*, 26 November 2002.

17 I Robertson, *Submission G209*, 29 November 2002.

6.31 Consequentialist ethics is frequently used to justify political and social judgments because of the impact on communities and society. Principlist ethics has long been associated with decisions affecting individuals, as in professional relationships, because of the close and historical alignment of ethical principles and professional duties to clients or patients.

Professional ethics

6.32 The social identification of health professionals, and the grant to them by society of the right to treat individuals, is accompanied by their acceptance of professional responsibilities. The health professions have developed enduring ideas of good professional conduct that are usefully referred to as professional ethics.

6.33 The need for professional ethics to focus on the relationship between individual professionals and their patients or clients lies in the typical imbalance of knowledge, and therefore power, between the two. Professional ethical obligations recognise the risk that professionals may misuse their power. Typical of these obligations are those to maintain the confidentiality of information about patients, to give priority to patients' interests over their own, and to avoid conflicts between personal interests and professional duties.

6.34 The development of health care ethics and bioethics in the past three decades has drawn heavily on professional ethics. The development of codes of professional practice has been through the formulation of simply stated principles. As a result, most health professionals are familiar with principlist ethics, and accept them. This history has another important feature—self-regulation. One submission argued that, in relation to genetic information, ethical obligations were too important to be left to professionals to formulate.¹⁸ On the other hand, the AMA argued that the knowledge and responsibility of health professionals gave them an essential role where citizens were asked to contribute genetic information.¹⁹

Critical ethics

6.35 Critical ethics questions the assumption that scientific progress is necessarily for the good of humanity. It criticises principlist bioethics for remaining silent on fundamental issues raised by the progress of health science (including genetics), arguing that bioethics frequently does little more than legitimise the activities of laboratories and governments. Critical ethics exposes the strong identification between principlist ethics and professional ethics. In place of the traditionally concern with patients' welfare, it seeks recognition of a wider set of responsibilities than those that professionals traditionally accept, and a wider set of interests than those of clients and patients.

18 ACROD Limited, *Submission G239*, 19 December 2002.

19 Australian Medical Association, *Submission G212*, 29 November 2002.

6.36 In relation to human genetic information, critical ethics highlights two key areas of concern. The first relates to the fact that genetic research depends on the medical-industrial complex and involves significant commercial interests. Critical ethics points to the risk that such interests may compromise compliance with ethical principles and values that are significant for the retention of an Australian ethos and society. For example, existing socio-economic inequalities may be exacerbated, to the detriment of society, if access to new health interventions based on uses of genetic information are determined solely by market forces. These concerns were voiced in numerous submissions to the Inquiry.²⁰

6.37 The second concern of critical ethics relates to the potential for abuses flowing from the creation of a human standard based on genetic discoveries. Here, critical ethics recognises fears about eugenics and other forms of genetics-based discrimination. In submissions to the Inquiry, the concern that genetic testing may lead to eugenic practices of selection, particularly in relation to sex and sexual orientation, was articulated.²¹ Critical ethics questions the motivations and assumptions that can be concealed and used to contain and limit more traditional ethical voices. Critical ethics is important when considering the design of regulatory frameworks so as to ensure transparency and openness in regulatory processes.

Ethics of discussion

6.38 Procedural ethics or the ethics of discussion attempts to integrate principlist and critical concerns. It recognises the pluralism of moral positions and emphasises the consequent need to ensure that moral discussion and debate is effective. It takes the view that the role of ethicists is not to establish standards to control biomedical developments but to promote discussion of the meaning and implications of these developments for communal life, and to identify (rather than prescribe) the moral questions that emerge from this debate.

6.39 An ethics of discussion is advocated as most appropriate to the field of bioethics to avoid the reduction of ethics to mere application of rules, the replication of hierarchical structures, and the focusing on technical issues, which may exclude lay participants. A shift away from a focus on ethical 'rules' toward an emphasis on discussion and critical reflection is said to be required, not only in the field of bioethics, but more generally in the contemporary approach to ethics:

20 Ms Kathy Liddell flagged the general question of how the benefits and costs of genetic technology should be shared: K Liddell, *Submission G141*, 23 March 2002. UnitingCare NSW & ACT submitted that the professional codes of ethics currently used to regulate ethical decision making relating to genetics are ill-adapted to the current context of increasing corporate involvement in health care and the distribution of genetic goods and services: UnitingCare NSW & ACT, *Submission G052*, 14 January 2002. Others suggested that it is naïve to assume that ethical self-regulation will be effective where significant commercial interests are at stake: *Confidential Submission G074BCON*, 13 January 2002.

21 C Somers, *Submission G021*, 14 November 2001. Others warned of the more general danger of 'genetic essentialism', or the reduction of personhood to genetic identity: I Barns, *Submission G056*, 8 January 2002; Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

Increasingly, the ethics of research and the ethics by which we live our lives will depend on negotiation. Stripped of the certainties of the past, we have to take responsibility for reconstructing the world and finding perspectives we can live with. This is a communal and political activity, in the broadest sense.²²

6.40 Some submissions to the Inquiry expressed support for a broader and more facilitative approach to ethics.²³ Nick Saunders and Paul Komesaroff identified the essential function of an ethics of discussion:

Ethical decision making involves communication and negotiation between individuals and groups in the community around issues of values. As is widely recognised, it is possible for individuals of integrity to hold widely different, even contradictory, views in relation to ethical questions. In a given situation, there is often no unique single, valid ethical decision or action. What makes a decision ethical is therefore not its substantive content but the process that generated it – namely the quality of the dialogues and reflection in which the protagonists engaged.²⁴

6.41 However, the important role for ethics in assisting society to reach decisions, and not merely ensure the quality of discussion, was also emphasised:

Discussion is most important, but if it does not come to a conclusion or a decision it remains an academic approach and unhelpful to both the individual case and society. Discussion ethics should inform members of ethics committees and lead them to make better decisions.²⁵

6.42 The Inquiry identified several areas where there are strong differences of opinion, not the least of which is those relating to parentage testing. In such contexts, attending to the ethics of discussion can assist in eliciting the underlying assumptions that contesting parties bring to the debate and identify agreements and differences more clearly.

6.43 An ethics of discussion sets standards for such a process. Those standards may not lead to a reconciliation of differing values or value based positions but, it is argued, will clarify in respectful, reasoned and effective ways, the differences that parties to the debate choose to maintain.

6.44 New discoveries are likely to emerge which will present new challenges to the regulatory regimes recommended in this Report. For this reason, among others, the Report recommends the establishment of the Human Genetics Commission of Australia (HGCA) (see Chapter 5). As these new issues emerge, recognition of and attention to an ethics of discussion will be important in identifying differing responses from existing ethical positions and articulating elements of an emerging community ethos that responds to new discoveries.

22 P McNeill, 'International Trends in Research Regulation: Science as Negotiation' in, *Research Ethics* (1999) Elsevier Science, Oxford, 243, 243, 263.

23 ACROD Limited, *Submission G239*, 19 December 2002; C Nagle, *Submission G196*, 26 November 2002; I Robertson, *Submission G209*, 29 November 2002.

24 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

25 C Nagle, *Submission G196*, 26 November 2002.

Civic ethics

6.45 While principlist and professional ethics focus on the ethics of decisions made by or about individuals, critical ethics and the ethics of discussion recognise the importance of considering community or civic perspectives in ethical matters. The use of genetic information has the potential to affect the interests of whole communities and so it is important to identify the fact that there will be community levels of ethical considerations. Accordingly, there is value in separately identifying a civic ethics.

6.46 In their submission, Saunders and Komesaroff pointed to the importance of diverse and extensive community input, channelled into responsive and democratic decision making processes.²⁶ They also emphasised the need for informed public debate and consultation on genetic issues. Community interests were also highlighted by UnitingCare NSW & ACT, which emphasised the need to consult groups who are directly affected by genetic-related illness or disability to ensure that ethical principles are applied in a way that meets the needs of vulnerable individuals and families.²⁷

6.47 The challenge to civic ethics is not only to consider the needs and perspectives of these people, but to provide them with appropriate services, such as counselling, to enable them to exercise in an informed way the responsibility they have to use genetic information.²⁸ A similar mutuality between community and individuals was recognised by Iain Robertson who argued that:

The ethical principle of mutuality recognises that the principle of individual autonomy is optimised when the strength of civil society is maximised, and that care should be taken when promoting the principle of autonomy to avoid limiting that autonomy by ill-considered constraints on public health action. At the same time, public health action is best advanced when individual autonomy is respected.²⁹

6.48 The difficult issues of consent for genetic research using human tissue illustrates an emerging tension between an ethical view that gives primacy to individual choice and an ethical view that gives equal or higher, priority to community value. One submission to the Inquiry identified the high value to the community, often difficult to specify in advance, of using tissue from existing collections for population-level genetic research.³⁰ There was concern that the opportunity to realise the value of this research could be denied by a traditional ethical insistence on fully detailed individual consent by every tissue donor to every use.

Narrative ethics

6.49 The need to apply ethical principles in ways that meet the needs of vulnerable families and individuals may be described by reference to yet another style in bioethics. Narrative ethics—the dependence on first hand accounts of those facing health care decisions as a source of reasons and justifications for those decisions—has

26 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

27 UnitingCare NSW & ACT, *Submission G052*, 14 January 2002.

28 Ibid.

29 I Robertson, *Submission G209*, 29 November 2002.

30 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

a well established place in contemporary bioethics,³¹ and was recognised in submissions to the Inquiry.³²

6.50 The dilemma surrounding the disclosure of a diagnosis to genetic relatives is one of the contexts in which narrative ethics is likely to shed more light than principlist or professional ethics. Accounts of how families have dealt with these situations—with resulting benefit or harm—will be a valuable source of ethical guidance for families that face similar challenges.

The international context

6.51 There is an international perspective to the relevant ethical considerations. While the Terms of Reference of this Inquiry refer only to consideration of ‘the range of Australian ethical opinion’, the Inquiry is also directed to have regard to ‘the global dimensions of issues relating to research, regulation and the protection of interests’. These include ethical issues.

6.52 Several submissions emphasised the relevance of existing international standards. Dr Nicholas Tonti-Filippini pointed to the importance of the UNESCO *Universal Declaration on the Human Genome and Human Rights*³³ in providing a framework for ethical practice within Australia.³⁴ The National Council of Jewish Women of Australia urged that the bioethics body that it recommended be established maintain constant contact with the UNESCO International Bioethics Committee.³⁵ The Australian Academy of Science stressed the need to remain sensitive to the international level of ethical standards and deliberations, in order to maintain and promote the profile of Australian science overseas,³⁶ while UnitingCare NSW & ACT identified several relevant provisions in international instruments relating to human rights.

6.53 In determining the balance of ethical considerations on issues relevant to the protection of genetic information, the status of these international standards deserves continued attention.

An ethics network

6.54 The Inquiry recognises the variety of relevant considerations in ethics and especially bioethics. Many of these are of relatively recent origin. The word ‘bioethics’ was coined only thirty years ago and the systematic application of human genetic knowledge is even more recent. However, traditions of medical ethics, to which

31 A Frank, *The Wounded Storyteller* (1995) University of Chicago Press, Chicago; T Chambers, *The Fiction of Bioethics* (1999) Routledge, New York.

32 I Barns, *Submission G056*, 8 January 2002. See also A Frank, *The Wounded Storyteller* (1995) University of Chicago Press, Chicago; T Chambers, *The Fiction of Bioethics* (1999) Routledge, New York.

33 *Universal Declaration on the Human Genome and Human Rights*, UNESCO, <www.unesco.org/ibc/en/genome/projet/>, 19 February 2003.

34 N Tonti-Filippini, *Submission G014*, 16 November 2001.

35 National Council of Jewish Women of Australia, *Submission G008*, 21 May 2001.

36 Australian Academy of Science, *Submission G097*, 21 January 2002.

modern bioethics owes much, have a longer history. The promises and risks of the use of genetic information in the future are imminent and to an important extent unforeseeable.

6.55 In this context, several submissions recognised the wisdom of not locating the source of ethical guidance, whether to individual citizens, professionals, governments or communities, in any one person or expert group.³⁷ Rather, the need for a functional, open and responsive network of citizens, experts, professionals and public agencies with interests in how and why genetic information is collected, stored, used and disclosed will be needed. Such a network would need to be open to the range of ethical considerations that have been referred to, and relied on, in this Report. The HGCA structure would be an important part of this network, if not its hub.

Education in ethics

6.56 Submissions to the Inquiry emphasised the need for educational measures to ensure that decisions relating to genetic information are informed by ethical dimensions.³⁸ At least two types of education are seen to be needed: specialised training and advice for those with public or professional responsibilities in relation to genetic information; and public education to inform and equip citizens for engagement in ethical discussion, debate and decision making about their genetic information.

6.57 Specific educational programs will be required to equip those handling genetic information in a professional capacity to keep pace with the ethical significance of developments in the area of genetics. The Australian Academy of Science recommended that professional training in ethics should be included in all medical science and allied health training programs at undergraduate and postgraduate level.³⁹ The AMA made a similar call for training in ethical issues for research workers and members of ethics committees.⁴⁰

6.58 A key group who requires specialised education in ethics is the individuals and families who are directly affected by genetically-related disease. Their interest and experience in the area also mean that their perspectives and insights into the ethical dilemmas posed by genetic information should be given particular weight in official and public discussions or deliberations on this topic, in a specific application of narrative ethics.⁴¹

37 ACROD Limited, *Submission G239*, 19 December 2002; C Nagle, *Submission G196*, 26 November 2002; I Robertson, *Submission G209*, 29 November 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

38 C Nagle, *Submission G196*, 26 November 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Australian Biospecimen Network, *Submission G238*, 19 December 2002.

39 Australian Academy of Science, *Submission G097*, 21 January 2002.

40 Australian Medical Association, *Submission G091*, 29 January 2002.

41 UnitingCare NSW & ACT, *Submission G052*, 14 January 2002.

6.59 Many submissions recognised the role of public perceptions, especially those that are uninformed, in determining the social significance and effects of genetic information.⁴² They identified the need to establish and maintain a healthy culture of discussion and debate, involving effective exchange between all interested parties on ethical issues relating to genetic information, a role that the Ethical, Legal and Social Implications Committee of the HGCA could play.

Ethics in a regulatory framework

6.60 Identifying the function of ethics in a regulatory framework can raise some profound issues about the relationship between ethics, governance, politics and law. Saunders and Komesaroff, noted that ‘a major issue that requires examination at the philosophical level is that of the relationship between law and ethics’ and suggested that:

the law should focus on the settings in which individuals engage in ethical decision making and seek to ensure that it is open and free from coercion and that adequate information is provided to allow individuals to make their own decisions after full and careful reflection.⁴³

6.61 Such a facilitative role is reflected in recommendations in this Report concerning the HGCA. Given this facilitation, the functions of ethics in a regulatory framework relate to individual decision making, justification of policy and regulatory choices and ongoing assessment of effectiveness and acceptability.

6.62 The necessity for individual decision making in a regulatory framework draws attention to an essential feature of ethical obligation. In its submission, the Department of Health of Western Australia noted, in relation to measures to govern the management of genetic databases, that:

.... the level of protection provided by such technical matters may still be compromised by failure in the integrity of the researchers and this is an important matter to be addressed by HRECs, institutions and the researchers themselves.⁴⁴

6.63 The integrity of researchers, and indeed of any decision makers in a regulatory environment, should be assessed by reference to the ethical values and principles on which that framework is based. The adherence to those values and principles is a matter of personal conviction not enforcement. This central feature of ethical obligation was described in IP 26:

It is intrinsic to the nature of ethical obligation that it be felt and followed because of an individual's commitment to it, whatever the source of that commitment: whether to a principle, because of a virtue or a felt obligation to a community. One motivation for behaviour that is not usually regarded as significant is the threat of enforcement or

42 ACROD Limited, *Submission G239*, 19 December 2002; C Nagle, *Submission G196*, 26 November 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

43 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

44 Department of Health Western Australia, *Submission G271*, 23 December 2002.

regulation. To ascribe the efficacy of an ethical code to the efficiency of its enforcement mechanism or to its regulatory force might be said to confuse the nature of ethical obligation with that of legal obligation. Evidence of the efficacy of ethics, or health care ethics, is perhaps better found in the conforming behaviour of the people taken to be subject to the obligations.⁴⁵

6.64 Ethics as the personal commitment to, and implementation of, values and principles operates to maintain the coherence and consistency of an ethically founded regulatory framework. Such a commitment is based on awareness and understanding of ethical principles derived from education and experience. It is important that ethics continues to promote, guide and justify good conduct and decisions by citizens, professionals and regulators in the handling of genetic information.

6.65 At a broader level than that of individual obligation, an essential role of ethics in a regulatory framework is that of justification.

In public policy debates, governments will make choices—even the decision to postpone and put off making a definitive choice is itself a choice. Making choices raises a central ethical issue: whether and how choices can be justified? That is, when people or institutions are faced with choices they want to make reasonable choices—choices supported by good reasons. But there are many different kinds of reasons for making particular choices ... The decision could be made on the basis of current policy, interest group lobbying, public opinion polls, administrative precedent, the personal feelings of senior bureaucrats or politicians, etc. But it is to be hoped that the decision will be made on the basis of good moral reasons which as indicated above are motivating reasons that are impartial, promote human well-being, non-arbitrary and overriding considerations of self-interest. But to justify the choice of one of these requires moral argumentation. That is, it is fair to ask whether a particular choice can be justified from the moral point of view. The moral point of view should be one that all the affected parties can reasonably endorse. It should not just reflect the interests of some of the parties, but all of them. That is, the choice should be justifiable interpersonally.⁴⁶

6.66 In providing reasons for such public policy choices, ethics can express the ethos of a society. In time, that ethos is frequently, though not invariably, expressed in the laws that a society chooses to make, or the regulatory frameworks that it chooses to devise and implement. However, as McDonald recognises, ethics tends to be expressed in widely shared principles so perfect justification may not always be demonstrated.

The ethical perspective urged here is to treat the use of ethics in public policy as a way of judiciously balancing or weighing relevant considerations—considerations usually identified by principles in common use. The objective, of course, is to make good ‘all things considered’ moral judgements that can be used to ground and formulate public policy. ...

45 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, 121.

46 M McDonald, *Biotechnology, Ethics and Government: A Synthesis*, Canadian Biotechnology Advisory Committee, <www.cbac-cccb.ca/>, 7 February 2003, 10.

In the sometimes messy and often times complex world of public policy-making the aim is not ideal or perfect justification but something more moderate and achievable—as it were ‘good enough’ policy-making, that is decisions reasonably supported by common moral principles (including principles of good governance).

While appeal in ethical reasoning is made to principles in common use, there must be openness to the idea that at least some commonly accepted principles are improperly used, restrictively applied, or otherwise inadequate. Otherwise, there would be no possibility of moral change or moral progress.⁴⁷

6.67 The function of ethics in policy making can be illustrated by the Report’s recommendation in Chapter 32 that genetic information from employees be collected and used to protect third party safety only where the danger cannot otherwise be avoided and the employee’s condition poses a risk of serious danger to the health and safety of third parties. The formulation of this recommendation reflects an attempt to strike a balance between, on the one hand, ethical protection of individual freedom and privacy and, on the other, ethical recognition of the need to protect innocent third parties from harm.

6.68 This recommendation may also be used to illustrate the role of ethics as a touchstone against which the continuing acceptability of regulatory frameworks may be assessed. As decision makers and regulators gain experience in the practical application of the recommended balancing test between individual freedom and third party safety, the effectiveness and acceptability of this formulation can be reassessed against the ethical foundations on which it was established.

6.69 In these ways, ethics can inform and justify the establishment of a regulatory framework, promote, guide and justify individual decisions in that framework and inform the continuing assessment of the framework’s effectiveness and acceptability.

47 Ibid, 10.

7. Information and Health Privacy Law

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Introduction

7.1 The Terms of Reference require the Inquiry to report on whether, and to what extent, a regulatory framework is required to protect the privacy of human genetic samples and information. Privacy is a concept capable of many meanings.

Privacy has been variously described as the right to be let alone, the right to personal space or autonomy, the right of people to exercise control over their personal information or the degree of interference with their personal life, a popular reaction to the spread of new technologies or, more recently, simply fair information practices.¹

7.2 There are increasingly few zones of privacy to be found in modern life. High density urbanisation means people have less space of their own. Surveillance cameras in public areas, shops and even in the workplace are increasingly common for security reasons.² Computers can track every movement and transaction, and facilitate cross-matching of information from disparate databases, and can directly monitor the activity of users (through 'cookies' and other means). Popular culture, especially as portrayed on television and on the Internet, is full of so-called 'reality programming' based upon intensive video surveillance of the sort that was first suggested—with horror, rather

1 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

2 See generally New South Wales Law Reform Commission, *Surveillance*, Interim Report 98 (2001).

than fascination—in George Orwell's classic novel *1984*. The heightened desire for border security has also encouraged the development of biometric technologies to track individuals as they move from one country to another.³

7.3 There is a very low rate of genetic variation among humans—99.9% of the human genome is identical. When we talk about ‘genetic privacy’ we are referring to only that very tiny part of the human genome that is unique to each individual, the single nucleotide polymorphisms that occur once in roughly 1,300 bases in each individual’s genetic code. In what has arguably become—with little public resistance in some areas, and strong encouragement in others—a ‘surveillance society’, there is genuine community concern about protecting the privacy of genetic information.

7.4 Importantly for the purposes of this Inquiry, the privacy of human genetic information is one aspect of the broader concept of information privacy. Information privacy can be defined as the right of individuals to control the collection, use and disclosure of information relating to them (personal information).

7.5 The High Court has confirmed that there is no enforceable general right to privacy in Australian common law and, in particular, there is no tort of invasion of privacy.⁴ With certain exceptions, such as where duties of confidentiality are breached, enforcement of rights to privacy must be based on statute, including information and health privacy legislation.

Information and health privacy legislation

7.6 The Inquiry has sought to assess the adequacy of existing privacy legislation as a framework for protecting the privacy of genetic samples and information and to make recommendations to better protect genetic privacy.

7.7 A key issue identified by the Inquiry is whether information and health privacy legislation should cover genetic samples as well as the genetic information derived from them. This issue is examined separately in Chapter 8. As discussed in more detail in that chapter, the Inquiry has concluded that the *Privacy Act 1988* (Cth) (*Privacy Act*) does not cover genetic samples, even where they are identifiable to an individual, for example, where they have a name or other identifier attached. The Inquiry is of the view that the Act should be extended to cover genetic samples by expanding the definitions of ‘personal information’ and ‘health information’⁵ to include bodily samples.⁶

7.8 This chapter briefly summarises the existing legislative framework for the protection of information and health privacy based on the *Privacy Act* and similar state and territory legislation and its application to the privacy of genetic information.

3 K Dearne, ‘Immature Biometric ID Systems Flawed’, *The Australian*, 4 March 2003, 28.

4 *Australian Broadcasting Corp v Lenah Game Meats Pty Ltd* (2001) 185 ALR 1.

5 *Privacy Act 1988* (Cth) s 6.

6 There are other mechanisms through which privacy protection of genetic samples could be pursued. These include recognising new property rights in genetic material or amending the Human Tissue Acts to regulate the collection, storage, use of, and access to, genetic samples. These options are examined further in Ch 20.

7.9 At present, national regulation of information privacy is provided by a complex, fragmented and overlapping set of federal, state and territory legislation. This chapter discusses whether there is a need for uniformity or greater harmonisation of laws concerning the privacy protection of human genetic information and, if so, on what basis.

7.10 The Inquiry has concluded that, while some inadequacies in the existing legislative privacy framework can be identified, these are best remedied through changes to general information and health privacy laws (including the *Privacy Act*), rather than through developing a new regulatory framework for the protection of genetic information specifically. This chapter examines the reasons for this conclusion.

7.11 On the assumption that the *Privacy Act* (amended as proposed in this Report) will continue to form the legislative underpinning for the privacy protection of genetic information, this chapter also examines a range of issues relating to the coverage of the *Privacy Act*.

Applying the Privacy Act to genetic information

7.12 The Inquiry has highlighted the privacy problems that arise in the practical application of the *Privacy Act* and the National Privacy Principles (NPPs) to genetic information.⁷ In particular, special issues may be raised by the familial nature of genetic information and by its predictive power, especially in relation to an individual's 'right not to know' about his or her long-term health prognosis.

7.13 These issues are examined in various contexts throughout this Report. More generally, submissions referred to possible problems in applying the NPPs in relation to consent requirements, the collection of clinical family history information, the disclosure of genetic information to genetic relatives of a patient or to genetic registers, and the de-identification of genetic information.⁸

7.14 Privacy NSW expressed a general concern that the NPPs are too widely drawn for the purposes of genetic information privacy. It also expressed specific concerns about consent to the use of genetic information in research.⁹ Similarly, the Australian Privacy Charter Council submitted generally that the NPPs contain too many exemptions and exceptions. The Council also raised specific concerns about the definition of 'health information', the exceptions in relation to collection and use and disclosure principles, and the application of the access principles to familial genetic information.¹⁰

7 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, Question 4–3.

8 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

9 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

10 Australian Privacy Charter Council, *Submission G120*, 18 March 2002.

The familial nature of genetic information

7.15 Many submissions referred to privacy issues arising from the fact that genetic information is familial, that is, details are shared between living and deceased relatives and the information revealed may have implications for all of them.¹¹ As the New South Wales Health Department observed:

Privacy principles have been designed with the individual's right to privacy paramount. Handling genetic information requires a different view concerning ownership of shared information. Information about a deceased relative may be of value to one individual but it may also reveal information about another relative whose identity could be ascertained, thereby conflicting with their right to privacy.¹²

7.16 Submissions confirmed that the familial or collective nature of genetic information is a characteristic that needs to be given special attention in considering the application of information privacy principles to genetic information.¹³ For example, Dr Graeme Suthers noted that privacy conflicts in the clinical use of genetic information

usually have to do with the familial nature of the disorder or information rather than its 'genetic' nature *per se*. It is not the DNA basis of the diagnosis that is of concern. Similar conflicts could arise in relation to familial disorders that are diagnosed by non-DNA means. In a society that places such a premium on individual autonomy, the fact that we are irrevocably linked to other people can cause difficulties.¹⁴

7.17 The Royal College of Pathologists of Australasia observed that privacy principles that apply to individuals present practical difficulties to health service providers who deal with genetic information that is, by its nature, shared.¹⁵ Similarly, Dr Ian Turnbull expressed concern that:

Given that genetic information obtained from one or more individuals immediately provides information, within defined probability limits, about other individuals, are the privacy and other rights of those other individuals (third parties) properly protected ...?¹⁶

11 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Australian Privacy Charter Council, *Submission G120*, 18 March 2002; Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002; I Turnbull, *Submission G059*, 14 January 2002; G Suthers, *Submission G026*, 30 November 2001; Royal College of Pathologists of Australasia, *Submission G144*, 25 March 2002.

12 NSW Health Department, *Submission G303*, 13 January 2003.

13 In this Report, the term 'information privacy principles' is used generally to refer to the Information Privacy Principles (IPPs) and National Privacy Principles (NPPs) set out in the *Privacy Act* as well as to similar sets of principles set out in state and territory information and health privacy legislation.

14 G Suthers, *Submission G026*, 30 November 2001.

15 Royal College of Pathologists of Australasia, *Submission G144*, 25 March 2002. It has even been suggested that the term 'genetic privacy' is a contradiction in terms and that the 'notions of interconnectedness and responsibility to others which are brought to the fore in the genetic sphere' challenge the primacy afforded to personal autonomy (including through the recognition of individual privacy rights): See A Sommerville and V English, 'Genetic Privacy: Orthodoxy or Oxymoron?' (1999) 25 *Journal of Medical Ethics* 144.

16 I Turnbull, *Submission G277*, 21 December 2002.

7.18 One means of balancing the interests of individuals in the privacy of their genetic information with the interests of family members in genetic information that they need for their own health care, is to differentiate between two aspects of genetic information. This approach is advocated by Professor Loane Skene. In her view, many misunderstandings about genetic information can be avoided if people understand that some information is familial and should be shared between genetic relatives, while some is personal to individuals and should be protected by the same privacy principles as any other personal information.¹⁷ The familial aspect of the information is that a mutation exists in a family. This will sometimes be known by family members in any event because earlier members will have died or suffered from the mutation.

7.19 However, family members may not keep in touch with one another or may not know family medical history. Also mutations can occur spontaneously so it is possible that a person might be the first in the family to have an inheritable mutation. If that occurs, it may be very important for other family members to have access to information about the affected relative, or even to test that person's tissue, to look after their own health. Professor Skene states that access to information or tissue for such purposes should be allowed. However, the person's own genetic status (namely, whether he or she is positive or negative for the family mutation) is personal information and should not be revealed to other family members without the person's consent.

7.20 Professor Skene has argued that a 'medical model' of regulation should apply to genetic testing, rather than a regulatory approach like the *Privacy Act*, which focuses solely on protecting an individual's right to privacy. The medical model is based primarily on what doctors consider to be best practice in providing medical care for patients and their families.¹⁸ Control of genetic samples and information, in so far as they are familial and not personal, would be shared among genetic relatives.

On this model, people would not have the ultimate right to 'control' their information and the use of their tissue taken for genetic testing (though the nature and use of the information and tissue will be fully discussed at the outset before testing is undertaken); and doctors will have a special role in providing and imparting genetic information that may appear contrary to their traditional obligation to maintain patient confidentiality.¹⁹

7.21 The Office of the Federal Privacy Commissioner (OFPC) responded that, while this approach had merit, it failed to take into account a number of other relevant factors, including the fundamental nature of an individual's right to maintain a degree of control over the handling of their health information, the need for individual rights

17 See L Skene, 'Patients' Rights or Family Responsibilities? Two Approaches to Genetic Testing' (1998) 6 *Medical Law Review* 1.

18 Ibid, 24.

19 Ibid, 24. A contrary view is expressed in D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130. See also L Skene, 'Genetic Secrets and the Family: A Response to Bell and Bennett' (2001) 9 *Medical Law Review* 162.

to be balanced with the legitimate interests of third parties and of the community in general, and the rights of individuals ‘not to know’.²⁰

7.22 However, Professor Skene’s approach to the two aspects of genetic information meets these concerns to a considerable degree. Individuals would retain the right to have their own genetic status kept private, although they could be compelled to reveal the familial aspect of the information where other family members have a legitimate need to know this for their own health care. Where the information can provide a clear clinical benefit for family members, it can usually be assumed that those members would want to know the information and their right not to know is somewhat rhetorical (see below).

7.23 The familial nature of genetic information is also discussed in the context of the Inquiry’s recommendations concerning how health professionals collect and deal with genetic information about genetic relatives and in relation to first-degree genetic relatives’ rights of access to genetic information (Chapters 21–22).

The right not to know

7.24 The right not to know has been stated as the right people should have to be protected from information that their own bodies can yield, based on the ethical principle of respect for autonomy. This principle may be seen as having particular application to genetic testing because of the predictive power, or perceived predictive power, of genetic information in relation to a person’s long-term health prognosis and other physical and behavioural characteristics.²¹

7.25 This right was a common topic in submissions that considered the application of information privacy principles to genetic samples and information.²² The Centre for Law and Genetics submitted that the right not to know should be recognised under privacy legislation. They submitted that, at present

[n]ot only is there no specific recognition or protection of the right not to know, there are provisions contained in the Act (in particular, NPP 1.5) which may encourage information to be inappropriately disclosed to the individual about whom it is collected in an over zealous attempt to comply with the requirements of the legislation.²³

20 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

21 Privacy Commissioner of Canada, *Genetic Testing and Privacy* (1992), Privacy Commissioner of Canada, Ottawa, 30–31. The right not to know is supported in a number of international instruments: *Universal Declaration on the Human Genome and Human Rights*, UNESCO, <www.unesco.org/ibc/en/genome/projet/>, 19 February 2003, Art 5; *Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine*, opened for signature 4 April 1997, ETS No 164, (entered into force on 1 December 1999), Art 10.

22 *Confidential Submission G051CON*, 14 January 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002.

23 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

7.26 Under the *Privacy Act* the right not to know is protected to some extent by the requirement that, in most circumstances, genetic testing will not be permitted without the consent of the individual concerned, given after appropriate information has been provided to them.²⁴

7.27 The specific concern raised by the Centre for Law and Genetics related to NPP 1.5, which provides that, if an organisation collects personal information about an individual from someone else, it must take reasonable steps to ensure that the person is or has been made aware of the collection. The concern is that if genetic information is collected from one individual then, in some circumstances, there may be an obligation to notify genetic relatives about this information, thereby revealing information about their own genetic status.

7.28 DP 66 noted that a Temporary Public Interest Determination (PID) had been issued by the federal Privacy Commissioner and that, at least where a health service is being provided, the Temporary PID may be sufficient to ease concerns that information will be inappropriately disclosed as part of a notification under NPP 1.5.²⁵

7.29 However, the final PIDs, issued on 15 October 2002, do not exempt organisations from their obligations to adhere to NPP 1.5.²⁶ Therefore, organisations remain obliged to take reasonable steps to ensure that third parties are informed about the collection of information. This is a matter that requires more consideration in the context of the development of a new PID dealing with the operation of genetic registers (see Chapter 22).

7.30 The right not to know is also examined in the context of the Inquiry's recommendations concerning the disclosure by health professionals of genetic information to genetic relatives without the consent of their patients (see Chapter 21).

The Privacy Act

7.31 There is a great deal of existing federal, state and territory regulation of information privacy. At the federal level, information privacy is regulated by the *Privacy Act*. While the *Privacy Act* is the major focus of consideration in this chapter, state and territory legislation is also discussed in the context of the need for greater harmonisation across Australian jurisdictions.

24 In the research context, the National Statement also requires that research participants be asked, at the time of giving consent, whether or not they wish to receive the results of the tests that relate to them as individuals: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.10]; [16.15]–[16.16].

25 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, [7.142]. See Office of the Federal Privacy Commissioner, *Temporary Public Interest Determination No. 2001-1*, <www.privacy.gov.au/act/public_interest/index.html#2>, 22 July 2002.

26 *Privacy Commissioner Public Interest Determination No. 9 2002* (Cth); *Privacy Commissioner Public Interest Determination No. 9A 2002* (Cth). These PIDs are discussed in more detail in Ch 21–22.

7.32 The *Privacy Act* is intended to protect the personal information of individuals and to give them greater control over how that information is collected, used and disclosed. The legislation sets out certain safeguards that government, private sector organisations and individuals must observe in collecting, storing, using and disclosing personal information. It also gives individuals rights to access and correct their own personal information.

7.33 The *Privacy Act* contains privacy safeguards set out in a number of Information Privacy Principles (IPPs) and NPPs, which have the force of law.²⁷ The IPPs cover collection, storage and security, use, disclosure and access to ‘personal information’, which is in a ‘record’ held by an ‘agency’, as those terms are defined in the *Privacy Act*. With limited exceptions, agencies include only Commonwealth and Australian Capital Territory (ACT) public sector entities.

7.34 Most private sector organisations are covered by the new private sector provisions of the *Privacy Act*.²⁸ The organisations covered include all health services holding ‘health information’ as defined by the *Privacy Act*. The Act extends privacy protection to personal information collected, used and disclosed by private sector entities such as private hospitals, health practitioners and insurance companies.²⁹

7.35 Private sector organisations must comply with the NPPs. The NPPs set out how organisations should collect, use and disclose personal information, maintain data quality, keep personal information secure, maintain openness, allow for access and correction of personal information, use identifiers, allow anonymity, conduct transborder data flows and collect sensitive information. Some of these principles are similar to the IPPs. However, among other differences, the NPPs contain special provisions for ‘sensitive information’ and ‘health information’, which is a subset of ‘sensitive information’.

State and territory privacy legislation

7.36 Most state and territory government bodies and local governments are not covered by the *Privacy Act*.³⁰ In particular, public hospitals and other state, territory or local government health service providers are not subject to the *Privacy Act*.³¹ Further,

27 *Privacy Act 1988* (Cth) s 14 (IPPs), Sch 3 (NPPs).

28 The *Privacy Amendment (Private Sector) Act 2000* (Cth) came into operation on 21 December 2001 and extended the coverage of the *Privacy Act* to much of the private sector. The new private sector provisions of the *Privacy Act* apply to ‘organisations’, which include partnerships, unincorporated associations and bodies corporate. An individual who is self-employed or a sole trader is considered an organisation for the purposes of the *Privacy Act*. Organisations are generally responsible for the actions of their employees, contractors and subcontractors, all of which are covered by the *Privacy Act*: ss 6C, 8.

29 To the extent that genetic information comprises ‘personal information’ and/or ‘health information’ as those terms are defined in the Act: *Privacy Act 1988* (Cth) s 6.

30 However, state or territory bodies that are incorporated companies, societies or associations are deemed to be organisations for the purposes of the *Privacy Act* and will be subject to the legislation. There is a provision in the legislation for these bodies to be prescribed out of the coverage of the *Privacy Act*: s 6C.

31 Although some Commonwealth services provided within state or territory public hospitals and those provided under contract to the Commonwealth, such as clinical services for the Department of Veterans’ Affairs may be covered by the *Privacy Act*: s 8.

private sector health service providers working under contract for a state, territory or local government agency are not covered by the *Privacy Act*.³²

7.37 Some States and Territories have information or health privacy legislation that is applicable to the handling of genetic information. This legislation applies privacy principles similar to those in the *Privacy Act* to ‘personal information’, ‘health information’ or ‘personal health information’ as those terms are defined in the various Acts. In summary, the coverage of this legislation is as follows:

- In New South Wales, the *Privacy and Personal Information Protection Act 1998* (NSW) regulates the privacy of health and other personal information handled by the New South Wales public sector. When it comes into force towards the end of 2003,³³ the *Health Records and Information Privacy Act 2002* (NSW) will regulate the privacy of health information handled by the New South Wales private and public sectors.
- In Victoria, the *Health Records Act 2001* (Vic) regulates the privacy of health information handled by the Victorian private and public sectors. The *Information Privacy Act 2000* (Vic) regulates personal information handled by the Victorian public sector.
- In the ACT, the *Health Records (Privacy and Access) Act 1997* (ACT) regulates the privacy of health information handled by the ACT private and public sectors.

7.38 In addition to privacy legislation, the Commonwealth, States and Territories have other legislation relating to the administration of public health services, which contains provisions to protect the confidentiality of genetic and other health information obtained by public sector health administrators and health service providers in the course of their employment.³⁴ The operation of this legislation is not the subject of specific consideration in this Report.

Harmonisation of health privacy law

7.39 While the *Privacy Act* creates a framework for national regulation of health information in the private sector, as well as protecting privacy in the Commonwealth public sector, there is no comprehensive framework for consistent national regulation of health information across public and private sectors, state and federal. Instead, regulation of information and health privacy is provided by complex, fragmented and overlapping federal, state and territory legislation. Health information is subject to different protection depending on whether it is held by a Commonwealth agency, state or territory agency or private sector organisation.

32 Although they may be covered by state or territory information or health privacy legislation.

33 Australian Health Ministers’ Advisory Council National Health Privacy Working Group, *National Health Privacy Code (draft) Consultation Paper* (2002), AHMAC, Canberra, 8.

34 For example, *Health Insurance Act 1973* (Cth); *National Health Act 1953* (Cth); *Health Administration Act 1982* (NSW); *Private Hospitals Regulations 1996* (NSW); *Nursing Homes Regulation 1996* (NSW); *Day Procedure Centres Regulation 1996* (NSW); *Health Services Act 1988* (Vic); *South Australian Health Commission Act 1976* (SA).

7.40 The situation is complicated by the fact that many different organisations may be responsible for delivery of health services to any one individual. Therefore, different legal regimes and privacy protection, with different privacy standards, may apply to different parts of the health information relating to a single individual.

7.41 Problems that arise from the lack of uniformity are discussed in specific contexts elsewhere in this Report³⁵ and were highlighted in submissions.³⁶ For example, the Centre for Law and Genetics noted that:

The privacy legislation within the various States and Territories is incomplete and lacking in uniformity. In Tasmania, for example, there is no privacy legislation. Where privacy legislation does exist, it is not necessarily compatible with either the public sector or private sector provisions in the federal Act.³⁷

7.42 The Commonwealth Attorney-General's Department observed that, as with most other areas of regulation, practical difficulties arise when organisations are required to comply with a number of related but conflicting laws:

It leads to greater expense when they have to seek professional advice regarding their legal obligations and implement different procedures for compliance. Where relevant it can also lead to forum shopping by consumers in relation to complaint-handling. This is an unsatisfactory situation and should be avoided by having national standards where possible.³⁸

7.43 Particular complexity arises where States and Territories have health privacy legislation purporting to cover the private sector, as is the case in New South Wales, Victoria and the ACT.³⁹ Various aspects of this state and territory legislation may be inconsistent with the *Privacy Act* and may create confusion and uncertainty for those organisations and individuals needing to comply with both sets of regulation.⁴⁰

7.44 For example, the NPPs in the federal *Privacy Act* permit personal information to be used or disclosed 'where the use or disclosure is required or authorised by or under law'.⁴¹ This provision would appear to allow disclosure permitted under state laws. State legislation, therefore, may effectively extend the circumstances under which the disclosure of health information is otherwise permitted by the NPPs.⁴² On the other hand, disclosure permitted under the NPPs but not under

35 See eg in relation to the coverage of genetic samples (Ch 8); regulation of human genetic research databases (Ch 18); genetic registers (Ch 22).

36 Centre for Law and Genetics, *Submission G048*, 14 January 2002; Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

37 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

38 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

39 *Health Records and Information Privacy Act 2002* (NSW); *Health Records Act 2001* (Vic); *Health Records (Privacy and Access) Act 1997* (ACT).

40 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

41 *Privacy Act 1988* (Cth) NPP 2.1(g).

42 See R Magnusson and C Clarke, 'Data Registers in Respiratory Medicine: A Pilot Evaluating Compliance with Privacy Laws and the National Statement on Ethical Conduct in Research Involving Humans' (2002) 10 *Journal of Law and Medicine* 69, 73-74.

state law may nevertheless be lawful under s 109 of the Constitution, which gives paramount operation to federal law where it is inconsistent with state law.⁴³

7.45 Differing submissions were made to the Inquiry regarding the relationship between federal privacy laws on the one hand, and state and territory privacy laws on the other. The *Privacy Act* provides that it is not to affect the operation of a law of a State or Territory that makes provision with respect to the collection, holding, use, correction, disclosure or transfer of personal information capable of operating concurrently with the Act.⁴⁴

7.46 The Commonwealth Attorney-General's Department stated that this provision is not intended to enable state and territory law to regulate the same types of personal information and organisations that are regulated by the *Privacy Act*.⁴⁵ In contrast, Privacy NSW submitted that the States should be free to 'enhance the Commonwealth's minimum standards in state legislation that provides for more stringent genetic privacy protection'. It was said that this might require a savings clause in the *Privacy Act* similar to those in other federal human rights legislation.⁴⁶

7.47 The proposition that there should be uniformity or greater harmonisation of federal, state and territory laws concerning the privacy protection of human genetic information met with widespread approval.⁴⁷ Privacy NSW summarised the desirability of a uniform approach in the following terms:

A uniform approach to genetic information privacy is essential to ensure that all persons have equal protection regardless of where they live and who handles their genetic information. Widely differing standards of protection not only undermine human rights, they also undermine public confidence in the way that institutions

43 See *Ibid*, 74.

44 *Privacy Act 1988* (Cth) s 3.

45 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

46 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

47 See *Ibid*; Centre for Law and Genetics, *Submission G048*, 14 January 2002; Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; New South Wales Nurses' Association, *Submission G090*, 21 January 2002; Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002; Sydney IVF Limited, *Submission G062*, 14 January 2002; Australian Medical Association, *Submission G091*, 29 January 2002; Queensland University of Technology, *Submission G109*, 14 March 2002; Disability Discrimination Legal Service, *Submission G146*, 28 March 2002; Australian Academy of Science, *Submission G097*, 21 January 2002; Australian Privacy Charter Council, *Submission G120*, 18 March 2002; Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; J Fleming, *Submission G241*, 20 December 2002; Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

handle their personal information, especially in an increasingly networked information environment. Lack of uniformity can also add to confusion for those responsible for handling personal information, as well as obstruct cross-border flows of information.⁴⁸

7.48 The Commonwealth Attorney-General's Department stated that:

The Commonwealth is concerned about the difficulties caused by the existence of different health information privacy laws across Australia. It is in the interests of both consumers and health service providers that clear and consistent health information privacy laws apply across Australia. The Commonwealth will discuss this issue with State and Territory Governments in the context of the development of the National Health Privacy Code and its implementation.⁴⁹

7.49 The means by which uniformity or greater harmonisation of health privacy law, as it applies to genetic information, should be pursued is problematic. Possible approaches to harmonisation include the development of a National Health Privacy Code, new federal, state and territory health privacy legislation, or the development of a regulatory framework specifically for genetic information.

A National Health Privacy Code

7.50 The Australian Health Ministers' Advisory Council (AHMAC) has formed a joint Commonwealth, State and Territory National Health Privacy Working Group to work towards the establishment of a nationally consistent regime for the protection of health information in both the public and private sectors.

7.51 The first step in this process is the development of a National Health Privacy Code, a draft of which was circulated for public comment in December 2002.⁵⁰ The Consultation Paper⁵¹ on the Code highlights the need for uniform rules regarding the handling of health information and the fact that this need has become 'even more pressing with the emerging developments in the management of electronic health records'.⁵²

7.52 The Code is intended to comprise a nationally integrated framework for the protection of personal health information. The framework aims to:

- achieve national consistency between the public and private sectors;
- safeguard the privacy and dignity of all individuals (whether they are health care consumers or health care providers); and

48 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

49 Commonwealth Attorney-General's Department, *Submission G228*, 12 December 2002.

50 Australian Health Ministers' Advisory Council National Health Privacy Working Group, *Draft National Health Privacy Code* (2002), AHMAC, Canberra.

51 Australian Health Ministers' Advisory Council National Health Privacy Working Group, *National Health Privacy Code (draft) Consultation Paper* (2002), AHMAC, Canberra.

52 *Ibid.*, 9. These developments include the proposed national health information network (*HealthConnect*) and the Better Medication Management System (BMMS).

- take into account initiatives proposed under the national health information network, *HealthConnect*.⁵³

7.53 The mechanism for implementing the Code is still under consideration and will be discussed by the AHMAC Working Group in more detail once the content of the Code has been agreed.⁵⁴

7.54 One option is for the National Health Privacy Code to operate as a code under Part IIIAA of the *Privacy Act*.⁵⁵ Part IIIAA provides for a process by which organisations may agree to be bound by a privacy code approved by the Privacy Commissioner,⁵⁶ which includes levels of privacy protection at least equivalent to the NPPs.⁵⁷ For the purposes of the *Privacy Act*, the term ‘organisation’ covers only private sector organisations. However, on the request of the government of a State or Territory, regulations may prescribe an instrumentality of a State or Territory as an organisation for the purposes of the Act.⁵⁸ This provides a mechanism by which the operation of an approved code might be extended to state and territory public sector health service providers.

7.55 The AHMAC process is the first attempt to develop a comprehensive approach to harmonisation of health privacy law. In DP 66, the Inquiry stated that it was unlikely that the AHMAC process would lead to uniformity or greater harmonisation of health privacy law in the short term.⁵⁹ From one perspective, recently enacted state and territory health privacy legislation might be seen as running counter to the proposal to develop a National Health Privacy Code to provide consistency across all jurisdictions.⁶⁰ However, implementation of these new state and territory laws could equally be seen as recognition of the need for a consistent national health privacy regime.⁶¹

Federal health privacy legislation

7.56 Other means of pursuing uniformity or greater harmonisation of health privacy law might include enacting new federal health privacy legislation to regulate the handling of health information in both the Commonwealth public sector and private sector,⁶² and to serve as a model for similar state and territory legislation.

53 Ibid, 11.

54 Ibid, 15.

55 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

56 As at January 2003, two codes had been approved by the Privacy Commissioner: Office of the Federal Privacy Commissioner, *Register of Approved Business Codes*, Office of the Federal Privacy Commissioner, <www.privacy.gov.au/business/codes/index.html#1>, 29 January 2003.

57 *Privacy Act 1988* (Cth) s 18BB.

58 Ibid s 6C.

59 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [7.33].

60 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002.

61 Department of Human Services Victoria - Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002.

62 At present, the handling of personal information (including health information) in the Commonwealth public sector is governed by the IPPs and in the private sector by the NPPs.

7.57 Differing views have been expressed about whether federal health privacy legislation is desirable. The OFPC opposed the idea and submitted that:

[a]dditional legislation of that nature will create problems of compatibility with the existing privacy framework. It would prejudice attempts to realise a consistent national standard for the protection of health information privacy. The introduction of a separate scheme to protect health information privacy, or even genetic information privacy, intended to co-exist with existing forms of regulation, would be likely to encourage forum-shopping or 'regulatory arbitrage'. In the interests of achieving a single uniform scheme of privacy regulation for health information, it would be preferable to concentrate on improving existing legislation within the current regulatory framework. Any special legislative protections for health information generally or genetic information in particular should be effected within this framework.⁶³

7.58 Other submissions suggested that there should be federal health privacy legislation, separate from the *Privacy Act*.⁶⁴ For example, the Centre for Law and Genetics considered that deficiencies identified in the privacy protection of genetic information should be addressed through new health privacy legislation:

Within the framework of privacy legislation specific to health, provisions could be included giving special recognition to the protection of genetic information to address perceived deficiencies in this area, as indeed could be done for other areas of health where there may be a need for particular protection. After all, genetic information is clearly health information and is best dealt with within this context, with the addition of specific provisions as appropriate. This would ensure a coherent approach is taken to the issue, in a manner consistent within a general health privacy framework.⁶⁵

7.59 While the House of Representatives Standing Committee on Legal and Constitutional Affairs ultimately recommended that health information be included in the Privacy Amendment (Private Sector) Bill 2000 (Cth),⁶⁶ the Centre for Law and Genetics observed that

the only reason that the committee ultimately decided to recommend that health information should remain part of the Bill, was because it thought it unlikely that a consensus could be achieved in the near future that would lead to the development of a separate legislative or regulatory code governing health services. Its recommendations were, instead, directed towards achieving such reforms in the future and therefore retaining the legislation's coverage of health information, at least on an interim basis, to ensure an acceptable level of privacy and access rights throughout Australia.⁶⁷

63 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

64 Centre for Law and Genetics, *Submission G048*, 14 January 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Australian Medical Association, *Submission G091*, 29 January 2002. The Australian Medical Association suggested that, because the *Privacy Act* is not health-specific, it does not deal adequately with the privacy of electronic health records or genetic privacy issues.

65 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

66 Standing Committee on Legal and Constitutional Affairs, *Advisory Report on the Privacy Amendment (Private Sector) Bill 2000* (2000), House of Representatives, Parliament of the Commonwealth of Australia, Canberra, Ch 6–7, rec 14–15.

67 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

7.60 Other submissions suggested that existing privacy legislation is adequate to protect human genetic information⁶⁸ or that the reform priority should be on selective changes to the existing regulatory framework.⁶⁹ For example, Privacy NSW favoured an approach that

identifies the weaknesses in the way existing privacy rules are expressed and which expands their scope to make them more responsive to the challenge proposed by genetic privacy issues.⁷⁰

Genetic privacy legislation

7.61 A third approach to harmonisation would involve the development of a regulatory framework specifically for genetic information. At the federal level, such an approach was taken in the Genetic Privacy and Non-discrimination Bill 1998 (Cth), introduced by Democrats Senator Natasha Stott Despoja.⁷¹ The Bill, which was last debated on 5 October 2000 in the Senate, was restored to the Notice Paper for the Senate on 14 May 2002. The Bill addressed genetic information and deals with information privacy, consent and genetic discrimination.⁷²

7.62 There was little support in consultations or submissions to this Inquiry for new legislation dealing specifically with genetic privacy. Most submissions that considered the issue opposed such an approach.⁷³ However, the Department of Human Services South Australia submitted that:

Given the past difficulties in establishing uniform privacy legislation, it may be more efficient and effective to develop specific legislation dealing with genetic information. This legislation could also establish the HGCA and provide for a separate entity to

⁶⁸ Life Sciences Network, *Submission G129*, 19 March 2002.

⁶⁹ Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Australian Defence Force, *Submission G291*, 23 December 2002.

⁷⁰ Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

⁷¹ The Genetic Privacy and Non-discrimination Bill 1998 (Cth) pre-dated the enactment of the *Privacy Amendment (Private Sector) Act 2000* (Cth). Senator Stott Despoja noted that there would have been no need for the former to deal with privacy if an effective legislated scheme for privacy protection had already been implemented. Senate Legal and Constitutional Legislation Committee, *Provisions of the Genetic Privacy and Non-discrimination Bill 1998*, The Parliament of Australia, <www.aph.gov.au/senate/committee/legcon_ctte/genetic/index.htm>, 21 August 2002, additional comments by Senator Natasha Stott Despoja, 34.

⁷² See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [4.124]–[4.131].

⁷³ Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002; K Liddell, *Submission G141*, 23 March 2002; R Magnusson, *Submission G039*, 10 January 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002; N Saunders and P Komesaroff, *Submission G084*, 9 January 2002; Australian Medical Association, *Submission G091*, 29 January 2002; Queensland University of Technology, *Submission G109*, 14 March 2002; Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Australian Society for Medical Research, *Submission G124*, 18 March 2002. The NSW Nurses' Association and the Genetic Support Council Western Australia suggested that specific genetic privacy legislation might be desirable: New South Wales Nurses' Association, *Submission G090*, 21 January 2002; Genetic Support Council WA, *Submission G112*, 13 March 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

license and regulate genetic testing laboratories. Recent precedents of positive uniform national legislation include food regulation and genetically modified organisms. This would also ensure that current “gaps” arising from the limited application of the Commonwealth Privacy Act (non-coverage of “employee records” and exempt small businesses etc) and the differences in the various states’ legislation could be filled without the need to amend ad hoc pieces of existing legislation.⁷⁴

Inquiry’s views

7.63 Given the plethora of existing regulation relating to the privacy protection of genetic information, it seems more appropriate to amend existing legislation to ensure that issues of genetic privacy are adequately covered rather than to add another layer of complexity by enacting genetic privacy legislation.

7.64 In particular, there would be considerable practical difficulty in defining the respective coverage of genetic privacy legislation and other information and health privacy legislation. Genetic information already forms part of ordinary clinical health information. It can be expected that genetic information will become increasingly important in the prevention, diagnosis and treatment of disease. As this occurs it will become increasingly difficult, if not meaningless, to distinguish between genetic information and other health information located, for example, in medical records held by health service providers.⁷⁵

7.65 While genetic information has some special characteristics that distinguish it from most other forms of personal information,⁷⁶ the Inquiry has concluded that genetic privacy issues and reform options are often similar to those applicable to information privacy generally and, in particular, to the privacy of medical records and other health information.

7.66 The Terms of Reference ask the Inquiry, in reporting on the regulatory framework required to protect the privacy of human genetic samples and information, to have regard to existing or proposed Commonwealth legislation and legislation in other jurisdictions.

7.67 Deficiencies in the existing regulatory framework for information and health privacy are a focus of the Inquiry only to the extent that they concern the protection of genetic samples and information specifically. The Terms of Reference do not anticipate that the Inquiry will review the adequacy of health information privacy laws more generally. Nor do they demand that the Inquiry reach a concluded view about whether privacy protection is best provided within the framework of the *Privacy Act* or in new information or health privacy legislation.

74 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

75 R Magnusson, *Submission G039*, 10 January 2002. Australian Society for Medical Research, *Submission G124*, 18 March 2002. The Society stated that ‘specific, all-encompassing genetic legislation would provide considerable burden and limited benefits’.

76 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [4.10]–[4.14].

7.68 Consultations and submissions have emphasised the importance of greater harmonisation in information and health privacy law, both within the federal sphere and between federal, state and territory laws. For example, the New South Wales Health Department observed that

due to the shared nature of genetic information, the current situation creates differing management of privacy issues for family members residing in different states or accessing services in the public or private sector.⁷⁷

7.69 Effective protection of genetic information requires that efforts continue to be made to achieve a harmonised approach. While the Inquiry expresses no view on the exact mechanism by which such harmonisation should be pursued, it considers that Commonwealth, state and territory governments should give priority to this policy aim. In this context, the AHMAC process is an obvious starting point for harmonisation initiatives.

Recommendation 7–1. As a matter of high priority, the Commonwealth, States and Territories should pursue the harmonisation of information and health privacy legislation as it relates to human genetic information. This would be achieved most effectively by developing nationally consistent rules for handling all health information. (See also Recommendation 8–1 in relation to genetic samples.)

Recommendation 7–2. States and Territories and privacy regulators should consider harmonising their privacy regimes, as applicable, in a manner consistent with the Recommendations in this Report. (See also Recommendations 7–4 to 7–7, 8–1 to 8–4, 21–1 to 21–3, and 22–1.)

Recommendation 7–3. The Commonwealth, States and Territories should take into account the Recommendations in this Report in developing the proposed National Health Privacy Code. (See also Recommendations 7–4 to 7–7, 8–1 to 8–4, 21–1 to 21–3, and 22–1.)

Genetic information and health information

7.70 The *Privacy Act* applies privacy protection to an individual's 'personal information'. The Act recognises that a subset of personal information is 'sensitive information', which due to its nature attracts some higher standards of privacy protection. 'Health information' is one type of 'sensitive information'.

7.71 Genetic information is not specifically referred to in the *Privacy Act*. The Explanatory Memorandum to the Privacy Amendment (Private Sector) Bill 2000 (Cth) stated that the definition of 'health information' covered genetic information, while at

77 NSW Health Department, *Submission G303*, 13 January 2003.

the same time noting that the NPPs were not designed to address the unique privacy issues associated with the handling of genetic information.⁷⁸

7.72 For the purposes of the private sector provisions, the *Privacy Act* creates a special category of ‘sensitive information’ and gives this a higher level of protection. Sensitive information is information or an opinion about an individual’s racial or ethnic origin; political opinion; political association membership; religious beliefs, affiliations or philosophical beliefs; professional or trade association membership; union membership; sexual preferences; criminal record; or is health information about an individual.⁷⁹

7.73 ‘Health information’ is separately defined as

- (a) information or an opinion about:
 - (i) the health or a disability (at any time) of an individual; or
 - (ii) an individual’s expressed wishes about the future provision of health services to him or her; or
 - (iii) a health service provided, or to be provided, to an individual;
 that is also personal information; or
- (b) other personal information collected to provide, or in providing, a health service; or
- (c) other personal information about an individual collected in connection with the donation, or intended donation, by the individual of his or her body parts, organs or body substances.⁸⁰

7.74 There are differences in the way the *Privacy Act* treats personal information, health information and other sensitive information. Health and other sensitive information are provided higher levels of protection than ordinary personal information.⁸¹ Generally, genetic information will receive the special protection afforded to sensitive information under the *Privacy Act* if it can be defined as health information or some other element of sensitive information, for example, if it constitutes information or an opinion about an individual’s racial or ethnic origin, their sexual preferences or their criminal record.

78 In the Senate, the Bill was amended to define ‘genetic information’ and to insert genetic information into the definition of health information. This amendment and other Senate amendments relating specifically to genetic information were not accepted by the House of Representatives. The reasons of the House for disagreeing with the Senates amendments included that it would be premature to accept the amendments proposed until the government had the benefit of the report of the present Inquiry: Commonwealth of Australia, *Parliamentary Debates*, House of Representatives, 5 December 2000, 1965 (Reasons of the House of Representatives for Disagreeing to the Amendments of the Senate presented by the Hon Darryl Williams Attorney-General). See also Senate Legal and Constitutional Legislation Committee, *Provisions of the Privacy Amendment (Private Sector) Bill* (2000), The Parliament of Australia, Canberra, 26–27.

79 *Privacy Act 1988* (Cth) s 6(1).

80 *Ibid* s 6(1).

81 Subject to some limited exceptions, NPP 10 requires consent for the collection of sensitive information: compare *Ibid*, NPP 1, which requires only that individuals be informed about various matters such as their access rights, the purposes of collection and to whom the organisation usually discloses information of that kind. The use and disclosure of sensitive information other than for the primary purpose of collection is more constrained than is the case with ordinary personal information—the secondary purpose must be directly related to the primary purpose: *Privacy Act 1988* (Cth), NPP 2.1(a).

7.75 There are circumstances in which genetic information may not be health information, as defined in the *Privacy Act*. This may occur either because the information is not about health, disability or the provision of a health service (as in the case of parentage or forensic testing) or because it is not about the health or disability of an existing individual (as may sometimes be the case with genetic carrier testing, where the information is primarily about the health of future children).⁸²

7.76 The Inquiry notes that some genetic information collected for criminal forensic purposes may fall within the definition of sensitive information if it is information about an individual's criminal record but that a range of genetic information will remain outside the definitions of sensitive information and health information. The Attorney-General's Department noted that the consequences of this gap include that commercial laboratories that currently offer parentage testing may be able to use genetic information for direct marketing purposes.⁸³

7.77 Submissions indicated general support for amending the *Privacy Act* to ensure that all genetic information is treated as health information or other sensitive information under the Act.⁸⁴

7.78 Some submissions referred to the definition used in the *Health Records Act 2001* (Vic) and the *Health Records and Information Privacy Act 2002* (NSW) as appropriate models.⁸⁵ The Victorian legislation defines health information to include

other personal information that is genetic information about an individual in a form which is or could be predictive of the health (at any time) of the individual or any of his or her descendants.⁸⁶

82 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [4.75]–[4.79].

83 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002. Direct marketing is a permissible secondary use of personal information but not health information: *Privacy Act 1988* (Cth) NPP 2.1(c).

84 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002; Royal College of Pathologists of Australasia, *Submission G144*, 25 March 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; UnitingCare NSW & ACT, *Submission G052*, 14 January 2002; Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003. AGSA opposed the proposed amendment on the basis that defining all genetic information as health information would lead to confusion: Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

85 Royal College of Pathologists of Australasia, *Submission G144*, 25 March 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002 (expressing the views of the AHMAC National Health Privacy Working Group).

86 *Health Records Act 2001* (Vic) s 3(1)(d). The Act came into effect from 1 July 2002.

7.79 The New South Wales legislation defines health information to include

other personal information that is genetic information about an individual arising from a health service provided to the individual in a form that is or could be predictive of the health (at any time) of the individual or of any sibling, relative or descendant of the individual.⁸⁷

7.80 Both of these definitions would be broad enough to encompass genetic carrier testing. It is unclear whether the Victorian definition covers parentage, research or forensic testing information.⁸⁸ The New South Wales definition clearly would not cover such information because of the required connection with the provision of a health service. The AHMAC Draft National Health Privacy Code adopts the Victorian formulation⁸⁹ and is intended to include all genetic information, whether or not it has been collected in relation to the provision of a health service.⁹⁰

7.81 The Inquiry is of the view that genetic information should receive the special protection afforded to health and other sensitive information under the *Privacy Act*. The existing definitions of health information and sensitive information do not provide that level of protection for all genetic information.

7.82 The Inquiry therefore recommends that the definition of health information in the *Privacy Act* be amended to make clear that it includes genetic information predictive of health, whether or not the information is collected in relation to the health of, or the provision of a health service to, the individual or a genetic relative.⁹¹ In this recommendation, the word ‘predictive’ does not bear the technical meaning used in some clinical contexts,⁹² but is chosen for the purpose of consistency with the existing legislative definitions referred to above. The term ‘genetic relative’ seems more appropriate than the Victorian or AHMAC formulations, which refer to ‘descendants’ of the individual and do not, therefore, encompass genetic information about an individual’s siblings.

7.83 An amendment to the definition of sensitive information is also necessary to cover genetic information derived from parentage or other identification testing that is not predictive of health.

87 *Health Records and Information Privacy Act 2002* (NSW) s 6(d).

88 The Victorian definition refers to information in a form which ‘is or could be’ predictive of health. It may not require that the information be obtained for health-related purposes.

89 Australian Health Ministers’ Advisory Council National Health Privacy Working Group, *Draft National Health Privacy Code* (2002), AHMAC, Canberra cl 1.

90 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Australian Health Ministers’ Advisory Council National Health Privacy Working Group, *National Health Privacy Code (draft) Consultation Paper* (2002), AHMAC, Canberra, 18.

91 And bodily samples from individuals whose identity is apparent or reasonably can be ascertained from the sample: See Ch 8.

92 See Ch 10.

Recommendation 7–4. The Commonwealth should amend s 6 of the *Privacy Act 1988* (Cth) (*Privacy Act*) to define ‘health information’ to include genetic information about an individual in a form which is or could be predictive of the health of the individual or any of his or her genetic relatives. (See also Recommendation 8–2 in relation to genetic samples.)

Recommendation 7–5. The Commonwealth should amend s 6 of the *Privacy Act* to define ‘sensitive information’ to include human genetic test information.

Deceased individuals

Extending the Privacy Act to deceased individuals

7.84 DP 66 asked whether the *Privacy Act* adequately deals with issues that may arise in relation to the genetic samples and information of deceased individuals.⁹³ A threshold question concerns the extent to which it makes sense to protect the privacy of deceased individuals through the application of information privacy principles. Logically, the privacy interests being protected can only be those related to surviving individuals or, more abstractly, to the memory of the deceased.⁹⁴

7.85 The *Privacy Act* regulates the handling of personal information about individuals. Section 6 of the Act defines an individual as ‘a natural person’. The *Privacy Act* does not cover genetic information about deceased persons. An individual may only make a complaint under the *Privacy Act* in relation to an interference to his or her own privacy.

7.86 This position may be contrasted with that under New South Wales and Victorian health privacy legislation, which cover personal information about individuals who have been dead for not more than 30 years⁹⁵—a period of protection

93 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Question 7–6. Related issues concerning individuals’ rights of access to the bodily samples of genetic relatives are also discussed in Ch 9.

94 Note that, in Australia, no action lies for defamation of a deceased person, however distressing to living relatives or friends: J Fleming, *The Law of Torts* (9th ed, 1998) LBC Information Services, Sydney, 585. See also Australian Law Reform Commission, *Unfair Publication: Defamation and Privacy* (1979), Australian Government Printing Service, Canberra [99]–[102]. The Commission recommended that a person representing a deceased defamed person should be able to sue, but only where the publication is made within 3 years of death and only to obtain a correction, declaratory order or injunction (and not damages). A patient’s right to confidentiality continues after the patient’s death, so that a doctor may not reveal confidential information after death except with proper legal authority: L Skene and J Savulescu, ‘Who Has the Right to Access Medical Information from a Deceased Person? Ethical and Legal Perspectives’ (2000) 8(August) *Journal of Law and Medicine* 81, 82.

95 New South Wales and Victorian health privacy legislation provides that the term ‘personal information’ ‘does not include information about an individual who has been dead for more than 30 years’: *Health Records Act 2001* (Vic) s 3; *Health Records and Information Privacy Act 2002* (NSW) s 5. The Victorian legislation also expressly provides that the Act applies ‘in relation to a deceased individual who has been dead for 30 years or less, so far as it is reasonably capable of doing so, in the same way as it applies in

that is consistent with the 30 year period after which government archival records are generally open to public access.⁹⁶

7.87 In Victoria, the rights or powers conferred on individuals under the *Health Records Act 2001* (Vic) are exercisable by a legal representative (executor or administrator) of the deceased individual.⁹⁷ New South Wales privacy legislation does not directly address the issue of obtaining consent in relation to deceased individuals.⁹⁸

7.88 The AHMAC Draft National Health Privacy Code covers personal information about individuals who have been dead for not more than 30 years.⁹⁹ The consultation paper notes that there are

many situations where information may be sought on deceased persons, for example a health provider could receive requests for such information from relatives, friends legal representatives, the media and medical researchers. Some of these requests may be legitimate and appropriate, while others may be motivated by reasons unrelated to the interests of the individual involved.¹⁰⁰

7.89 The Commonwealth Department of Health and Ageing noted that, because the genetic health information of a deceased person is also information about their family members or descendants, there may be an argument for protecting the genetic information long after a person's death.¹⁰¹ Other submissions suggested that the *Privacy Act* should be amended to cover genetic information about,¹⁰² and samples from,¹⁰³ deceased individuals.¹⁰⁴

relation to an individual who is not deceased': *Health Records Act 2001* (Vic) s 95. ACT legislation also covers personal information about deceased individuals but has no 30 year limitation on coverage: See *Health Records (Privacy and Access) Act 1997* (ACT) s 4 (definition of 'consumer'); s 27. In the United States, federal health privacy regulations, effective on 26 February 2001, extend protection of protected health information about deceased individuals for as long as the covered entity maintains the information: *Health Insurance Portability and Accountability Act 1996: Standards for the Privacy of Individually Identifiable Health Information 45 CFR Part 164 1996* (USA) s 164.502(f).

96 For example, *Archives Act 1983* (Cth) s 3(7).

97 *Health Records Act 2001* (Vic) s 95(2); s 3. See also *Health Records and Information Privacy Act 2002* (NSW) s 8.

98 Privacy NSW submitted amendments to the federal *Privacy Act* should include provisions for decision making, either by a next-of-kin or an authorised person, regarding the human tissue and health information of deceased individuals: Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002.

99 Australian Health Ministers' Advisory Council National Health Privacy Working Group, *Draft National Health Privacy Code* (2002), AHMAC, Canberra cl 1.

100 Australian Health Ministers' Advisory Council National Health Privacy Working Group, *National Health Privacy Code (draft) Consultation Paper* (2002), AHMAC, Canberra, 42.

101 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

102 Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; I Turnbull, *Submission G277*, 21 December 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

103 Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

104 Extension of the *Privacy Act* to information about deceased individuals was opposed in some submissions: Queensland Government, *Submission G274*, 18 December 2002.

7.90 The Inquiry believes that it is desirable that information privacy protection extend to genetic information about deceased individuals because of the implications that the collection, use or disclosure of this information may have for living genetic relatives. It appears preferable for representatives of the deceased to be able to consent to the collection, use or disclosure rather than to leave decisions about these matters outside the *Privacy Act*.

7.91 It would also be desirable for Commonwealth, state and territory privacy legislation to have consistent coverage in relation to information about deceased individuals. At present, where genetic information about a deceased individual is transferred between, for example, a Victorian clinical genetics service and a private sector clinical geneticist, the extent of privacy protection will change. While the clinical geneticist will be permitted under the federal *Privacy Act* to collect and disclose the information without the consent of the deceased's legal representative, the Victorian service may not be able to do so.

Recommendation 7–6. The Commonwealth should amend the *Privacy Act* to provide that 'health information' includes information about an individual who has been dead for 30 years or less. These amendments should include provision for decision making by next-of-kin or an authorised person in relation to the handling of a deceased individual's health information. (See also Recommendation 8–2 in relation to genetic samples.)

Access by genetic relatives

7.92 The Inquiry has been informed about situations in which individuals have been denied optimal medical care because of difficulty in obtaining access to the medical records of deceased relatives. The following example illustrates the problem:

The client is a 24 year old individual whose mother died at age 40 of bowel cancer and polyps. The father subsequently remarried and there was falling out between the children and the father so they have lost contact with him. They present for genetic advice. It is essential to know whether there were a few polyps or [whether] a condition known as Familial Adenomatous Polyposis was present. The histopathology would state this. The offspring are genetically related and therefore have a greater need for the information than the spouse who has 'moved on'. In both the ACT and Queensland there is now a blanket rule in medical records departments to prohibit access without the executor's permission. In the current case [the client] was not aware of who that was and now does not know whether annual colonoscopy or 3–5 yearly colonoscopy is necessary.¹⁰⁵

7.93 If the law requires that access to genetic information about a deceased individual can be granted only with the consent of that person's legal or other authorised representative, genetic relatives may still have problems in gaining access.

105 K Barlow-Stewart, *Correspondence*, 18 July 2002.

7.94 Whether or not privacy legislation applies to genetic information about deceased individuals, some rights in respect of genetic information may be exercisable by genetic relatives if they can show that the information is also ‘about’ them.¹⁰⁶ This is one implication of the shared or familial nature of genetic information. An individual’s genetic information is information about that individual, but it is also ‘about’ the individual’s immediate family, other genetic relatives, ethnic group, racial group, and ultimately humanity itself.

7.95 Under the existing provisions of the *Privacy Act*, it should be possible for an individual to argue that he or she has rights in relation to genetic information that is primarily about a genetic relative on the ground that the information is also, at least in part, ‘about’ the individual seeking to assert those rights (and identifies him or her).¹⁰⁷

7.96 One advantage of differentiating between the familial and personal aspects of genetic information is that it assists in this analysis. In the example related above, the familial information is that the gene for Familial Adenomatous Polyposis is present in the family. This information is ‘about’ each family member. The mother’s genetic status, though it may be inferred from her death from bowel cancer, is personal—it is ‘about’ her. The father should not be permitted to withhold the familial information from the daughter who needs to know it in order to monitor her own health.

7.97 In some circumstances, the principles for fair information handling codified in the *Privacy Act* may provide a flexible basis for dealing with this issue without further amendment. For example, if the matter were tested, it may well be that the Privacy Commissioner or a court might find that a person has a right of access to familial genetic information about his or her deceased parents or siblings (on the ground that the information is also information ‘about’ the person seeking access), but no right of access to the same information about a deceased second cousin.¹⁰⁸

7.98 However, in practice the need to show that the information is also identifying of the living genetic relative may constitute a barrier to the exercise of access rights. Therefore, other recommendations made in this Report are intended to ensure that first-degree genetic relatives are able to access genetic samples or information where such access is vital to their health. In Chapter 8 and Chapter 21 the Inquiry recommends that the *Privacy Act* be amended to provide individuals with rights to access genetic samples and familial information about their first-degree genetic relatives in certain exceptional circumstances.

106 Commonwealth Attorney-General’s Department, *Submission G158*, 7 May 2002.

107 A similar position may exist under the *Data Protection Act 1998* (UK): G Laurie, *Genetic Privacy* (2002) Cambridge University Press, Cambridge, 253–254.

108 Where it is held by public sector agencies, access to information about deceased individuals may also be sought under Commonwealth, state or territory freedom of information legislation. Similar balancing tests are applicable to FOI requests: eg *Freedom of Information Act 1982* (Cth) s 41(1) states that ‘A document is an exempt document if its disclosure under this Act would involve the unreasonable disclosure of personal information about any person (including a deceased person)’.

Small business exemption

7.99 Under the small business exemption, some small business operators are excluded from the definition of ‘organisation’ and are therefore entirely exempt from the operation of the *Privacy Act*.¹⁰⁹ Organisations cannot qualify for this exemption if, for example, they:

- provide a health service to another individual and hold any health information except in an employee record; or
- disclose personal information about another individual to anyone else for a benefit, service or advantage; or
- provide a benefit, service or advantage to collect personal information about another individual from anyone else.¹¹⁰

7.100 Dr Tim Smyth has observed:

While health service providers who hold health information are subject to the Act, irrespective of their turnover, a small business that is not a health service provider can remain exempt from the Act even though it might hold health information. A business that simply stores genetic samples or acts as a data repository, providing no health service, may not be subject to the Commonwealth Act.¹¹¹

7.101 It has also been suggested that research undertaken by a genomics company may fall outside the definition of a health service¹¹² although such an organisation might still be caught by the provisions relating to collecting or disclosing personal information for a benefit, service or advantage.¹¹³

7.102 In DP 66, the Inquiry noted that the acts and practices of small business operators that hold genetic information pose a potential risk to the privacy of both the individual concerned and his or her genetic relatives. The Inquiry expressed the view that small business operators that hold genetic information should be subject to the provisions of the *Privacy Act*, whether or not they provide a health service.¹¹⁴

109 In summary, to qualify for the small business operator exemption, an entity (i) must have an annual turnover of \$3 million or less; (ii) cannot be related to a business with an annual turnover of greater than \$3 million; (iii) must not provide a health service and hold health records; (iv) must not disclose personal information about an individual for a benefit, service or advantage; (v) must not provide a benefit, service or advantage to collect personal information; (vi) cannot be a contracted service provider for a Commonwealth contract (even if the entity is not a party to the contract). See *Privacy Act 1988* (Cth) s 6C–6E.

110 See *Ibid* s 6D(4)(b)–(d).

111 T Smyth, ‘Protecting Human Genetic Information and Its Use’ (2002) 10(6) *Health Law Bulletin* 64, 66.

112 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

113 These provisions of the *Privacy Act* commenced on 21 December 2002.

114 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [7.126].

7.103 This proposal was generally supported by submissions,¹¹⁵ although the Commonwealth Attorney-General's Department questioned whether, in practice, there would be any genetic information held by small businesses that is not governed by the *Privacy Act*.¹¹⁶ The Commonwealth Department of Health and Ageing agreed that small business operators holding genetic information should be subject to the *Privacy Act* and noted that this would be consistent with the AHMAC Draft National Health Privacy Code, which does not exempt small business operators.¹¹⁷

7.104 The Inquiry has concluded that there is sufficient doubt about the coverage of *Privacy Act* to justify amending the Act to make it clear that all small business operators that hold genetic information¹¹⁸ are subject to its provisions.

Recommendation 7–7. The Commonwealth should amend the *Privacy Act* to ensure that all small business operators that hold genetic information are subject to the provisions of the Act. (See also Recommendation 8–2 in relation to genetic samples.)

115 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003. The OFPC submitted that, insofar as they apply to all forms of health information, the exemptions from coverage under the Act presently afforded to employee records and to small business operators should be repealed. However, the OFPC expressed concern that limiting the reform to 'genetic information' would introduce 'unnecessary complexity into the regulatory framework applying to small businesses': Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003. See also Ch 34.

116 That is, given the effect of *Privacy Act 1988* (Cth) s 6D(4)(b)–(d): Commonwealth Attorney-General's Department, *Submission G228*, 12 December 2002.

117 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

118 Or bodily samples from individuals whose identity is apparent or reasonably can be ascertained from the sample. See Ch 8.

8. Privacy of Genetic Samples

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Introduction

8.1 The Terms of Reference for the Inquiry refer to the privacy of ‘human genetic samples and information’. These words distinguish genetic ‘samples’ from information that may be derived from them. Almost all human biological samples can reveal genetic information by DNA analysis.¹ In effect, therefore, the Inquiry must consider the privacy of all bodily samples that may be identified with an individual.²

8.2 This chapter begins by examining the existing coverage of the *Privacy Act 1988* (Cth) (*Privacy Act*) as it relates to genetic samples. The Inquiry has concluded that the Act does not cover genetic samples, even where they are identifiable to an individual, for example, where they have a name or other identifier attached.

1 Provided the samples contain cells with nuclei.

2 Including genetic material derived from samples taken from the bodies of individuals—as in the case of immortal cell lines, to the extent that these cell lines contain the DNA of an identifiable individual.

8.3 The chapter then examines why, in the Inquiry's view, the coverage of the *Privacy Act* should be extended to cover genetic samples. These reasons include the following:

- genetic samples are closely analogous to other sources of personal information that are covered by the *Privacy Act* and should be protected by rules that are consistent with those applying to the genetic information derived from samples;
- there are gaps in the existing framework for protecting the privacy of individuals from whom genetic samples are taken or derived;
- these gaps might be usefully remedied if the National Privacy Principles (NPPs) or a set of similar privacy principles were to apply to genetic samples; and
- no circumstances have been identified in which applying the *Privacy Act* to genetic samples would lead to adverse consequences for existing practices involving the collection and handling of genetic samples.

Does the Privacy Act cover genetic samples?

8.4 A key term in defining the coverage of the *Privacy Act* is 'personal information'. The Information Privacy Principles (IPPs) and NPPs apply when government, private sector organisations and individuals collect, store, use and disclose personal information. Under s 6(1) of the *Privacy Act* 'personal information' is defined as follows:

personal information means information or an opinion (including information or an opinion forming part of a database), whether true or not, and whether recorded in a material form or not, about an individual whose identity is apparent, or can reasonably be ascertained, from the information or opinion.

8.5 The definition of a 'record' is also important to the coverage of the Act. In general, the *Privacy Act* applies only to the handling of personal information in a record.³ Under s 6(1) of the *Privacy Act* a 'record' is relevantly defined as follows:

record means:

- (a) a document; or
- (b) a database (however kept); or
- (c) a photograph or other pictorial representation of a person; ...

8.6 Therefore, the extent to which the *Privacy Act* covers a genetic sample depends on whether:

- the genetic sample may be considered to be 'information';
- the information is held in a 'record'; and

3 See *Privacy Act 1988* (Cth) s 16B (in relation to private sector organisations) and the IPPs (in relation to Commonwealth and ACT government agencies).

- the information is about an individual whose identity is apparent, or can reasonably be ascertained, from the information.

Is a genetic sample ‘information’?

8.7 DP 66 stated that the *Privacy Act* does not cover genetic samples, even where they are identifiable (for example, where they have a name or other identifier attached), because the samples are not themselves ‘information’.⁴

8.8 This interpretation is supported by common law principles of statutory interpretation, which require that general words should be given their plain and ordinary meaning, unless the contrary is shown.⁵ The plain and ordinary meaning of the word ‘information’ is unlikely to extend to a genetic sample, as opposed to the information that is derived by sequencing the DNA that the sample contains.⁶

8.9 The fact that it was thought necessary in New South Wales legislation to expressly refer to ‘body samples’ in the definition of ‘personal information’ also supports the view that samples are not covered by the term ‘information’.⁷

8.10 However, different interpretations have been expressed. For example, the Office of the Victorian Privacy Commissioner has stated that the definition of personal information contained in s 3 of the *Information Privacy Act 2000* (Vic) may be interpreted to mean that the Victorian legislation

will apply to DNA samples that are collected, for example, by an organisation (such as police) who have the means available to them to analyse the sample in order to ascertain a person’s identity. At a simpler level, a body sample stored or able to be linked with a unique identifier of the person from whom it was extracted is also personal information.⁸

4 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [7.54]. The Inquiry noted that the Office of the Federal Privacy Commissioner (OFPC) has taken this interpretation: Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Office of the Federal Privacy Commissioner, *Submission G164*, 27 June 2002.

5 See eg D Pearce and R Geddes, *Statutory Interpretation in Australia* (5th ed, 2001) Butterworths, Sydney, 35–36.

6 In a related vein, it has been suggested that a bodily sample, such as a newborn screening card, is not a ‘document’ for the purposes of the *Freedom of Information Act 1982* (Vic) s 5(1). Therefore, there is no right of access to the card: L Skene, ‘Access to and Ownership of Blood Samples for Genetic Tests: Guthrie Spots’ (1997) 5(2) *Journal of Law and Medicine* 137, 140.

7 *Privacy and Personal Information Protection Act 1998* (NSW) s 4(2).

8 Office of the Victorian Privacy Commissioner, *Submission to the Forensic Procedures Review Committee on its Review of Part 1D of the Crimes Act 1914 (Cth)*, 5 September 2002, 5.

Is a genetic sample a ‘record’?

8.11 The second question is whether a genetic sample is a record. The plain and ordinary meaning of the word ‘record’ would not generally extend to a genetic sample.⁹ Under the *Privacy Act*, a record is relevantly defined to mean a document, database or photograph.

8.12 Further assistance in interpreting the terms ‘record’ and ‘document’ is provided by the *Acts Interpretation Act 1901* (Cth), which provides that:

25. In any Act, unless the contrary intention appears:

document includes:

- (a) any paper or other material on which there is writing;
- (b) any paper or other material on which there are marks, figures, symbols or perforations having a meaning for persons qualified to interpret them; and
- (c) any article or material from which sounds, images or writings are capable of being reproduced with or without the aid of any other article or device.

record includes information stored or recorded by means of a computer.

writing includes any mode of representing or reproducing words, figures, drawings or symbols in a visible form.¹⁰

8.13 As with the term ‘information’, it is difficult to argue that the meaning of the word ‘record’ is capable of extending to a genetic sample.¹¹

Is a genetic sample about an identifiable individual?

8.14 The third issue concerns the circumstances in which a genetic sample may be considered to contain information about an identifiable individual. The *Privacy Act* does not apply to information unless it is ‘about an individual whose identity is apparent, or can reasonably be ascertained, from the information or opinion’.¹² Such a standard, based on reasonableness, means that in some cases it will not be clear

9 A newborn screening card is one category of bodily sample that clearly constitutes a ‘record’. However, a blood spot excised from the card is not ‘information’ and, therefore, is not covered by the NPPs or IPPs.

10 *Acts Interpretation Act 1901* (Cth) s 25.

11 It might be possible to argue that a bodily sample is a document because blood is material from which an image (a DNA profile) may be reproduced eg when Restriction Fragment Length Polymorphism (RFLP) analysis and Southern blotting are used to create an image on X-ray film. However, it appears doubtful that the legislation would support such an extended interpretation. In a related context, it has been suggested that a bodily sample, such as that stored on a newborn screening card, is not a ‘document’ for the purposes of the *Freedom of Information Act 1982* (Vic) s 5(1): L Skene, ‘Access to and Ownership of Blood Samples for Genetic Tests: Guthrie Spots’ (1997) 5(2) *Journal of Law and Medicine* 137, 140.

12 From the definition of ‘personal information’ in *Privacy Act 1988* (Cth) s 6(1). Relevant state and territory information and health privacy legislation contains similar definitions.

whether particular information is identifiable and, therefore, whether or not the IPPs and NPPs apply to how it is handled.¹³

8.15 The concept of de-identification arises in different ways under the NPPs. For example, under NPP 10.3, an organisation may collect health information about an individual for research purposes without consent only where the research cannot be conducted with de-identified information.

8.16 In relation to the conduct of research involving humans, the National Health and Medical Research Council's *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) also makes distinctions between identified, potentially identifiable and de-identified personal information or material.¹⁴ However, under the National Statement, information or material is 'de-identified' only if the process of de-identification is irreversible—for example because the identifiers have been removed permanently or because the data have never been identified.¹⁵

8.17 Some submissions questioned whether it can ever be said that a genetic sample is truly de-identified,¹⁶ at least as long as the individual from whom the sample was taken, or their body, still exists. As collections of genetic samples and information proliferate, the chances that any given genetic sample may be able to be re-identified by matching it with samples held on human genetic databases increases.¹⁷

Genetic information, unlike other health information, is inherently linked to a particular individual. This fact, in combination with computer technology, makes the linkage of genetic information to an identifiable individual always a possibility ... Given these concerns, it is not advisable to completely exempt 'anonymous' genetic information from data protection regimes.¹⁸

13 A particular issue involves the extent to which coded information can be considered to be de-identified. Where an independent person (eg a gene trustee) holds the code and it is not possible for others to ascertain the identity of the individual concerned (without the assistance of the independent person), is the information de-identified so that the *Privacy Act* does not govern it or does it depend on the circumstances and, in particular, the practices or policies of the gene trustee? In this context, the Australian Health Ministers' Advisory Council (AHMAC) National Health Privacy Working Group has stated that privacy protection will depend on whether 'an individual's identity could be reasonably ascertained, in the particular situation in which the information is being collected, used or disclosed': Australian Health Ministers' Advisory Council National Health Privacy Working Group, *National Health Privacy Code (draft) Consultation Paper* (2002), AHMAC, Canberra, 50. See also R Magnusson and C Clarke, 'Data Registers in Respiratory Medicine: A Pilot Evaluating Compliance with Privacy Laws and the National Statement on Ethical Conduct in Research Involving Humans' (2002) 10 *Journal of Law and Medicine* 69, 74–75.

14 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 9.

15 The National Statement states that 'It should be recognised that the term "de-identified" is used frequently, in documents other than this Statement, to refer to sets of data from which only names have been removed. Such data may remain "potentially identifiable"': *Ibid*, 9.

16 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002.

17 N Tonti-Filippini, *Correspondence*, 17 May 2002.

18 T Lemmens and L Austin, 'The Challenges of Regulating the Use of Genetic Information' (2001) 2(3) *Isuma: Canadian Journal of Policy Research* 26, 33–34.

8.18 On the other hand, the Commonwealth Attorney-General's Department observed, in relation to the argument that genetic information is, by its nature, identifiable, that the same could be said of fingerprints, dental records or any other uniquely identifying features of an individual.¹⁹

8.19 The Inquiry does not believe that genetic samples should be considered inherently identifiable for the purposes of the *Privacy Act*. Whether they are *reasonably* identifiable or not will depend on the surrounding context. In most circumstances, an unlabelled and uncoded sample may still be considered to be de-identified, despite a theoretical possibility of re-identification.

Conclusion—Coverage of the Privacy Act

8.20 The existing application of the *Privacy Act* to bodily samples is not entirely clear. However, the Inquiry has concluded that the only well-known category of genetic sample that is clearly covered by the *Privacy Act* is the newborn screening card. Newborn screening cards are made of blotting paper on which spots of blood have been absorbed. The child's name, date of birth, hospital of birth and birth weight are recorded on the card, together with the mother's name and date of birth. It seems clear that newborn screening cards contain personal information in a record and must be handled in accordance with the IPPs or NPPs, as applicable.²⁰ This conclusion does not stem from the fact that a card contains a genetic sample, but from the fact that it contains other personal information about an identifiable individual and the genetic sample forms part of the same documentary record.

8.21 It seems equally clear that genetic samples, in whatever form, that are not labelled with names or other identifiers are not covered by the *Privacy Act*, even in situations where the identity of the individual from whom the sample is derived is reasonably ascertainable by testing. Such samples are not information and will not usually be held in a 'record', for example, where they are stored as microscope slides.

8.22 The position with regard to labelled samples is more complex. In some cases, while the sample itself may not be covered by the *Privacy Act*, the information attached to it (that is, the label) may be, and must be handled in accordance with information privacy principles.

8.23 Many, if not most, bodily samples held by organisations in Australia are labelled with names or other identifiers, such as alpha-numeric codes. This appears to be the case with most samples held by pathology laboratories. Codes are used so that laboratory technicians and others may handle samples without becoming aware of the identity of individuals. DP 66 proceeded on the basis that a name or a code by itself is not 'information about an individual'. This view is consistent with opinions expressed by the Attorney-General's Department and the Australian Government Solicitor in

19 Commonwealth Attorney-General's Department, *Submission G158*, 7 May 2002. The Department submitted that 'to be identifiable, the information must usually be accompanied by other identifying information'.

20 However, a blood spot excised from the card is not information and, therefore, is not covered by the NPPs or IPPs.

relation to the identical definition of ‘personal information’ in the *Freedom of Information Act 1982* (Cth).²¹

8.24 Personal information that may be derived indirectly from the context in which the identifier is found—for example, that the named individual has had blood taken from them, which is now stored in a laboratory that tests for certain genetic disorders—is not covered by the *Privacy Act*. This is because, while personal information need not be recorded in a material form, it must be in a ‘record’ to be governed by the IPPs or NPPs,²² and not be merely a matter of inference.

8.25 However, the legal position is not clear. An alternative view is that a name, by itself, may constitute information about an individual and therefore, in effect, labelled samples always have to be handled in accordance with the IPPs or NPPs.²³ Another view is that the application of the Act in this regard may depend on the exact circumstances of the case, for example, on whether the name on a labelled sample is a common name or not. As with DP 66, this Report proceeds on the basis that a name or a coded name by itself is not ‘information about an individual’. The implications for the Inquiry’s recommended reforms, if this approach is not correct, are discussed at the conclusion of this chapter.

8.26 Different approaches have been taken to the coverage of samples in state and territory information and health privacy legislation. The *Privacy and Personal Information Protection Act 1998* (NSW) and *Health Records and Information Privacy Act 2002* (NSW) define personal information to include ‘such things as an individual’s fingerprints, retina prints, body samples or genetic characteristics’.²⁴ Individually identifiable genetic samples are therefore covered by New South Wales privacy legislation.²⁵ Other state and territory legislation contains definitions of personal information and health information which, in relevant respects, more closely follow the wording contained in the federal *Privacy Act*. Genetic samples are generally not covered by such legislation.²⁶

21 Commonwealth Attorney-General’s Department, *Submission G315*, 19 February 2003. See Attorney-General’s Department — Security Law and Justice Division, *Freedom of Information (FOI) Memorandum No.98: Exemptions Sections in the FOI Act*, Attorney-General’s Department, <www.ag.gov.au/www/securitylawHome.nsf/Web+Pages/F6D7D4157FE35630CA256C17000AA2BD?OpenDocument>, 24 February 2003.

22 See *Privacy Act 1988* (Cth) s 16B (in relation to private sector organisations) and the IPPs (in relation to Commonwealth and ACT government agencies).

23 Yet another possible interpretation is that the labelled sample in its entirety should be considered as a ‘record’. However, this interpretation, by extension would mean that any thing may be transformed into a record covered by the *Privacy Act* simply by labelling it (notwithstanding that it is not a document, database or photograph) eg clothes or other personal possessions.

24 *Privacy and Personal Information Protection Act 1998* (NSW) s 4(2); *Health Records and Information Privacy Act 2002* (NSW) s 5(2).

25 Privacy NSW noted that *Privacy and Personal Information Protection Act 1998* (NSW) s 4(2) was inserted ‘because the former Privacy Committee was acutely aware of concerns regarding a number of issues involving bodily samples in the NSW context, for instance, the non-consensual access to and disclosure of newborn screening cards for forensic testing and law enforcement purposes’: Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002.

26 See *Health Records Act 2001* (Vic) s 3 ‘health information’; ‘personal information’; *Information Privacy Act 2000* (Vic) s 3 ‘personal information’; *Health Records (Privacy and Access) Act 1997* (ACT) s 4 ‘personal health information’; ‘personal information’.

The analogy between genetic samples and information

8.27 DP 66 suggested that genetic samples are closely analogous to other immediate sources of information that are protected by information privacy principles.²⁷

8.28 DNA is often popularly referred to as a ‘genetic code’ and the genome as a ‘book’. The four bases of DNA (A-G-C-T)²⁸ are sometimes called the ‘genetic alphabet’. Genetic science itself is replete with the language of information and information technology—for example, there are bases, codons, messenger RNA, transcription and translation.²⁹

The idea of the genome as a book is not, strictly speaking, even a metaphor. It is literally true. A book is a piece of digital information, written in linear, one-dimensional and one-directional form and defined by a code that transliterates a small alphabet of signs into a large lexicon of meanings through the order of their groupings. So is a genome.³⁰

8.29 One view expressed to the Inquiry was that a distinction needs to be drawn between privacy protection of personal information and the sources of that information.³¹ A basis for this distinction might be that, unlike a book or other written information, technology must intervene to create genetic information from a genetic sample. However, if a book exists in electronic form, technology will also be required to intervene. Computerised information, whether on a hard drive, a CD-ROM or some other format, requires technological intervention before information may be derived from the bytes recorded. Yet there is no question that personal data on an encrypted CD-ROM is considered to be ‘information’ in a ‘record’ for the purposes of the *Privacy Act*. Modern genetic sequencing technology may make genetic samples as immediate a source of information as, for example, a computer disk or database, which are already covered by the *Privacy Act*.

Gaps in existing privacy protection

8.30 The Inquiry has identified gaps in the existing framework for protecting the privacy of the individuals from whom genetic samples are taken or derived, including gaps that might be remedied if information privacy principles were to be applied to bodily samples.³² Examples of these gaps are outlined below, with particular reference to the NPPs.

27 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [7.58]–[7.67].

28 Adenine; guanine; thymine; cytosine.

29 See the genetics primer in Ch 2.

30 M Ridley, *Genome: The Autobiography of a Species in 23 Chapters* (1999) Fourth Estate, London, 7–8.

31 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

32 For a more detailed examination of these gaps in privacy regulation and the possible benefits of new regulation see Australian Law Reform Commission, *Working Paper: Applying Privacy Principles to Genetic Samples* (2002) ALRC.

8.31 Information privacy principles, such as the NPPs, ensure that organisations handle personal information responsibly and give people some control over the way in which it is handled.³³ In the context of health services, the NPPs promote greater openness between health service providers and consumers regarding the handling of health information.³⁴ The Inquiry has concluded that a similar set of principles should apply to the handling of identifiable bodily samples. Samples, as well as the information derived from them, should be handled fairly, responsibly and with adequate recognition of the expectations of the individuals concerned.

8.32 In many circumstances, genetic samples and the information derived from them travel in tandem—the samples are used, transferred, and stored for the same purposes as the information derived from them. Therefore, it might reasonably be expected that consistent rules should apply to both, unless there is some compelling reason for divergence.

8.33 For example, if personal information derived from the analysis of a bodily sample may not be used for a particular purpose, then it might be expected that the transfer of the sample itself to an organisation that intends to use it for that purpose should not be permitted. However, as discussed below, this is not always the case.

Collection of genetic samples

8.34 The core obligations contained in the NPPs relating to collection of genetic information³⁵ include obligations to collect this information only with consent (except in specified circumstances) and to inform individuals, at the time of collection, about how their information is to be handled.³⁶

8.35 At present, no similar obligations apply to the collection of genetic samples. While aspects of these obligations sometimes arise under other laws, protection is not comprehensive. In particular, existing laws do not require the individual to be fully informed about the purposes for which the samples are being collected or the organisations or persons to whom they are transferred. For example, the general law relating to battery and consent to medical treatment requires consent to the physical taking of samples. However, this does not require the consent to be informed by information about the purposes for which the sample may be used, or the persons to whom the samples may be transferred. While health service providers may in practice inform patients about these matters,³⁷ there is no legal obligation to do so.

33 Office of the Federal Privacy Commissioner, *Guidelines to the National Privacy Principles* (2001), OFPC, Sydney, 2.

34 For example, by providing a general right of access for consumers to their own health records, and requiring health service providers to have available documentation that clearly sets out their policies for the management of personal information: Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, Foreword.

35 That is, health information as defined in *Privacy Act 1988* (Cth) s 6(1).

36 *Ibid*, NPP 1, NPP 10.

37 The Inquiry understands that most hospital consent forms include advice that bodily samples taken during treatment are retained by the hospital and may be used in research.

8.36 Where the samples are taken primarily for research, the Human Tissue Acts³⁸ require that consent be given to that use. However, these requirements do not apply where the samples are taken in the course of medical treatment³⁹ and in any case relate only to consent to the removal of the tissue, rather than to its future storage, use or transfer. The Human Tissue Acts do not require the donor to be informed about what will be done with the sample.

8.37 The Centre for Law and Genetics, in supporting the extension of the *Privacy Act* to genetic samples, stated:

It is appropriate that the sample collector should be under an obligation to explain the purpose of collection, primary and related secondary uses, the persons to whom the samples are usually transferred, access rights etc at the time of collection of the sample. This is consistent with the obligations of researchers to explain *future uses* of genetic information when conducting human genetic research. If these matters are properly explained to sample providers, growing concerns about the use of samples may well be alleviated. ... In the existing climate, with increasing concerns about personal privacy and increasing capacity to extract personal information from genetic samples, it makes good sense that sample providers should be told about what happens to their samples after removal. In many instances, all that may be required is for the sample provider to be notified that their sample will be stored for a particular period and then destroyed.⁴⁰

Example A. *A private hospital collects blood samples from its patients for clinical purposes. Part of the sample is sent to the pathology laboratory for analysis. Without the knowledge or consent of the patients, part of the sample, labelled only with the name of the patient from whom the blood has been taken, is sent to the hospital's research institute for storage in a tissue bank.*

The current law does not proscribe the collection of samples in these circumstances. The *Privacy Act* does not apply to the collection of the sample, although it does apply to the collection of any information associated with the sample.

The hospital has no obligation under NPP 1 to inform the patient about the purposes for which the sample is collected or the fact that the hospital transfers samples to the research institute. Further, because the sample is collected in the course of medical treatment, the consent requirements of the Human Tissue Acts do not apply.⁴¹ This gap in privacy protection is remedied only once the research institute analyses the sample (collecting health information) and uses this information.

38 *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1979* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1985* (Tas); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplant Act 1982* (WA); *Transplantation and Anatomy Act 1978* (ACT); *Human Tissue Transplant Act 1979* (NT).

39 That is, the obligation to obtain consent to the use of the sample for 'medical purposes or scientific purposes'. See eg *Human Tissue Act 1983* (NSW) ss 7, 34.

40 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

Transfer of genetic samples

8.38 The core obligation contained in NPP 2 is that organisations can disclose personal information only for the primary purpose for which it was collected or for directly related secondary purposes. At present, no similar obligation applies to the disclosure (that is, ‘transfer’)⁴² of genetic samples.

8.39 This has significant consequences. While the *Privacy Act* prohibits organisations from disclosing health information derived from a genetic sample without the individual’s consent (other than for the primary purpose of collection), no similar privacy protection attaches to the sample itself. There may be professional, regulatory, contractual or other consequences for the organisation transferring the samples,⁴³ but there is no general legal obligation not to transfer possession (or sell) a sample without the consent of the individual from whom the sample comes.

Example B. *A public health authority holds newborn screening cards. An officer of the authority is approached by a private investigator who wishes to obtain a sample in order to establish paternity in a family dispute. The officer punches out a section of the blood spot and provides it to the private investigator.*

Newborn screening cards are clearly covered by the *Privacy Act* because they are ‘personal information’—the cards contain identifying information, such as date of birth, as well as the blood sample, and the card is a ‘record’ for the purposes of the Act.⁴⁴ Disclosure of the card itself in these circumstances would breach the Act. However, once a section of the blood spot is punched out and physically detached from the personal information and the record on which it was stored, the *Privacy Act* no longer governs how the genetic sample is dealt with and it may be transferred without breaching the Act.

8.40 The *Privacy Act* sets out specific obligations that apply when an organisation transfers personal information outside Australia. Briefly, NPP 9 prohibits the transfer of personal information unless the recipient of the information is subject to a law, binding scheme or contract that upholds principles substantially similar to the NPPs, or the organisation has taken reasonable steps to ensure that the information will not be dealt with inconsistently with the NPPs.

41 That is, the obligation to obtain consent to the use of the sample for ‘medical purposes or scientific purposes’. See eg *Human Tissue Act 1983* (NSW) ss 7, 34.

42 *Privacy Act 1988* (Cth) NPP 2 refers to the ‘disclosure’ of personal information. While it may be possible to interpret the passing of possession of genetic samples from one organisation to another as a ‘disclosure’, the term ‘transfer’ seems more apt.

43 For example, for a pathology laboratory, these consequences could include liability for breach of a contractual undertaking or fiduciary duty, loss of National Association of Testing Authorities Australia accreditation and eligibility to obtain Medicare fees for medical services, penalties for sale of human tissue under the Human Tissue Acts, and disciplinary action by health registration authorities.

44 *Privacy Act 1988* (Cth) s 6(1).

8.41 It is common for Australian pharmaceutical companies to send genetic samples overseas for analysis in conducting clinical trials. Research groups also conduct collaborative projects and transfer samples to scientists overseas. Applying NPP 9 to this situation would permit the export of genetic samples to continue, but subject to safeguards.⁴⁵ This would be consistent with the National Statement, which requires that research conducted overseas under the aegis of an Australian institution must comply with the provisions of the National Statement.⁴⁶

8.42 The Commonwealth Department of Health and Ageing supported a nationally consistent approach to regulating the export of genetic samples and agreed that NPP 9 was an appropriate model.⁴⁷ Other submissions supported the use of NPP 9, as the starting point for regulating the export of genetic samples⁴⁸—although some submissions suggested that the provisions of NPP 9 may need strengthening to provide adequate protection for genetic samples.⁴⁹

Example C. *Without the consent of the individuals concerned, an Australian pharmaceutical company (C Ltd) conducting a clinical trial sends genetic samples for analysis by D Ltd in an overseas jurisdiction (E). C Ltd also sends associated personal information about the individuals from whom the samples come. Country E has no law restraining the secondary use or transfer of the personal information or genetic samples. There are no contractual terms dealing with this matter in the contract between C Ltd and D Ltd.*

At present the *Privacy Act* would prohibit the transfer of the information but not the transfer of the samples themselves, even if the samples were identifiable to the individuals concerned. Applying NPP 9 to samples would mean that similar protection applies to the transborder transfer of both information and samples.

45 And subject to other relevant legislation—eg the import or export of a ‘human embryo clone’ is an offence under s 11 of the *Prohibition of Human Cloning Act 2002* (Cth).

46 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [1.21].

47 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

48 Centre for Genetics Education, *Submission G232*, 18 December 2002; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Peter MacCallum Cancer Institute Tissue Bank, *Submission G263*, 20 December 2002; Department of Human Services Victoria - Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002.

49 Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Department of Human Services Victoria — Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002. Privacy NSW recommended that it should be a precautionary practice to de-identify genetic samples and genetic information intended for export for research or other purposes: Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002.

Access to genetic samples

8.43 NPP 6 of the *Privacy Act* states that, subject to some exceptions, if an organisation holds personal information about an individual, it must provide the individual with access to the information on request. There is no similar right to obtain access to a genetic sample.

8.44 It may be argued that extending rights of access to genetic samples serves no useful purpose because an individual always has access to a new sample because a new sample may always be taken from their body. However, some genetic samples, such as samples of cancerous tissue, may not be substitutable by new samples.

8.45 Individuals may have a legitimate need to obtain access to their own genetic samples or those of a genetic relative (see below). The reasons for such a request might include an intention to transfer the samples to a new medical practitioner for re-testing or to arrange new testing of the samples for health care purposes.

Example D. *An oncologist holds medical records concerning the diagnosis and treatment of patient D. The oncologist also holds a sample of cancerous tissue surgically excised from D in the course of her treatment. D requests access to her medical records and to the excised tissue in order to transfer them to her new specialist so that the tissue can be re-tested.*

NPP 6 of the *Privacy Act* provides a legally enforceable right for patients to obtain access to their medical records held by private medical practitioners. In this example, the oncologist is required to provide access to the medical records and, if D requests a copy of the original records, these will ordinarily have to be provided by the oncologist.⁵⁰ However, there is no similar right of access to the tissue itself.

8.46 It is problematic to apply NPP 6 to genetic samples held by medical practitioners or pathology laboratories. The content of the right of access would have to make it clear that it involves only a right to obtain access to part of the sample. It should not imply a right to require that a sample be re-analysed, de-identified or destroyed. Further exceptions to the right of access would need to be developed so that access may be refused where:

- it is not physically possible to provide part of a sample;
- providing part of a sample means that the remaining portion is insufficient for the purposes of the organisation retaining it; or

⁵⁰ See eg Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 32.

- releasing a sample to an individual raises public health concerns.⁵¹

8.47 The Inquiry's view is that the right of access to genetic samples should be exercisable only where the samples are needed in connection with the present or future provision of medical testing, diagnosis or treatment to the individual seeking access. It is reasonable to expect that, in most cases, individuals seeking access will do so for such purposes. In the Inquiry's view, no other purposes for seeking access are sufficiently compelling to justify a right of access under the *Privacy Act*.

Rights of genetic relatives

8.48 The Inquiry also recommends that the right of access to an individual's bodily samples should extend to his or her 'genetic relatives'. This term will require further definition, limiting it to siblings, parents or children (first-degree genetic relatives).⁵² The Inquiry understands that there are cases in which access to samples taken from genetic relatives may be important, especially in relation to the detection and diagnosis of familial cancers.⁵³ In most of these cases, the individual from whom the sample is derived is deceased.⁵⁴

8.49 First-degree genetic relatives' rights of access should also be limited to access for medical testing, diagnosis or treatment purposes,⁵⁵ and to circumstances where access is necessary to lessen or prevent a serious threat to their life, health, or safety, even where the threat is not imminent.⁵⁶

8.50 When an organisation receives a request from a first-degree genetic relative for access to an individual's bodily sample, the organisation should be obliged, unless it is impracticable, to first ask the individual from whom the sample comes whether he or she consents to the organisation providing access. Granting access without the

51 Some bodily samples may be infected or be likely to decompose if improperly stored. Therefore, the right to access may need to be drafted to ensure samples must be transferred to an appropriate intermediary such as a pathology laboratory, medical practitioner or hospital, rather than to the individual who has sought access. As is the case with information under NPP 6, an access principle relating to samples should allow for release to a mutually agreed intermediary. Generally access to a sample will be sought because the individual wishes to have the sample analysed or tested. Therefore, release of the sample to an intermediary should in most circumstances meet the needs of the individual seeking access.

52 Professor Bob Williamson has noted that 'Only blood relatives have a direct interest. The further you go, the lower the "yield". First degree relatives (parent, child, sib) have a 50% chance of sharing genes. As you move to grand-children, or first cousins, this goes down to 25% or less. Once you are at second cousins or more removed, the chances are really little different from those in the general population for many diseases. But of course for high penetrance dominant disorders, the key thing is that a test can often be offered to anyone on a linear path to the person affected, but it is only of value if the "intermediate" person has the mutation': B Williamson, *Correspondence*, 27 September 2002.

53 One estimate is that there would be around 100 cases each year in Victoria where access to familial samples might be clinically required: Ibid. Where a disease is caused by a known mutation such as Huntington's disease and cystic fibrosis there is no need for access to samples from genetic relatives. The DNA of the person who is being diagnosed is sufficient.

54 B Williamson, *Correspondence*, 28 February 2003.

55 For example, where access is 'likely to give relevant information of his or her risk of a disease, assuming that risk is raised significantly above that of the general population, or if the age of onset is earlier, or the course of the disease is more severe': B Williamson, *Correspondence*, 27 September 2002.

56 This formulation is consistent with the Inquiry's recommendations in relation to access to genetic information by first-degree genetic relatives: See Ch 21.

consent of a living individual from whom the sample has been taken should only occur in exceptional circumstances.

8.51 When seeking consent for access, the organisation should tell the individual from whom the sample comes that the sample will be handled only by health professionals and that, in most cases, the only information given to genetic relatives will be familial genetic information. In general, no details of the individual's own genetic status will be given to genetic relatives without the individual's consent.⁵⁷

8.52 Where the individual does not give consent, the organisation should be able to refuse access if providing access would have an unreasonable impact upon the privacy of the individual from whom the sample comes.

8.53 The Inquiry has recommended that the *Privacy Act* should protect information about (and samples from) deceased individuals (see Recommendation 7–6). However, an important factor in determining whether providing access would have an unreasonable impact upon the privacy of any individual is whether or not the individual from whom the sample comes is deceased. Where the sample is from a deceased individual, the privacy impact of providing access is likely to be minor.

8.54 Access to the sample should be available only through a nominated medical practitioner who is in a position to arrange testing of the samples for health care purposes and to handle the sample in an appropriate way.

Extending the Privacy Act to fill the gaps

Benefits of extending the Act

8.55 There are some clear benefits in applying information privacy principles to the handling of genetic samples. In addition to those highlighted above, organisations would have to comply with legally enforceable standards for the physical security of holdings of genetic samples and would not be permitted to retain samples without a clearly defined purpose.⁵⁸ Genetic samples would not be able to be sent outside Australia unless reasonable steps were taken to ensure that the privacy of the samples is adequately protected by the recipient in the overseas jurisdiction.⁵⁹

8.56 The need to comply with privacy principles would have the salutary effect of requiring organisations to articulate their policies with regard to the use, transfer and storage of genetic samples.⁶⁰ There are benefits in promoting more openness about, and public understanding of, the ways in which samples are dealt with. Openness and accountability may reduce the need for other sector-specific regulation—including, for example, the licensing or registration of human genetic research databases.⁶¹

57 Although in some cases, this status may be able to be inferred.

58 See *Privacy Act 1988* (Cth) NPP 4.

59 See *Ibid*, NPP 9.

60 See *Ibid*, NPP 5.

61 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 15–1.

8.57 It may be argued that the benefits of such reform may be limited because, as soon as information is derived from samples, the protection of the *Privacy Act* will immediately apply in any case.⁶² However, regulating the genetic samples brings regulation closer to the point of collection and may make it more effective in practice. The Centre for Law and Genetics stated that extending privacy protection to samples

will ensure that all of the people who come into possession of genetic samples are bound by privacy obligations, irrespective of whether or not they, themselves, extract genetic information. This will improve the capacity to keep track of the use and transfer of genetic samples from the source to the end user of genetic information.⁶³

8.58 If a genetic sample is passed through a chain of hands, the organisation that ultimately uses it may breach the *Privacy Act*, but the individual from whom the sample comes is less likely to be aware of the offending use or to be able to enforce his or her rights under the Act. Further, the ultimate user may be an organisation or individual not covered by the Act.⁶⁴

8.59 As noted above, samples and associated information often travel in tandem, but there may be situations in which they come into different hands. For example, when a sample is sent for analysis to a pathology laboratory it may be accompanied by information about the gender and Medicare number of the patient, the conditions to be tested for, the name of the treating physician and so on. At the laboratory, the sample may be separated from this information and have only an identifier (a name or code) attached. The sample will not be covered by the Act, yet the potential exists for the sample to become re-associated with the medical records or other personal information of the patient at some future time.⁶⁵

8.60 Extending the *Privacy Act* to cover samples also has the advantage of using an existing and well-developed regulatory framework, under the oversight of the Office of the Federal Privacy Commissioner (OFPC). This regulatory framework includes mechanisms for complaint investigation, conciliation and determination, the approval of industry privacy codes, the publication of privacy guidelines and the making of public interest determinations.

8.61 Privacy protection for samples may also best be achieved by building on concepts that are already becoming familiar to health professionals and others involved in handling identifiable bodily samples. The culture of privacy compliance is well-established in the professional groups most involved with the handling of bodily samples. For example, respect for the privacy of persons and information is well

62 Assuming the organisation concerned is covered by the *Privacy Act*.

63 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

64 For example, because the user is an individual testing the sample 'other than in the course of a business': *Privacy Act 1988* (Cth) s 7B(1). However, testing a sample without consent might nevertheless constitute a breach of the new criminal offence proposed by the Inquiry: see Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 5-4, 5-5.

65 As discussed earlier in this Report, if a name or other identifier, by itself, can constitute information about an individual in terms of the *Privacy Act*, the effective gaps in the privacy protection of genetic samples may be more limited than indicated above.

understood as an underpinning of ethical conduct in medical research, notably as expressed in the National Statement, which governs how researchers deal with both genetic samples and genetic information in research.

8.62 While it would be possible to draft an appropriate set of minimum privacy standards for the fair handling of bodily samples from scratch, the framework provided by existing information privacy principles is a logical place to start.

Other jurisdictions

8.63 With the exception of New South Wales,⁶⁶ no other Australian jurisdiction applies information privacy principles explicitly to bodily samples. DP 66 noted that, while the New South Wales legislation has been in operation since 1 July 2000, the coverage of bodily samples has not led to noticeable controversy.

8.64 The idea of applying information privacy protection to genetic samples is beginning to attract attention in other jurisdictions, including in the United States and the United Kingdom. In the United States, an April 2002 report prepared by the Health Privacy Project, Georgetown University, noted that the *Health Insurance Portability and Accountability Act 1996* (US) ‘does not protect tissue, blood, or any other bodily source of a person’s genetic information’—despite the fact that samples are relatively easy to obtain. The report concluded that ‘genetic source materials’ need privacy protection.⁶⁷

8.65 In the United Kingdom, a November 2002 report by William Lowrance for the Nuffield Trust considered whether genetic materials should be considered as ‘personal data’. The report noted that an analogy often suggested is with fingerprints, which are treated as personal data under most data protection (information privacy) regimes.⁶⁸ However, the report concluded that:

Medical specimens containing DNA and linked to personal identifiers probably should not be considered personal data just because they contain DNA, but they should be held in medical confidentiality as is customary.⁶⁹

Reaction to the reform proposal

Support for reform

8.66 The proposal to extend the coverage of the *Privacy Act* to identifiable genetic samples received broad support in submissions and consultations.⁷⁰ The Centre for Law and Genetics summarised the position as follows:

66 *Privacy and Personal Information Protection Act 1998* (NSW); *Health Records and Information Privacy Act 2002* (NSW).

67 J Hustead, A Cunningham and J Goldman, *Genetics and Privacy: A Patchwork of Protections* (2002), California HealthCare Foundation, Oakland, 27.

68 W Lowrance, *Learning from Experience: Privacy and the Secondary Use of Data in Health Research* (2002), The Nuffield Trust, London, 34.

69 *Ibid.*, 35.

There is some academic support for the view that genetic samples are information, or, more likely, that they are records containing information. On this basis, samples would be interchangeable with the information they contain. Technologies such as bioinformatics are firmly establishing the linkage between computer technology and genetic technology and it is likely that human tissue samples will, over time, be seen increasingly as living databases of information. However, this stage has not yet been reached and the argument remains speculative. There is sufficient uncertainty about this argument at the present time to justify clarification through amendment of [the *Privacy Act*].⁷¹

8.67 The New South Wales Health Department stated that:

We are in an age where health information is stored and accessed using technological means such as electronic health records, or encrypted data from which information is potentially derivable. It is therefore appropriate to consider an identifiable sample similarly, ie, not only as the source of genetic information but also as the information itself since it is technologically accessible.⁷²

8.68 The Office of the Victorian Privacy Commissioner stated that:

In the present—and particularly in the likely future—a bodily sample is personal information and should be so defined. While a bodily sample and the genetic data derived from it will almost always be health information, it will also be personal information with relevance to matters other than health. Proper privacy protection in relation to genetic information should not fail for any individual through the narrowness of a definition.⁷³

8.69 Similarly, the Office of the Health Services Commissioner Victoria expressed the opinion that ‘samples are health information and this should be made clear in legislation’.⁷⁴ The Australian Privacy Charter Council agreed that the *Privacy Act* should ensure that bodily samples from which identity can be ascertained are covered by the terms ‘personal information’ and ‘record’ and noted

70 WA Research Tissue Network, *Consultation*, Perth, 28 October 2002; Health Consumers' Council, *Submission G174*, 18 September 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; A Gesche, *Submission G264*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003: The proposal was opposed by some: Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Queensland Government, *Submission G274*, 18 December 2002. The Department of Human Services, South Australia expressed concern about the level of protection that would be provided by the *Privacy Act*, given that the Act does not ‘cover the deceased, de-identified samples and the fact that genetic information is always inherently identifiable’: Department of Human Services South Australia, *Submission G288*, 23 December 2002.

71 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

72 NSW Health Department, *Submission G303*, 13 January 2003.

73 Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002.

74 Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

[w]e cannot see any particular difficulties in relation to the complaints and enforcement aspects of the Privacy Act ... , or with the relationship with other laws dealing with bodily samples ...⁷⁵

8.70 However, other submissions expressed a range of reservations about the efficacy of the proposed reforms, possible duplication or complexity resulting from the interplay between an amended *Privacy Act* and other laws dealing with bodily privacy or the handling of body parts, and possible adverse consequences for existing practices involving the collection and handling of genetic samples.⁷⁶ These issues are addressed below.

Efficacy of reform

8.71 The OFPC raised concerns about whether the inclusion of ‘bodily samples’ within the definition of ‘personal information’ and ‘record’ in the Act is justified by the genetic properties of the sample, and whether this is an example of unwarranted genetic exceptionalism.⁷⁷ The OFPC also questioned whether, given that use and disclosure of information derived from samples is covered by the Act, there is any need to bring all bodily samples within its coverage:

Since almost any body sample with cellular properties contains all of an individual’s genes, a body sample, however large or small, can be a ‘genetic sample’. If that bodily or ‘genetic sample’ is collected and perhaps stored in a refrigerator, the Act does not apply. At the moment any information, whether genetic or otherwise, contained in that bodily sample is derived from the sample, all ‘uses’ and ‘applications’ involving that information are covered by the Act. These include any subsequent uses or disclosures of that information. The reasoning which supports bringing *all* bodily samples within the coverage of the Act to protect the collection, and possibly the storage, of bodily samples may need to be re-visited.⁷⁸

8.72 However as discussed above, the potential regulatory benefits of reform are not limited to those relating to the collection and storage of samples, but extend to the transfer of samples (including to overseas jurisdictions), the promotion of transparency in organisational practices, and individual rights of access to samples.

8.73 Importantly, the Inquiry has not been able to identify any circumstances in which applying the principles contained in the *Privacy Act* to samples would lead to clearly undesirable consequences for legitimate existing practices, nor have such circumstances been clearly identified in submissions or consultations.⁷⁹

75 Australian Privacy Charter Council, *Submission G304*, 21 January 2003.

76 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Office of the Federal Privacy Commissioner, *Submission G164*, 27 June 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Queensland Government, *Submission G274*, 18 December 2002.

77 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

78 *Ibid.* Other concerns raised by the OFPC included the need for detailed consideration of the possible retrospectivity of amendments to the Act to cover all bodily samples, possible adverse consequences for research and ethics review of research involving bodily samples and the impact of OFPC compliance functions.

79 For a detailed examination of the implications of the proposed reform for existing practices see: Australian Law Reform Commission, *Working Paper: Applying Privacy Principles to Genetic Samples* (2002) ALRC.

Information and bodily privacy

8.74 In DP 66, the Inquiry recognised that the proposed reform would represent a fundamental change to the coverage of the *Privacy Act*, extending it beyond information privacy as currently conceived.⁸⁰ The OFPC suggested that problems may arise from blurring the distinction between ‘information privacy’ and ‘bodily privacy’, to the detriment of regulatory efficacy and public understanding of genetics.⁸¹

8.75 In general, the views of the OFPC were not shared by other privacy regulators. For example, Privacy NSW submitted that privacy legislation need not be confined to the protection of personal information held in records:

With appropriate legislative reform, privacy laws could be further developed to provide the necessary legal protection for bodily privacy. Conversely, an attempt to maintain a clear demarcation between different types of privacy protection may be problematic in light of new technologies which involve the merging of biology, mathematics and computer science, namely, biometrics and bioinformatics. Such developments give rise to new forms of body templates or records which further blur the distinction between personal information and its source in individual humans, rendering the concepts of information privacy and bodily privacy inherently inter-related.⁸²

8.76 It is not the intention of the Inquiry to reform existing laws dealing with the protection of the individual’s person and their body parts. These existing laws include those dealing with physical violence and assault, the ownership or theft of body parts,⁸³ and controls over the transplantation of organs or the donation of human tissue. The focus of the present recommendations remains on information privacy and on protecting genetic samples as an immediate source of personal information.

8.77 The Inquiry recognises that, as a consequence of its recommendations, aspects of bodily privacy may be imported into the *Privacy Act*. There are contexts in which human bodies or body parts are dealt with that have nothing to do with their potential as sources of genetic information. For example, after death bodies must be buried, interred or cremated. Bodies and parts of bodies may be subject to post-mortem examination, autopsy or coronial procedures. Body parts from living or dead individuals may be used in transplantation.

8.78 It is also possible to envisage circumstances in which a body part, from a living or dead individual, is preserved as a memento, or displayed as a curiosity or relic. Regulating the ways in which bodily parts may be used or transferred may be seen as extending the *Privacy Act* to protect against affronts to human dignity.

80 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [7.84].

81 Office of the Federal Privacy Commissioner, *Submission G164*, 27 June 2002.

82 Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002. Privacy Victoria stated that ‘a bodily sample is personal information and should be so defined’: Office of the Victorian Privacy Commissioner, *Submission G266*, 20 December 2002.

83 The possible use of property rights in genetic material as a means of protecting genetic privacy is discussed (and rejected) in Ch 20.

8.79 If the Inquiry's recommendations are implemented, the handling of bodies and body parts in these contexts would be regulated by the *Privacy Act*, as well as by existing legislation that regulates specific aspects of the handling of bodies and body parts. These other laws include the Human Tissue Acts,⁸⁴ and legislation relating to coronial procedures and the use of bodies in anatomy teaching and research.⁸⁵

Overlapping law

8.80 The Inquiry does not consider that this overlap would necessarily produce problems in practice. The additional legislation referred to above deals with the handling of bodies or body parts in specific contexts. In the language of the *Privacy Act*, these practices are 'required or authorised by or under law',⁸⁶ and may be taken into account in applying the NPPs.

8.81 Privacy NSW noted that privacy legislation often overlaps with other more specific forms of regulation. Privacy NSW was 'not aware of any major or insuperable legislative inconsistencies which would arise if the *Privacy Act* were to be amended as proposed'.⁸⁷

8.82 The baseline standards provided by the *Privacy Act* can co-exist with more specific obligations with regard to the handling of bodily samples. For example, a New South Wales Department of Health report into the retention of tissue and organs following post-mortems has recommended that the *Human Tissue Act 1993* (NSW) and the *Anatomy Act 1977* (NSW) be amended to require all collections to be centrally registered and specific standards adopted for cataloguing, identification and recording of consent to retain samples.⁸⁸ Such requirements would not conflict, but augment, the application of information privacy principles to bodily samples.

8.83 If coronial authorities transfer the possession of a body or body part to another organisation (without consent of the executor) this would constitute a criminal offence under coronial legislation.⁸⁹ Similarly, the collection of bodily samples for research or transplantation without consent is an offence under the Human Tissue Acts.⁹⁰ It is no objection that the same conduct might also give rise to a complaint of interference with privacy, if the NPPs were to be applied to bodily samples.

84 *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1979* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1985* (Tas); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplant Act 1982* (WA); *Transplantation and Anatomy Act 1978* (ACT); *Human Tissue Transplant Act 1979* (NT).

85 For example, in NSW the *Human Tissue Act 1983* (NSW); *Coroners Act 1980* (NSW); *Anatomy Act 1977* (NSW).

86 *Privacy Act 1988* (Cth) NPP 2.1(g).

87 Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002. Privacy NSW suggested that it may be necessary to clarify the respective application of privacy law and the Human Tissue Acts and to review the savings clause in *Privacy Act 1988* (Cth) s 3 to ensure that NSW legislation remains valid and applicable in respect of the coverage of identifiable bodily samples.

88 NSW Department of Health, *Interim Report into the Retention of Tissue and Organs Following Post-mortems in NSW* (2001), NSW Department of Health, Sydney, 12.

89 *Coroners Act 1980* (NSW) s 53A.

90 For example, *Human Tissue Act 1983* (NSW) s 36.

8.84 There may also be rare circumstances in which an extended *Privacy Act* would provide a remedy where no existing remedy exists—for example where an individual’s identifiable body part is put on display without consent (a modern manifestation of the ‘Buddha’s finger’ phenomenon).⁹¹

8.85 If it were considered that *Privacy Act* coverage of bodily samples was undesirable in principle or for practical reasons—for example, because of the number and nature of complaints that might have to be dealt with by the OFPC—it would be possible to exclude certain matters from the ambit of the amended Act. For example, the handling of bodies in the course of post-mortem or coronial procedures or of body parts that have been surgically removed might be excluded from the application of the Act.

8.86 In particular, the Inquiry has concluded that the provisions of the NPPs, recast as proposed, appear capable of operating in harmony with the Human Tissue Acts.⁹² In this context, the Centre for Law and Genetics observed that:

There may be overlap between the provisions in the *Privacy Act* relating to bodily samples and other laws, including the Human Tissue Acts, the tort of invasion of privacy (should one be created in the future by the High Court), property law ... etc. However, in our view the overlap does not create inconsistencies. It is important to provide adequate coverage with no gaps and therefore the fact that there may be overlap does not preclude the proposed extension of the *Privacy Act*.⁹³

Implementing the reform

8.87 Although the drafters of the *Privacy Act* may not have had genetic samples in mind, the NPPs are drafted as high level principles capable of flexible interpretation in a myriad of circumstances. The NPPs do not prescribe exactly what an organisation must do to comply with them. Rather, they apply broad standards—for example, based on whether an organisation has taken ‘reasonable steps’ to do something, whether certain possible actions are ‘reasonable and practicable’ or ‘impracticable’ and whether information is ‘necessary’ for certain purposes.⁹⁴

8.88 It has been stated that one strength of the principles is that they are ‘technology neutral’,⁹⁵ that is, principles of fair information handling can be applied evenly, no matter the form in which information is held or stored:

The result is that the NPPs apply equally to conventional, electronic and digital environments. This neutrality also aims to ensure that the legislation will not date and will work in practice now and for many years to come.⁹⁶

91 For example, BBC News ‘*Buddha’s ‘Finger’ Beckons Taiwan Crowds*, BBC News, <news.bbc.co.uk/1/hi/world/asia-pacific/1879607.stm>, 18 March 2002. An individual from whom a body part comes may have no property rights in the body part. See Ch 20.

92 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [7.87]–[7.91].

93 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

94 See eg *Privacy Act 1988* (Cth) NPP 1.

95 See Attorney-General’s Department, *Submission to Australian Senate Select Committee on Information Technologies Inquiry into E-Privacy*, 1 August 2000.

8.89 Given the close analogies between genetic samples and forms of data or information, the Inquiry has concluded that the NPPs are sufficiently flexible to extend sensible and balanced privacy protection to genetic samples. The question therefore becomes how best to change privacy laws to make them apply sensibly to the handling of samples. Four possible options are as follows:

1. Amend the Act to define ‘personal information’ and ‘record’ to include bodily samples and leave the courts and the Privacy Commissioner to interpret how the NPPs are to apply to samples in practice;⁹⁷
2. In addition to Option 1, insert new interpretation provisions in the *Privacy Act* to assist in applying the existing NPPs to bodily samples, or amend the NPPs to better adapt them to the handling of bodily samples, as well as to personal information;⁹⁸
3. Insert a new set of privacy principles into the *Privacy Act* dealing specifically with the fair handling of bodily samples; or
4. Enact a new set of privacy principles in the Human Tissue Acts, other existing legislation, or in new stand alone legislation.

8.90 Option 1 would leave the NPPs in their current form. This is the approach taken in New South Wales, where the legislation covers ‘body samples’⁹⁹ but the privacy principles make no special provision for the application of these principles to such samples.¹⁰⁰ The Inquiry is not aware of any situation in which the New South Wales privacy principles have been applied to a bodily sample. However, there is uncertainty as to how a court would, for example, apply the access principle.

8.91 While it may be possible to apply some of the NPPs coherently to samples without further interpretative assistance, in relation to other principles, amendment or interpretative aid appears necessary to ensure predictability of application and desirable regulatory outcomes. This is particularly true in relation to the access principle. The changes necessary to implement a right of access to samples, including rights exercisable by first-degree genetic relatives,¹⁰¹ appear to be substantial and are not easily accommodated within the existing wording of the NPPs.

96 Office of the Federal Privacy Commissioner, *Guidelines to the National Privacy Principles* (2001), OFPC, Sydney, 2.

97 The Privacy Commissioner might develop guidelines to assist organisations to apply the NPPs to genetic samples. Such guidelines are not legally binding but indicate how the Privacy Commissioner will interpret and apply the NPPs: *Privacy Act 1988* (Cth) s 27(1)(e).

98 For example, by referring throughout to ‘disclosure, use or transfer’ rather than to ‘use or disclosure’.

99 *Privacy and Personal Information Protection Act 1998* (NSW) s 4(2); *Health Records and Information Privacy Act 2002* (NSW) s 5(2).

100 See *Privacy and Personal Information Protection Act 1998* (NSW) Pt 2; *Health Records and Information Privacy Act 2002* (NSW) Sch 1.

101 Genetic relatives should also be able exercise similar rights of access to genetic information, other than samples.

8.92 Under Option 3, a new set of privacy principles dealing with the handling of bodily samples would be inserted into the *Privacy Act*. A range of consequential changes to other parts of the *Privacy Act* would also be necessary—notably to the provisions that define what acts or practices constitute an ‘interference with the privacy of an individual’.¹⁰² This approach may be criticised as adding to the proliferation of privacy principles. In addition to the IPPs and NPPs, the *Privacy Act* already contains distinct regimes for regulating the handling of credit reporting information¹⁰³ and tax file numbers.¹⁰⁴

8.93 Option 4 is considered, and rejected, in Chapter 20, particularly in so far as the Human Tissue Acts might be used as the vehicle for reform. Enacting a new set of privacy principles in other existing legislation, or in new stand alone legislation also presents significant problems. The new rules would have to be consistent with information privacy legislation and would introduce even more complexity into the regulation of the handling of bodily samples.

8.94 Under the Inquiry’s favoured option, Option 2, new interpretation provisions could be inserted into the *Privacy Act* to assist in applying the existing NPPs to bodily samples. For example, a new section could be inserted to explain how the access principle (NPP 6) is to be applied to a bodily sample and to state that ‘disclosure’ in relation to a bodily sample means transfer of possession or control of the sample. Because the handling of personal information by Commonwealth or Australian Capital Territory public sector agencies is governed by the IPPs, rather than the NPPs, it would also be necessary to give detailed consideration to new provisions applying the IPPs to bodily samples.¹⁰⁵

8.95 Constitutional limits on federal legislative power are an important consideration in examining how best to extend privacy protection of genetic samples at the federal level. The external affairs power¹⁰⁶ is an important constitutional underpinning for the *Privacy Act*. The Act gave effect to Australia’s agreement to implement the Organisation for Economic Cooperation and Development’s 1980 Guidelines for the Protection of Privacy and Transborder Flows of Personal Data (OECD guidelines) and to its obligations under Article 17 of the *International Covenant on Civil and Political Rights 1966*. To the extent that extending the coverage of the *Privacy Act* to genetic samples may be characterised as protecting information privacy, the amending legislation may also be able to rely on the external affairs power, along with other powers such as the corporations power.¹⁰⁷ In any case, state and territory legislation will also be necessary, especially given that many existing

102 *Privacy Act 1988* (Cth) ss 13, 13A.

103 *Ibid* Pt IIIA.

104 *Ibid* s 17. There are also data-matching guidelines issued under the *Data-matching Program (Assistance and Tax) Act 1990* (Cth) and the Medicare and Pharmaceutical Benefits Program privacy guidelines issued under s 135AA of the *National Health Act 1953* (Cth).

105 The language of the IPPs may be less suited to such application than that of the NPPs. For example, while the NPPs refer simply to organisations dealing with ‘personal information’, the IPPs refer to ‘record-keepers’ having ‘possession or control of a record that contains personal information’.

106 Australian Constitution s 51(xxix).

107 Australian Constitution s 51(xx).

collections of genetic samples are held in state public health systems and lie beyond the reach of Commonwealth legislative power.

8.96 The Inquiry acknowledges that reform will require review of the audit, investigation, complaints handling and enforcement provisions of the Act to determine whether they continue to be appropriate where the protection of genetic samples is at issue.¹⁰⁸

8.97 The resource and policy implications for the OFPC, as the regulator of a future regime that covers samples, will also require more detailed consideration. The OFPC submitted that the implications of the proposed reform for the OFPC generally, and for the compliance unit in particular, are substantial and not limited to major resourcing issues.¹⁰⁹ These implications include that the OFPC would need to:

- develop new expertise to audit compliance;
- address how the regulation of bodily samples would interact with other regulatory standards and OFPC functions; and
- review complaints handling functions and education strategies.¹¹⁰

8.98 Any regulator given new responsibility for regulating the privacy of genetic samples would face similar challenges, whatever the exact mechanisms of regulation. The Inquiry considers the OFPC to be well suited to the task, by reason of its long-standing experience in the regulation of information privacy in Australia. Privacy NSW noted that the proposed reform

would expand the responsibilities of the OFPC and necessitate adequate funding and resources to enable it to carry out all its regulatory responsibilities and functions. However this expanded role would have a positive benefit in better enabling the OFPC to address a range of privacy issues which the current restricted definition inhibits them from dealing with.¹¹¹

Inquiry's views

8.99 Modern data protection or information privacy legislation (of the kind enacted in Australia, New Zealand, Canada, the United Kingdom and most western European countries) was a response to concerns that arose in the 1970s about the privacy implications of computerised data processing. The 1980 OECD guidelines,¹¹²

108 Office of the Federal Privacy Commissioner, *Submission G164*, 27 June 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

109 The OFPC stated that 'the resource requirements for the Office to enforce the Privacy Principles for body samples in any meaningful way across the health sector alone would run into millions of dollars for an Office whose total budget is currently less than \$5 million': Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

110 *Ibid.*

111 Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002.

112 Organisation for Economic Co-operation and Development, *Guidelines Governing the Protection of Privacy and Transborder Flows of Personal Data* (1980).

which formed the template for legislative privacy principles, recognised that the advent of computer technology was creating powerful new capabilities to collect and use information. In a similar way, the new capabilities of genetic technology to collect information now need to be recognised.¹¹³

8.100 Bodily samples constitute such an immediate source of personal information (a 'virtual medical record') that they demand similar comprehensive privacy protection. While a dearth of legislative precedent may suggest a need for care, the Inquiry considers that this is not in itself a reason not to proceed.

8.101 The Inquiry has concluded that legally enforceable privacy standards for the handling of genetic samples (including their collection, storage, use and transfer) are needed. The options for implementing such standards include amendments to information and health privacy legislation, amendments to the Human Tissue Acts or new stand-alone legislation. For the reasons discussed above,¹¹⁴ the Inquiry has concluded that the best way to proceed is through amendment of existing information and health privacy legislation. The standards should be consistent with those that apply to the handling of genetic information derived from the analysis of genetic samples under existing information and health privacy legislation such as the *Privacy Act*.

Recommendation 8-1. The Commonwealth, States and Territories should enact legislation to provide legally enforceable privacy standards for handling genetic samples, including in relation to the collection, storage, use and transfer of samples. The standards should be consistent with those that apply to the handling of genetic information derived from the analysis of genetic samples under existing information and health privacy legislation such as the *Privacy Act 1988* (Cth) (*Privacy Act*).

Recommendation 8-2. The Commonwealth should amend the *Privacy Act* to extend the coverage of the Information Privacy Principles and National Privacy Principles (or similar privacy principles) to identifiable genetic samples. This may be done by:

- (a) defining 'personal information' and 'health information' to include bodily samples from an individual whose identity is apparent or can reasonably be ascertained from the sample; and
- (b) defining a 'record' to include a bodily sample.

113 See eg T Lemmens and L Austin, *Of Volume, Depth and Speed: the Challenges of Genetic Information* (2001), Report prepared for the Canadian Biotechnology Advisory Committee.

114 See also Ch 20 in relation to reform of the Human Tissue Acts.

Recommendation 8–3. The Commonwealth should amend the *Privacy Act* to provide that an individual has a right to access his or her own bodily samples, through a nominated medical practitioner, for the purpose of medical testing, diagnosis or treatment. The right of access should be limited to a right to obtain access to part of the sample. Access may be refused where:

- (a) it is not physically possible to provide part of a sample;
- (b) providing part of a sample means that the remaining portion is insufficient for the purposes of the organisation retaining it; or
- (c) releasing a sample to an individual raises public health concerns.

Recommendation 8–4. The Commonwealth should amend the *Privacy Act* to provide that an individual has a right to access bodily samples of his or her first-degree genetic relatives, through a nominated medical practitioner, where access is necessary to lessen or prevent a serious threat to his or her life, health, or safety, even where the threat is not imminent. The right of access should be limited to a right to obtain access to part of the sample. Where an organisation subject to the *Privacy Act* receives a request for access, the organisation should be obliged to seek consent from the genetic relative, where practicable, before determining whether to provide access. Access may be refused where:

- (a) it is not physically possible to provide part of a sample;
- (b) providing part of a sample means that the remaining portion is insufficient for the purposes of the organisation retaining it;
- (c) releasing a sample to an individual raises public health concerns; or
- (d) providing access would have an unreasonable impact upon the privacy of the individual from whom the sample comes.

Implications of the alternative view of existing coverage

8.102 This chapter has proceeded on the basis that a name or a coded name (by itself) is not ‘information about an individual’ for the purposes of the *Privacy Act*. As discussed above, another possible interpretation of the existing provisions of the *Privacy Act* is that a name, by itself, may constitute information about an individual. Therefore, labelled samples, in effect, may already have to be handled in accordance with the IPPs or NPPs (assuming that the organisation that holds them is covered by the *Privacy Act*). For example, by transferring a labelled sample, an organisation will also be disclosing information (the name of the individual) and will have to comply with NPP 2.

8.103 If this interpretation is correct the gaps in the privacy protection of genetic samples may be more limited than indicated in this chapter. However, problems would remain. The obligations in the IPPs and NPPs relate to the handling of the information, not the sample itself. This leads to illogical results. For example:

- Under NPP 1.3, an organisation collecting labelled samples would have to take reasonable steps to ensure that the individual is aware that his or her name has been collected for the purpose of labelling a sample, but would be under no obligation to inform the individual that a sample has been collected or about the purpose of collection or what will happen to it.
- Under NPP 2.1(b), when an organisation intends to use or disclose labelled samples for a secondary purpose, the organisation would have to obtain consent to the use or disclosure of the information on the labels, but not to the use or transfer of the samples themselves. For example, an organisation could seek and obtain consent from an individual to the disclosure of ‘your personal details and health information’ for research purposes without disclosing that the individual’s sample will also be transferred.
- Under NPP 6.1, an individual would have a right of access to the label on the sample, but not to the sample itself.
- A fundamental gap in coverage would remain in relation to the handling of samples that are not labelled but are identifiable by other means, for example by matching DNA profiles.

9. Anti-Discrimination Law

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Introduction

Officially they are called 'In-valids'. ... They are the 'healthy ill'. They don't actually have anything yet — they may never have. But since few of the pre-conditions can be cured or reversed, it is easier to treat them as if they were already sick.¹

1 A Niccol, *GATTACA* (1997), Columbia Pictures.

9.1 This quote, from the film GATTACA, describes an imaginary future world, but discrimination on the ground of genetic status is no longer science fiction. Recognising this, the Inquiry's terms of reference asked whether a regulatory framework is required to provide protection from inappropriate discriminatory use of human genetic samples and information.

9.2 Many submissions received by the Inquiry included statements about the undesirable effects of inappropriate and discriminatory use of genetic information. The Anti-Discrimination Board of NSW stated:

Rather than acting as an impediment to the development and application of genetic technology, effective anti-discrimination and privacy legislation are critical to realising the public health benefits of genetic information. Conversely, if we fail to provide such protection, discrimination and privacy concerns will act as disincentives to testing and research participation and have negative consequences for individual and public health outcomes.²

9.3 The Human Genetics Society of Australasia provided a topical example:

One concern is that people will avoid having potentially important tests that may help in their clinical management for fear of discrimination. For example, a test for factor 5 Leiden would enable a person to know if they were at increased risk of developing a deep vein thrombosis when on a long flight. However, fear of not being allowed to fly or not obtaining insurance may prevent them taking the test.³

9.4 In some circumstances, discrimination on the ground of a person's genetic status may already be unlawful under existing race, sex, or disability anti-discrimination laws. This chapter explores possible deficiencies in this protection, particularly in relation to disability discrimination. The Inquiry has concluded that genetic discrimination should be addressed within the existing legal framework, subject to a range of recommendations intended to ensure comprehensive protection.

9.5 Australia has anti-discrimination legislation at the federal level as well as legislation in all States and Territories.⁴ This chapter focuses on the federal legislation but reference is made to the state and territory legislation in the context of discussing the need for greater harmonisation across Australian jurisdictions and for the purpose of examining alternative models.

9.6 The primary pieces of federal anti-discrimination legislation are the:

- *Sex Discrimination Act 1984* (Cth) (SDA);
- *Racial Discrimination Act 1975* (Cth) (RDA);

2 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

3 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

4 *Racial Discrimination Act 1975* (Cth); *Sex Discrimination Act 1984* (Cth); *Disability Discrimination Act 1992* (Cth); *Human Rights and Equal Opportunity Commission Act 1986* (Cth); *Anti-Discrimination Act 1977* (NSW); *Equal Opportunity Act 1995* (Vic); *Anti-Discrimination Act 1991* (Qld); *Equal Opportunity Act 1984* (WA); *Equal Opportunity Act 1984* (SA); *Anti-Discrimination Act 1998* (Tas); *Discrimination Act 1991* (ACT); *Anti-Discrimination Act 1992* (NT).

- *Disability Discrimination Act 1992* (Cth) (DDA); and
- *Human Rights and Equal Opportunity Commission Act 1986* (Cth) (HREOC Act).

9.7 In addition, the *Workplace Relations Act 1996* (Cth) (WRA) contains provisions that prohibit discrimination on a range of grounds in terminating employment. The WRA is discussed in more detail in Chapter 30. The Inquiry also notes that, on 9 January 2003, the Attorney-General, the Hon Daryl Williams AM QC MP, released an information paper on proposed age discrimination legislation and called for submissions on the proposal. A Core Consultative Group has been established to help progress the development of the legislation.⁵

Constitutional issues

Commonwealth constitutional powers

9.8 The federal Parliament's power to legislate is set out in, and limited by, the Commonwealth Constitution. The relevant provisions do not expressly refer to 'human rights' or 'discrimination' and the Commonwealth's legislation in this area is based on a suite of powers, including the 'external affairs' power in s 51(xxix). The High Court has interpreted this provision to mean that the Commonwealth may enact laws to implement its international legal obligations,⁶ subject to certain implied and express constitutional limitations, and provided the laws are reasonably appropriate and adapted to implement the obligations.⁷

9.9 In addition, the DDA and the SDA expressly identify a number of other heads of constitutional power that support the legislation. The intention of these provisions is to extend the reach of the legislation as far as possible, given the absence of an express power to enact laws with respect to discrimination. For example, under s 12 of the DDA, the Act extends to:

- matters covered by specified international conventions;
- matters external to Australia or of international concern;⁸
- discrimination by foreign, trading or financial corporations;
- discrimination in the course of carrying on the business of insurance or banking;
- discrimination in the course of interstate or international trade and commerce; and

5 Attorney-General, *Media Release: Public comment on age discrimination laws*, Attorney-General, <www.law.gov.au/ageinfopaper>, 9 January 2003.

6 *Commonwealth v Tasmania* (1983) 158 CLR 1 (The Tasmanian Dam Case).

7 *Richardson v Forestry Commission* (1988) 164 CLR 261.

8 See *Soulitopoulos v LaTrobe University Liberal Club* [2002] FCA 1316; *O'Connor v Ross (No 1)* [2002] FMCA 210, confirming that the prohibition of disability discrimination is a matter of international concern.

- discrimination against Commonwealth employees in connection with their employment.

State and territory laws

9.10 The States and Territories do not have such constitutional limitations. Each jurisdiction has enacted anti-discrimination legislation and, while the Acts are not identical, there are considerable similarities between them. In many situations involving claims of discrimination, state and territory anti-discrimination legislation overlaps with federal laws. Where this occurs, an individual may have a choice of legislation under which to seek redress.

9.11 Under s 109 of the Constitution, in the event of an inconsistency between federal and state laws, the federal law prevails and the state law is inoperative to the extent of the inconsistency.⁹ This caused problems in the early days of anti-discrimination legislation in Australia. All federal anti-discrimination Acts now contain provisions expressly indicating that the federal Act is not to be taken to exclude or limit the operation of any state or territory law capable of operating concurrently with the federal Act. Some problems of articulation between federal and state laws still remain, for example in the field of insurance.¹⁰

International context

9.12 Australia's federal anti-discrimination legislation makes reference to a wide range of international instruments, as well as 'matters external to Australia' (in the case of the DDA and SDA) and 'matters of international concern' (in relation to the DDA).¹¹ These instruments do not specifically address discrimination on the basis of genetic status. However, as noted in Chapter 4, the international community has, in recent years, been turning its attention to this matter in some detail. The UNESCO *Universal Declaration on the Human Genome and Human Rights 1997* recognises that:

research on the human genome and the resulting applications open up vast prospects for progress in improving the health of individuals and of humankind as a whole, but ... that such research should fully respect human dignity, freedom and human rights, as well as the prohibition of all forms of discrimination based on genetic characteristics.¹²

9.13 The Declaration is not a binding legal instrument but is evidence of growing international concern and an indication of the general approach of the international community in this area. Article 2 of the Declaration states that:

⁹ *Dao v Australian Postal Commission* (1987) 162 CLR 317.

¹⁰ The High Court has held that provisions of the *Life Insurance Act 1945* (Cth) (allowing life insurers to classify risks according to actuarial evidence) are inconsistent with provisions of the *Anti-Discrimination Act 1977* (NSW) (precluding insurers from discriminating against insurance applicants in respect of disability). See *Australian Mutual Provident Society v Goulden* (1986) 160 CLR 330.

¹¹ These references were included to help ensure the widest possible constitutional basis for this legislation following the reasoning of the High Court in *Koowarta v Bjelke-Petersen* (1982) 39 ALR 417.

¹² *Universal Declaration on the Human Genome and Human Rights*, UNESCO, <www.unesco.org/ibc/en/genome/projet/>, 19 February 2003.

Everyone has a right to respect for their dignity and for their rights regardless of their genetic characteristics. That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity.

9.14 Article 6 goes on to declare that:

No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.

9.15 The Council of Europe's Convention on Human Rights and Biomedicine, which is a legally binding instrument and has been signed and ratified by 15 countries to date, gives a clear indication of the approach adopted in Europe in relation to this issue. Article 11 states that:

Any form of discrimination against a person on grounds of his or her genetic heritage is prohibited.¹³

9.16 It is against this background that the Inquiry was asked to consider whether the protection offered by existing legislation in Australia is adequate.

Australian anti-discrimination law framework

9.17 Despite the differences of detail between Australian jurisdictions, all legislation dealing with discrimination embodies the same paradigm or framework for identifying unlawful discrimination. Generally, for discrimination to be unlawful, an act or omission must:

- be based on one of the grounds or attributes set out in the legislation, such as race, sex or disability;
- fall within an area of activity set out in the legislation, such as employment, education, or the provision of goods or services;
- result in some harm or less favourable treatment, whether by direct or indirect discrimination; and
- not fall within an exception, exemption or defence.

Specified ground or attribute

9.18 In order to be unlawful, the discrimination must be based on one of the grounds or attributes set out in the legislation. This means that the statutory definitions of these grounds are crucial to the operation of the legislation. In Australia, these grounds vary from jurisdiction to jurisdiction and include race, sex, sexuality, pregnancy, marital status, parental status, age, disability, religion, political belief or

13 *Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine*, opened for signature 4 April 1997, ETS No 164, (entered into force on 1 December 1999).

activity, and trade union activity. If a person is discriminated against on the basis of an attribute that is not listed in the legislation, for example, simply because the discriminator does not like them personally, the victim has no remedy under anti-discrimination law.

9.19 Most of the grounds listed in anti-discrimination legislation reflect the community's belief that the attribute in question is, or at least should be, an irrelevant consideration in the particular context in which the question arises. For example, discrimination between a man and a woman in employment is permitted on the basis of their qualifications, skills or experience, but not generally on the basis of their sex. This legislative judgment is based on the belief that sex does not usually have a relevant impact on the ability of the individual to perform the job.

9.20 However, exceptions to anti-discrimination provisions recognise that, in some circumstances, grounds or attributes may be relevant and may be taken into account. Under the SDA, for example, it is not unlawful for an insurer to discriminate on the basis of sex in the terms on which an insurance policy is offered if the discrimination is based on reasonable actuarial and statistical data. In the same way, the DDA recognises the relevance of a person's disability in certain circumstances by including a number of exceptions, including an insurance exception. In addition, the DDA does not simply require that employers and others disregard a person's disability. Employers and others are expressly required to make reasonable efforts to accommodate disabled individuals so that they are, for example, able to perform the job, despite their disability. The DDA and the duty to provide reasonable accommodation are discussed further below and in Part H of this Report.

9.21 Potentially, any of the grounds or attributes listed in Australian anti-discrimination law may be relevant to discrimination on the basis of genetic status. The relevance of these grounds will depend upon our expanding understanding of genetics, and of the way in which genes may influence attributes such as race, sexuality and so on. Currently, the most obviously relevant ground is disability or impairment and this is discussed in detail below.

Specified area of activity

9.22 In order to be unlawful, discrimination must occur in a field of activity set out in the legislation. The areas specified in Australian anti-discrimination legislation vary from jurisdiction to jurisdiction and include employment, education, the provision of goods and services (for example, insurance), the provision of accommodation, the disposition of land, membership of clubs, and the administration of laws and government programs. This coverage is wide, but does not generally include acts done in the private sphere, reflecting the public/private distinction that runs through much of Australian anti-discrimination law.

9.23 Some legislation has adopted a slightly different approach in indicating those areas of public life covered. The RDA refers to specific areas,¹⁴ but also contains a more general provision, based on the language of the *International Convention on the Elimination of all Forms of Racial Discrimination 1966*. Section 9(1) of the RDA states in part that:

It is unlawful for a person to do any act involving a distinction, exclusion, restriction or preference based on race ... which has the purpose or effect of nullifying or impairing the recognition ... of any human right ... in any field of public life.¹⁵

9.24 The Genetic Privacy and Non-discrimination Bill 1998 (Cth), which is discussed below, also adopted this approach. Clause 17 of the Bill was in the same terms as s 9 of the RDA, with the substitution of ‘genetic information’ for ‘race’.

Direct and indirect discrimination

9.25 Australian law recognises two ways in which discrimination may occur. These are direct discrimination and indirect discrimination.

9.26 Direct discrimination occurs when a person is treated less favourably than another person who does not share the first person’s attribute. For example, refusing to admit non-Caucasians to a cinema amounts to unlawful racial discrimination. This is so whether the discriminatory policy is worded positively (‘Whites Only’) or negatively (‘No Non-Whites’).

9.27 This type of discrimination is the most obvious to identify. The intention of the discriminator is irrelevant: a person who believes he or she is doing the right thing (for example, dismissing a pregnant woman ‘for her own good’) is liable in the same way as someone who is blatantly biased and actively discriminatory.

9.28 If an action is done for more than one reason, one of which is discriminatory on its face and the other of which is not (for example, refusing service in a hotel to someone of a particular race who is also drunk or improperly dressed), there may still be liability for unlawful discrimination, but this differs between jurisdictions. Under the RDA a complainant need only show that it was one of the reasons for the act ‘whether or not it is the dominant reason or a substantial reason’.¹⁶

9.29 Indirect discrimination is less obvious and more difficult to identify. It is sometimes called ‘adverse impact’ discrimination because it focuses on the effect of the discriminator’s action rather than on the attributes of the person towards whom the action is directed, although the latter are still relevant. Australian law is not uniform with respect to the elements comprising indirect discrimination. Generally, it must be shown that a requirement or condition is imposed which, even though neutral on its face, has an adverse impact on people with a particular attribute, in circumstances where this is unreasonable. As with direct discrimination, an intention to discriminate is not necessary.

14 *Racial Discrimination Act 1975* (Cth) ss 11–15.

15 *Ibid* s 9(1).

16 *Ibid* s 18.

9.30 For example, a requirement that all users of public transport buy tickets that are validated for travel by scratching off segments may appear to be non-discriminatory. However, in *Waters v Public Transport Corporation of Victoria*¹⁷ it was held that this requirement has a greater adverse impact on people who have visual, motor or intellectual impairments than it does on people without such impairments. Such a requirement, where unreasonable, amounts to indirect disability discrimination.

Exemptions, exceptions and defences

9.31 Anti-discrimination laws contain a number of exemptions, exceptions and defences. If they apply, an otherwise valid complaint of discrimination cannot be sustained. Exemptions, exceptions and defences in Australian anti-discrimination legislation include ‘unjustifiable hardship’ in accommodating a person’s disability, and acts done to comply with other legislation, such as occupational health and safety legislation. These issues, which arise in the employment context, are examined further in Chapters 30, 31, 32 and 33. Broad exceptions relating to insurance, which raise important issues about the use of genetic information by the insurance and financial services industry, are discussed in detail in Chapters 26 and 27.

9.32 In some circumstances it may be possible for a person or body to apply to the agency administering the anti-discrimination legislation for an exemption with respect to a particular activity. In relation to the DDA and SDA, for example, a person may apply to the Human Rights and Equal Opportunity Commission (HREOC) for a temporary exemption from the operation of the legislation. HREOC may grant an exemption for a period of up to five years, provided it is not inconsistent with the objects of the legislation.¹⁸

Principal federal legislation

Sex Discrimination Act

9.33 The SDA prohibits discrimination on the basis of sex. The Act also extends to discrimination on the basis of a characteristic that generally relates to people of a particular sex or that is generally imputed to people of a particular sex.¹⁹ The Act also prohibits discrimination on the ground of marital status, pregnancy or potential pregnancy and, in the area of employment, family responsibilities.

9.34 In certain circumstances, discrimination on the basis of genetic status may amount to sex discrimination. For example, refusing to employ people with a genetic predisposition to prostate cancer or a family history of breast cancer may give grounds for a complaint of indirect discrimination under the SDA. This is because apparently neutral requirements such as these may have an unreasonable adverse impact on men, in the first case, and on women, in the second case. Such practices may also amount to direct disability discrimination.

17 *Waters v Public Transport Corporation of Victoria* (1991) 173 CLR 349.

18 See eg *Disability Discrimination Act 1992* (Cth) s 55.

19 *Sex Discrimination Act 1984* (Cth) s 5(1).

Racial Discrimination Act

9.35 The RDA prohibits discrimination based on race, colour, descent or national or ethnic origin. Discrimination on the basis of genetic status may, in some circumstances, amount to racial discrimination because some disorders are known to be more prevalent in particular races and communities. For example, Tay Sachs disease is prevalent in some Jewish populations and sickle cell disease is prevalent in certain African populations. Refusing to provide goods and services to people with Tay Sachs disease or sickle cell anaemia may give grounds for a complaint of indirect racial discrimination under the RDA.

9.36 Refusing to employ people from a particular racial group because that racial group is more likely to have a genetic predisposition to a particular disorder may also give grounds for complaint of direct discrimination under the RDA. Another possibility is that the ‘descent’ element of the definition of race might be relied on to ground a claim of discrimination based on genetic status.

Disability Discrimination Act

9.37 The DDA is the most relevant piece of anti-discrimination legislation in this area. The DDA prohibits discrimination on the basis of disability. The definition of ‘disability’, and whether it is wide enough to cover discrimination on the basis of genetic status, is discussed in detail below.

9.38 The DDA covers a disability which a person:

- has now;
- had in the past (for example, a past episode of mental illness);
- may have in the future (for example, a late onset genetic disorder); or
- is imputed to have (for example, a genetic disorder that a person is assumed to have on the basis of that person’s family medical history).

9.39 Discrimination is prohibited in employment, education, access to premises used by the public, provision of goods, services and facilities, accommodation, buying land, activities of clubs and associations, sport and the administration of Commonwealth government laws and programs. Discrimination on the ground of genetic status may, potentially, arise in many of these contexts. Consistently with the emphasis in the Inquiry’s Terms of Reference, and with the level of concern expressed in submissions, this Report focuses on discrimination in employment and insurance. These areas are considered in detail in Chapters 26, 27, 30 and 31. The Report also considers the use of genetic information in sport in Chapter 38.

9.40 Under s 31 of the DDA, the Attorney-General may formulate Disability Standards which, once tabled in Parliament for a certain period, gain the force of law. Such standards are intended to provide greater detail and more certainty in relation to

rights and responsibilities under the Act. Standards can be made in the areas of employment, education, public transport services, access to premises, accommodation and the administration of Commonwealth laws and programs. The first Disability Standards for Accessible Public Transport were tabled in Parliament for the required period and took effect on 23 October 2002.

9.41 In addition, HREOC may issue guidelines under the DDA to assist persons and organisations with responsibilities under the legislation to avoid discrimination and comply with their responsibilities. HREOC has, for example, issued Guidelines for Providers of Insurance and Superannuation.²⁰ Unlike standards, these guidelines are not legally binding.

9.42 Part 3 of the DDA also provides for the development and implementation of Action Plans by those who provide goods, services or facilities. Action Plans are intended to set out the policies and programs a service provider has put in place to achieve the objects of the DDA. In developing Action Plans, service providers must review their practices to identify and attempt to eliminate discriminatory practices. Action Plans are one of the factors that HREOC is required to consider in attempting to conciliate complaints of disability discrimination against service providers. Although an Action Plan does not constitute a defence to a complaint, if it establishes the respondent's commitment to do everything possible to eliminate discrimination within a reasonable period of time, and if the respondent is actively implementing the Action Plan, it is possible for HREOC to find that it would impose unjustifiable hardship to require the respondent to do more.²¹

9.43 Finally, s 30 of the DDA makes it unlawful to request or require information in connection with, or for the purposes of doing, an act of discrimination. The meaning and scope of this provision is not entirely clear. This provision is discussed in detail in Chapter 31.

9.44 In February 2003, the federal government announced a review of the DDA by the Productivity Commission. The Commission has been asked to undertake a cost-benefit analysis of the legislation and, in particular, of any restrictions on competition in the Act. The Commission is expected to report by February 2004.

Human Rights and Equal Opportunity Commission Act

9.45 The HREOC Act establishes HREOC and enables it to handle complaints under the SDA, the RDA and the DDA. HREOC may accept complaints under these three Acts, inquire into them, and attempt to settle them by conciliation. Where this is not possible, a complainant may choose to apply to the Federal Court or the Federal Magistrates Court to seek a binding determination.

20 Human Rights and Equal Opportunity Commission, *Guidelines for Providers of Insurance and Superannuation*, <www.hreoc.gov.au/disability_rights/standards/Insurance/insurance_adv.html>, 19 February 2003.

21 Copies of Action Plans are registered with HREOC, and some are available on its web site: Human Rights and Equal Opportunities Commission, *Developing an Effective Action Plan*, HREOC, <www.hreoc.gov.au/disability_rights/action_plans/Effective_Plan/effective_plan.html#1>, 7 February 2003.

9.46 In addition, HREOC may inquire into any act or practice, including any systemic practice, which impairs equal opportunity in employment on a wide range of grounds that might be relevant to genetic information. These include race, colour, sex, national extraction, social origin, age, medical record, sexual preference, impairment, physical, mental, intellectual or psychiatric disability. The definition of disability for the purposes of such an inquiry differs from the definition of disability used in the DDA. HREOC has used these powers to inquire into systemic age discrimination practices in employment.

9.47 The HREOC Act also provides HREOC with the power to inquire into alleged breaches of human rights by or on behalf of the Commonwealth, for example, by a government department.

9.48 In contrast to complaints under the SDA, RDA and DDA, individual complaints of discrimination brought under these provisions of the HREOC Act cannot be dealt with by a court or tribunal and therefore cannot lead to an enforceable remedy. For this reason, complaints tend to be lodged under one of the other Acts where an enforceable remedy is available.

Existing legal framework or new legislation?

9.49 A preliminary question is whether it would be better to amend existing anti-discrimination laws to clarify their application, or enact new legislation dealing specifically with genetic discrimination. The question has been prompted, in part, by the Genetic Privacy and Non-discrimination Bill introduced into the federal Parliament by Senator Natasha Stott Despoja in 1998.²² The Bill was largely based on a Bill then before the United States Congress. For reasons explained further below, the Inquiry has concluded that discrimination on the ground of genetic status should continue to be dealt with under the framework of existing federal, state and territory anti-discrimination laws, subject to the legislative amendments and other safeguards recommended in this Report.

Submissions and consultations

9.50 Several submissions to the Inquiry, including Senator Stott Despoja's submission, expressed support for separate legislation.²³ Although ultimately rejecting this approach, the Centre for Law and Genetics set out some of the potential advantages of separate legislation in its submission:

There would undoubtedly be some advantages in enacting legislation dealing specifically with genetic discrimination, including the advantage of greater certainty of protection, heightened visibility, and the consequential effect of public consciousness raising.²⁴

22 The Australian Bill was returned to the Senate Notice Paper on 14 May 2002.

23 Department of Health Western Australia, *Submission G271*, 23 December 2002; Health Consumers' Council, *Submission G174*, 18 September 2002; N Stott Despoja, *Submission G198*, 27 November 2002.

24 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

9.51 The ‘stand alone’ legislation approach has been adopted in a number of States in the United States and in some European countries. The Human Genetics Commission (HGC) in the United Kingdom also recommended separate legislation in its final report. In relation to the *Disability Discrimination Act 1991* (UK), the HGC was of the view that it would be too difficult to amend the definition of disability in the Act, which adopts a very different approach to the DDA, to address genetic disorders, especially presymptomatic disorders.²⁵

9.52 However, the majority of submissions received in response to DP 66, including those from the acting Disability Discrimination Commissioner,²⁶ the Anti-Discrimination Board of NSW,²⁷ the Institute of Actuaries of Australia,²⁸ the Centre for Genetics Education²⁹ and the Human Genetics Society of Australasia³⁰ expressed support for dealing with the issue within the existing legislative framework. The Genetic Discrimination Project Team, for example, commented:

We support the proposal to work within the existing legal framework (Proposal 8-1). This would appear to us to be the most logical, least interventionist approach. This is far preferable to developing a whole new legislative framework: this is both unnecessary and undesirable and would attract justifiable criticism of genetic exceptionalism.³¹

9.53 In its 1999 report, the Senate Legal and Constitutional Committee concluded in relation to the Genetic Privacy and Non-discrimination Bill 1998:

5.28 The committee considers that it would be more appropriate to amend, where necessary, existing privacy and discrimination legislation to ensure that issues raised by genetic technology are adequately covered under that legislation, for example, the various federal anti-discrimination acts, the *Human Rights and Equal Opportunities Act 1986*, and the *Privacy Act 1988*.

5.29 The committee believes that such an approach provides a clearer legislative base. It avoids the administrative and legal confusion created by having different sets of rules applying to genetic information than to other personal information ...

5.30 Creating specific legislation such as the bill would also cut across a number of regulatory systems already in place, or in the process of being established, that are themselves the product of extensive consultation and negotiation between stakeholders and state, territory and federal governments.³²

25 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London [6.31].

26 Acting Disability Discrimination Commissioner — Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

27 Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002.

28 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

29 Centre for Genetics Education, *Submission G232*, 18 December 2002.

30 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

31 Genetic Discrimination Project Team, *Submission G252*, 20 December 2002.

32 Senate Legal and Constitutional Legislation Committee, *Provisions of the Genetic Privacy and Non-discrimination Bill 1998*, The Parliament of Australia, <www.aph.gov.au/senate/committee/legcon_ctte/genetic/index.htm>, 21 August 2002 [5.28]–[5.30].

9.54 The Anti-Discrimination Board of NSW shared the Senate Committee's concerns about administrative and legal confusion and went on to state in its submission:

There are numerous benefits to retaining genetic discrimination within [the] conceptual framework of existing anti-discrimination legislation. Many of the issues discussed above in relation to uniformity of legislation also apply to this issue including:

- greater clarity about people's rights and responsibilities under anti-discrimination law where there are fewer pieces of legislation
- a reduction in the complexity of jurisdictional decisions for would-be complainants
- increasing the likelihood that case law from one jurisdiction is applicable in another and for precedent to be applied ...

Retaining genetic discrimination within [the] conceptual framework of existing anti-discrimination legislation will ensure that we do not afford different levels of protection to people with disabilities diagnosed by genetic testing, or future or imputed disabilities based on predictive genetic testing compared with other people with disabilities.³³

Inquiry's views

9.55 The Inquiry considers these submissions to be persuasive. The Inquiry recommends that discrimination on the grounds of genetic status be dealt with under existing anti-discrimination legislation subject to the legislative amendments and other safeguards recommended in this Report. Working within the existing legal framework will promote certainty and consistency and will build on existing understanding and practice in this field. The Inquiry does not support the development of separate genetic discrimination legislation.

Recommendation 9–1. Discrimination on the ground of genetic status should continue to be dealt with under the framework of existing federal, state and territory anti-discrimination laws, subject to the legislative amendments and other safeguards recommended in this Report.

Genetic status and disability in the DDA

9.56 If discrimination on the grounds of genetic status is to be dealt with under existing legislation, as recommended, the most relevant piece of federal legislation is the DDA. While some cases of discrimination on the ground of a person's genetic status may give rise to issues under the RDA and the SDA, most cases will involve

33 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

discrimination on the basis that a person's genetic status is disabling in some way, or is perceived to be disabling. It is important to ensure that the DDA protects individuals from discrimination in these circumstances.

9.57 There are a number of ways in which the DDA could be clarified in relation to genetic discrimination. It would be possible, for example, to amend the short title or the objects clause of the Act to indicate that it was intended to apply to discrimination on the ground of genetic status. Another option would be to clarify the existing definition of 'disability' to ensure that it includes genetic status or to include genetic status as a new and separate ground under the Act. Each of these options is considered below.

Short title and objects clause

9.58 DP 66 sought feedback on the proposal that the name of the DDA be changed to the *Disability and Genetic Discrimination Act 1992*. The proposal was put forward on the basis that the change would bring some of the advantages cited in the Centre for Law and Genetics' submission quoted above, that is, heightened visibility and public awareness and that it would emphasise that genetic status did not necessarily equate with disability.

9.59 DP 66 also sought feedback on s 3 of the DDA, which sets out the objects of the legislation. At present this provision does not expressly address the harm caused by discrimination on the basis of imputed, past or possible future disabilities. An amended objects provision could make clear that the DDA is intended to prohibit discrimination of this kind, including discrimination on the basis of genetic status.

Submissions and consultations

9.60 Opinion in submissions was divided on changing the name of the DDA. The Anti-Discrimination Commission of Queensland, the Genetic Support Council of WA, the Human Genetics Society of Australasia and others expressed the view that a change of name would assist with community awareness. However, the Centre for Genetics Education, although expressing support for the name change, cautioned that including the term 'genetics' in the title of the DDA would more closely link the concepts of genetics and disability:

The concern is that it will increase the possibility of linking a positive genetic test result with a "disability"—already that is occurring and much effort is put into correcting this misconception both at the individual clinical level and in genetics education generally.³⁴

9.61 The Anti-Discrimination Board of NSW did not support amending the name of the DDA:

34 Centre for Genetics Education, *Submission G232*, 18 December 2002.

In our view this would suggest that a distinction should or does exist where people are discriminated against on the basis of future or imputed conditions detectable by genetic testing, as compared with people who are discriminated against on the basis of future or imputed conditions which are not detected by genetic testing. Concerns about access to and use of health information are no different in relation to people with conditions or predisposition to conditions which can be determined by genetic testing than for other people with disabilities, whether actual, future or imputed.³⁵

9.62 The Centre for Law and Genetics was also opposed to changing the name of the legislation:

Not only would this be likely to attract criticism of genetic exceptionalism (ie singling out genetic conditions for special treatment), it would run counter to the essential framework of the legislation. The legislation already covers discrimination on the basis of a risk of disability in the future or on grounds which are imputed to a person: to deal with genetic status separately (on the grounds that this is not a disability which presently exists and indeed, may never materialise) would give the wrong impression that the *Disability Discrimination Act* 1992 (Cth) is only concerned with existing disability and may therefore undermine much of the good work that has been done in educating the community about avoiding disability discrimination in all its forms.³⁶

9.63 The acting Disability Discrimination Commissioner, while remaining open to community views on this issue, noted that the DDA applies in numerous cases that do not involve a disability, as that term is generally understood. He concluded:

It is clearly not possible for a concise title to reflect all of the other areas where 'disability' may not be a clear enough sign that the legislation applies, and concerns may be raised as to why genetic issues alone should be highlighted.³⁷

9.64 There was more consistent support for an amendment to the objects clause in the DDA,³⁸ although the Anti-Discrimination Board of NSW expressed the view that, as the objects clause was drafted in a very general way, it may not be appropriate to include a provision specifically about genetic discrimination.

Inquiry's views

9.65 The Inquiry is of the view that, on balance, the arguments against changing the name of the DDA are stronger than those in favour of change. The Inquiry has generally resisted making recommendations in this Report based on genetic exceptionalism. Discrimination on the ground of genetic status fits within the existing

35 Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002.

36 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

37 Acting Disability Discrimination Commissioner - Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

38 N Stott Despoja, *Submission G198*, 27 November 2002; Anti-Discrimination Commission Queensland, *Submission G214*, 2 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Acting Disability Discrimination Commissioner - Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

conceptual framework of the DDA. Including ‘genetic’ in the title of the Act may cause confusion in relation to the legal meaning of the term ‘disability.’ The Inquiry believes that community awareness on this issue can be raised in other ways, for example, through education and publicity.

9.66 However, the Inquiry is of the view that it would be desirable to make a statement in s 3 of the DDA, which sets out the objects of the Act, indicating that the Act applies to discrimination on the basis of past, present, possible future or imputed disabilities, including discrimination on the ground of genetic status. A statement of this kind would sit more comfortably with the existing high level statements in s 3 and could make clear that discrimination on the basis of genetic status is simply one example of discrimination on the basis of imputed or possible future disability.

Recommendation 9–2. The Commonwealth should amend the objects clause of the *Disability Discrimination Act 1992* (Cth) (DDA) to clarify that the Act applies to discrimination in relation to past, present, possible future or imputed disability, including discrimination on the ground of genetic status.

Genetic status as a new ground

9.67 In some cases a person’s genetic status may give rise to a disability in the generally understood sense. As noted above, however, this is not always the case, particularly in relation to genetic information that is merely predictive and which may indicate nothing more than an elevated level of risk of developing a disorder at some time in the future. An editorial in the *British Medical Journal* has remarked that:

On a fundamental level, genetic science is forcing a re-examination of the concept of normality itself, by showing that everyone’s genome is different and that we are all in some sense ‘abnormal’. We each carry genetic variants, many of which will have no detectable impact in normal circumstances, but some undoubtedly will alter our risk of disease or may, with a partner carrying similar variations in their genomes, result in the birth of a child with a recessive genetic disorder.³⁹

Submissions and consultations

9.68 The Centre for Law and Genetics expressed the view that adding genetic status as a separate ground in the DDA

is not a justifiable response given the range of conditions and disorders (past, present, future and imputed) that this legislation is intended to cover. Singling out one category, in the form of genetic status would create an imbalance in the legislation and would probably give rise to claims of unfair special treatment for those who are at risk of genetic disease.⁴⁰

39 R Zimmern, ‘Genetics and Medicalisation’ (2002) 324 *British Medical Journal* 863. See also F Fukuyama, *Our Posthuman Future: Consequences of the Biotechnology Revolution* (2002) Profile Books, London, 209, who notes that what society considers to be pathology or disease is actually a socially constructed phenomenon in which deviation from some presumed norm is stigmatised.

40 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

Inquiry's views

9.69 The legal concept of disability as it is used in the DDA is wide and does not necessarily equate with the general understanding of the term. This is because the DDA is not intended to address only discrimination against people with existing disabilities but to address the harm caused by incorrect assumptions made about the existence, or impact, of disabilities. Discrimination based on an incorrect assumption that a person has a disability has the potential to cause as much harm as discrimination on the basis of an actual disability. This is why the definition of disability in the DDA extends to imputed disabilities, as well as disabilities that existed in the past or may exist in the future, in addition to existing disabilities.

9.70 The Inquiry is of the view that discrimination on the basis of genetic status fits within this wider legal concept of disability discrimination. Although adding a separate ground to the Act may help to emphasise that genetic status does not necessarily equate with disability, it would also add considerably to the complexity of the legislation. On balance, the Inquiry does not support including genetic status as a separate ground under the DDA. However, as discussed in the following section, the definition of 'disability' should be amended to clarify its application to discrimination based on genetic status.

Definition of disability***Current law***

9.71 If discrimination on the ground of genetic status is to be dealt with under the framework of existing legislation, and if genetic status is not to be an independent ground of discrimination, it is important to ensure that the definitions in the DDA are wide enough to cover genetic issues. Section 4(1) of the DDA provides as follows:

'disability' in relation to a person means –

- (a) total or partial loss of the person's bodily or mental functions; or
- (b) total or partial loss of a part of the body; or
- (c) the presence in the body of organisms causing disease or illness; or
- (d) the presence in the body of organisms capable of causing disease or illness; or
- (e) the malfunction, malformation or disfigurement of a part of the person's body; or
- (f) disorder or malfunction that results in the person learning differently from a person without the disorder or malfunction; or
- (g) a disorder, illness or disease that affects a person's thought processes, perception of reality, emotions or judgement or that results in disturbed behaviour;

and includes a disability that:

- (h) presently exists; or
- (i) previously existed but no longer exists; or
- (j) may exist in the future; or
- (k) is imputed to a person.

9.72 The term ‘disability’ is defined to a high level of specificity in the DDA. For example, para (d) (the presence in the body of organisms capable of causing disease or illness) was included in the definition to make clear that asymptomatic conditions such as HIV were covered. The term ‘impairment’ is also defined in the regulations made under the HREOC Act but the term ‘physical or mental disability’ is not defined in the WRA.

9.73 There is little doubt that the existing definition of disability in s 4 of the DDA covers genetic conditions that are manifested by current symptoms. Such symptoms may result, for example, in the partial loss of a person’s bodily or mental functions (para (a)) or in the malfunction of a part of a person’s body (para (e)). Under these paragraphs it is not necessary to consider the cause of the disability, only the effect on the individual.

9.74 The more problematic issue is whether the definitions in the DDA and in other anti-discrimination legislation are wide enough to address discrimination on the basis of genetic status where a person is presently asymptomatic. The DDA specifically covers disabilities that ‘may exist in the future’ or are ‘imputed to a person’, as well as past or present disabilities. The legislation in New South Wales and Tasmania is similar to the DDA in this respect.⁴¹ However, not all Australian legislation has such wide coverage.

9.75 The Northern Territory legislation contains an inclusive definition of impairment but does not expressly refer to future impairments.⁴² It may be open to a court to find that impairments arising in the future fall within this definition. In Western Australia, South Australia and the Australian Capital Territory, the legislation includes impairments imputed to a person and the imputation is not limited to past or present impairments.⁴³ It is also possible that a court may interpret these provisions to include future impairments. In other States the legislation has exclusive definitions of disability or impairment, which do not refer to future or imputed disabilities or impairments.

9.76 The definition of disability in the DDA is divided into two parts—the physical description of what amounts to a disability is set out in para (a)–(g), while some of the circumstances in which disabilities will be recognised for the purposes of

41 *Anti-Discrimination Act 1977* (NSW) s 49A; *Anti-Discrimination Act 1998* (Tas) s 3.

42 *Anti-Discrimination Act 1992* (NT) s 4(1), which states that ‘impairment ... includes ...’.

43 *Equal Opportunity Act 1984* (WA) s 4(1); *Equal Opportunity Act 1984* (SA) s 66(a); *Discrimination Act 1991* (ACT) s 7(2).

the Act are set out in para (h)–(k). These circumstances include present, past, possible future and imputed disabilities. The difficulty with the definition is that para (h)–(k) must relate to a type of physical or mental manifestation in the terms of para (a)–(g) of the definition.

9.77 Is a genetic mutation that increases a person's risk of heart disease, for example, an 'organism capable of causing disease or illness' (para (d))? Is it a 'malfunction, malformation or disfigurement of a part of the person's body' (para (e))? While it might be possible to argue that a genetic mutation is a malformation of part of a person's body, it seems clear that these provisions were not drafted with this issue in mind and that genetic mutations of this sort do not fit neatly into the existing terminology.

9.78 It is more likely that discrimination on the basis of a genetic mutation that increases the risk of a person developing a particular disorder is covered by para (j) of the definition of disability, coupled with para (a), (b) or (e). To take the case of a genetic mutation that increases the risk of heart disease, under the DDA the 'disability' does not arise directly because of the person's present genetic mutation, but because that mutation indicates that a 'partial loss of the person's bodily functions' (para (a)) 'may exist in the future' (para (j)). In short, the disability is not the genetic mutation itself but the possible future expression of that mutation through the malfunctioning of a part of the person's body.

9.79 The terms 'impairment' and 'disability' are also used in the regulations made under the HREOC Act and in the WRA. Both pieces of legislation, which are discussed in more detail in the employment context in Chapter 30, use general language such as mental, intellectual or psychiatric disability and physical disability without defining these terms. The *Human Rights and Equal Opportunity Commission Regulations 1989* (Cth) (HREOC Regulations) do, however, include a definition of impairment and expressly cover past and imputed disabilities. The WRA does not. Neither piece of legislation specifically includes possible future disabilities.

Submissions and consultations

9.80 A number of submissions suggested that there was no need to amend the existing definition in the DDA. The Australian Chamber of Commerce and Industry, for example, expressed the view that any change would be premature.⁴⁴ The Law Society of NSW,⁴⁵ the acting Disability Discrimination Commissioner⁴⁶ and the Anti-Discrimination Board of NSW submitted that the existing definition in the DDA is likely to cover discrimination on the basis of genetic status.⁴⁷ The Anti-Discrimination Board of NSW stated in its submission that:

44 Australian Chamber of Commerce and Industry, *Submission G170*, 2 March 2002.

45 Law Society of New South Wales, *Submission G285*, 18 December 2002.

46 Acting Disability Discrimination Commissioner - Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

47 For the position under the *Americans with Disabilities Act* (US), see P Miller, 'Is There a Pink Slip in My Genes? Genetic Discrimination in the Workplace' (2000) 3 *Journal of Health Care Law & Policy* 225, 240.

In our view the definition of disability in both the DDA and the ADA already adequately covers discrimination on the ground of a person's genetic make up. Given the breadth of the definition of disability in the DDA, we cannot conceive of a condition or predisposition to a condition discernible by genetic testing which would not fall within the current definition.⁴⁸

9.81 While the Law Society did not support amending the definition, the acting Disability Discrimination Commissioner, the Anti-Discrimination Board of NSW and a significant number of other submissions including the Genetic Support Council of Western Australia,⁴⁹ the Australian Medical Association,⁵⁰ the Australian Council of Trade Unions,⁵¹ the Human Genetics Society of Australasia,⁵² the Centre for Genetics Education,⁵³ the Investment and Financial Services Association⁵⁴ and the Queensland Government⁵⁵ expressed the view that this should be put beyond doubt. The Anti-Discrimination Board of NSW set out the benefits of clarification:

Although the ADB considers the definition of disability in the ADA and DDA covers genetic discrimination, there is a strong public interest rationale for making such coverage explicit in anti-discrimination legislation. Such clarification would:

- reflect the current state of the law under the DDA and ADA;
- have an educative effect;
- serve a symbolic function in clarifying that such discrimination is unlawful conduct under anti-discrimination law; and
- provide certainty regarding people's rights and responsibilities under anti-discrimination law.⁵⁶

9.82 The Centre for Law and Genetics expressed the view that, not only should the definitions in the DDA and the regulations made under the HREOC Act be amended to clarify that they include genetic status, the regulations should be brought into line more generally with the definition of disability in the DDA. The Centre questioned the use of the different terms 'disability' and 'impairment' and suggested that the same terminology should be used in both pieces of legislation. The acting Disability Discrimination Commissioner noted that differences between the two provisions had the potential to give rise to confusion.

48 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

49 Genetic Support Council WA, *Submission G243*, 19 December 2002.

50 Australian Medical Association, *Submission G091*, 29 January 2002.

51 Australian Council of Trade Unions, *Submission G037*, 14 January 2002.

52 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

53 Centre for Genetics Education, *Submission G232*, 18 December 2002.

54 Investment and Financial Services Association, *Submission G244*, 19 December 2002.

55 Queensland Government, *Submission G274*, 18 December 2002.

56 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

9.83 Most submissions that addressed this issue were also supportive of amending the definition of impairment in the regulations so that the HREOC Act applies to discrimination on the ground of a disability that may exist in the future.⁵⁷ The acting Disability Discrimination Commissioner noted that any amendment to the grounds of discrimination set out in the regulations may require consultation with representative employers' and workers' organisations under Article 1(b) of the International Labour Organization Convention 111 on Discrimination in Employment and Occupation 1958.

9.84 The Department of Employment and Workplace Relations noted that the Australian Industrial Relations Commission is required by s 93 of the WRA to have regard to the principles embodied in the DDA in the exercise of its functions. The Department did not support inserting a definition into the WRA on the basis that it would limit the flexibility of the courts to consider this issue on a case-by-case basis.⁵⁸ The Anti-Discrimination Commission of Queensland suggested, however, that the WRA should be amended so that the term 'disability' is defined by reference to the DDA to ensure consistency.⁵⁹ The acting Disability Discrimination Commissioner was also of this view.⁶⁰

Options for reform

9.85 A range of approaches to clarifying the definition of 'disability' in the DDA were put forward in submissions, including a return to the use of simple, undefined terms; the addition of paragraphs to the existing definition; and the inclusion of a descriptive preface to the definition.

9.86 ACROD Ltd was supportive of a return to a simpler definition of 'disability' on the basis that this would bring greater transparency and predictability.⁶¹ It is not clear, however, that this would assist in clarifying the operation of federal anti-discrimination law. Indeed, there is a danger that under the WRA, in particular, as currently drafted, it would be difficult to bring an action for unlawful termination on the basis of genetic status indicating an increased risk of future ill health.

9.87 The Anti-Discrimination Board of NSW, while holding the view that the existing definitions in the DDA and the *Anti-Discrimination Act 1977* (NSW) were wide enough to cover genetic status, went on to suggest in its submission to the Inquiry:

That the definition of disability in the DDA and all State/Territory anti-discrimination legislation be amended to make clear that disability includes genetic mutations or chromosome abnormalities:

57 Including Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Acting Disability Discrimination Commissioner - Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

58 Commonwealth Department of Employment & Workplace Relations, *Submission G305*, 22 January 2003.

59 Anti-Discrimination Commission Queensland, *Submission G214*, 2 December 2002.

60 Acting Disability Discrimination Commissioner - Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

61 ACROD Limited, *Submission G239*, 19 December 2002.

- causing or capable of causing disease, illness, malfunction, malformation or disfigurement of a part of the person's body, or
- resulting in the person learning differently from a person without the disorder or malfunction, or
- affecting a person's thought processes, perception of reality, emotions or judgment or that results in disturbed behaviour.⁶²

9.88 This suggestion makes clear that not all genetic variation should be covered by the DDA. Many genetic variations result in differences that could not be described as disabilities, such as eye or hair colour. Some genetic variations that can be described as abnormal do not result in any disabling difference and indeed may result in an individual being placed at an advantage in relation to the rest of the community. Some genetic mutations, for example, appear to confer increased immunity to particular diseases. The object of the DDA is not to protect those who are different, because we all differ from each other, but to protect those who are, or who are perceived to be, disabled.

9.89 The Anti-Discrimination Commission of Queensland made the following suggestion:

paragraph (ga) could be added to the definition of "disability" in section 4 of the DDA, along the lines of the following:-

"(ga) the person's genetic status as defined in section 4A of this Act."

Section 4A could then define "genetic status" with reference to a person's genetic predisposition to developing one of the conditions set out in the definition of "disability" in section 4 .

"4A "genetic status" means variations in a person's DNA, RNA, genes or chromosomes that predispose that person to developing: -

- *total or partial loss of the person's bodily or mental functions; or*
- *total or partial loss of a part of the body; or*
- *the malfunction, malformation or disfigurement of a part of the person's body; or*
- *a disorder or malfunction that results in the person learning differently from a person without the disorder or malfunction; or*
- *a disorder, illness or disease that affects a person's thought processes, perception of reality, emotions or judgement or that results in disturbed behaviour."*⁶³

9.90 Another possible approach is adopted in the preface to the definition of 'disability' in s 3 of the *Anti-Discrimination Act 1998* (Tas) which provides that:

⁶² Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

⁶³ Anti-Discrimination Commission Queensland, *Submission G214*, 2 December 2002.

"disability" means any of the following that presently exists, previously existed but no longer exists, may exist in the future, whether or not arising from an illness, disease or injury or from a condition subsisting at birth.

It could be argued that 'a condition subsisting at birth' includes a person's genetic status, although this is not express.

9.91 A number of submissions suggested that the definition should be amended in consultation with genetic scientists,⁶⁴ genetic support groups,⁶⁵ the Australian Institute of Health and Welfare,⁶⁶ the Investment and Financial Services Association⁶⁷ and other relevant stakeholders.⁶⁸

Inquiry's views

9.92 The Inquiry considers that there is value in providing greater certainty and raising awareness in relation to the issue of genetic discrimination. There is a possibility that the existing definition in the DDA will be construed narrowly by the courts so as to exclude predictive genetic information. The Inquiry is of the view that there is no policy justification for excluding discrimination based on possible future genetic conditions from coverage by the DDA. As well as having an educative effect, an appropriate amendment would put the matter beyond doubt and would ensure that the question did not need to be tested in the courts. The Inquiry recommends, therefore, that the definition of disability in the DDA be amended to specifically include genetic status.

9.93 The definition of impairment in the regulations made under the HREOC Act should also be amended to make clear that the HREOC Act applies to discrimination on the ground of genetic status. The Inquiry also supports amendments that would make the definitions in the DDA and the HREOC Regulations consistent in a more general sense. The Inquiry is of the view that the term 'disability' in the WRA should be expressly defined by reference to the definition in the DDA. This would be consistent with the policy underpinning s 93 of the WRA, discussed above, and would not, in the Inquiry's view, unduly limit the discretion of the courts.

9.94 The Inquiry does not propose to recommend a specific form of words for the amendments but is of the view that they should be developed in consultation with stakeholders, including the Human Genetics Commission of Australia, to ensure that the language is clear, appropriate and comprehensive. The Inquiry notes that, in developing a form of words, it will be important to limit the definition to those aspects of genetic status that are associated with a past, present, future or imputed disability.

64 Ibid.

65 Centre for Genetics Education, *Submission G232*, 18 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

66 ACROD Limited, *Submission G239*, 19 December 2002.

67 Investment and Financial Services Association, *Submission G244*, 19 December 2002.

68 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

Recommendation 9–3. In order to provide a consistent approach to addressing discrimination on the basis of genetic status, the Commonwealth, in consultation with the Human Genetics Commission of Australia and other stakeholders, should:

- amend the definitions of ‘disability’ in the DDA and ‘impairment’ in the regulations made under the *Human Rights and Equal Opportunity Commission Act 1986* (Cth) (HREOC Act) to clarify that the legislation applies to discrimination based on genetic status;
- amend the definition of ‘impairment’ in the regulations made under the HREOC Act to clarify the application of the legislation to a disability that may exist in the future; and
- define ‘disability’ in the *Workplace Relations Act 1996* (Cth) by reference to the definition of ‘disability’ in the DDA.

Medical records

9.95 The HREOC Act, as well as the Tasmanian and the Northern Territory anti-discrimination legislation, includes provisions relating to discrimination on the basis of medical records. Under the HREOC Act it is possible to lodge a complaint of discrimination in employment on the basis of a distinction, exclusion or preference made on the ground of a medical record. As noted above, the complaint procedures under these provisions do not give rise to a legally enforceable remedy. Section 16 of the Tasmanian anti-discrimination legislation and s 19 of the Northern Territory legislation provide that a person must not discriminate on the basis of an irrelevant medical record.⁶⁹ DP 66 sought feedback on whether the DDA and other relevant legislation should be amended to include discrimination on the ground of medical record.

Submissions and consultations

9.96 A number of submissions expressed support for including medical records as a further ground of discrimination in the DDA.⁷⁰ No examples were provided, however, of situations in which this would extend the protection offered by the existing provisions.

⁶⁹ *Anti-Discrimination Act 1998* (Tas); *Anti-Discrimination Act 1992* (NT).

⁷⁰ National Council of Women Australia, *Submission G095*, 31 January 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Anti-Discrimination Commission Queensland, *Submission G214*, 2 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

9.97 The Anti-Discrimination Board of NSW, on the other hand, expressed the view that:

The ADB does not consider that the prohibition in relation to discrimination on the ground of ‘irrelevant medical record’ in Tasmanian and Northern Territory anti-discrimination legislation adds anything additional to that which is already covered by the prohibition of discrimination on the ground of disability, combined with adequate provisions in relation to unlawful questions and requests for information and privacy protection in relation to health information.⁷¹

9.98 In consultation, the Attorney-General’s Department noted that ‘medical record’ would sit oddly with the other elements of the definition of disability and that an amendment of this kind would move the focus of the legislation away from the attributes of the individual to the form in which those attributes are recorded.⁷² Several submissions expressed the view that the legislation should not be amended if there was no demonstrated need.⁷³

Inquiry’s views

9.99 The Inquiry has not been able to identify any situations in which discrimination on the ground of a person’s genetic medical record would not be covered by the existing provisions in relation to discrimination on the ground of disability, amended in accordance with the recommendations in this Report. For example, discrimination on the ground of an accurate medical record of a genetic condition or predisposition would be covered by discrimination on the ground of existing or possible future disability. Discrimination on the ground of an incorrect medical record would be covered by imputed disability. On the basis that such an amendment would not extend the protection provided by existing anti-discrimination legislation, the Inquiry does not recommend that this amendment be made.

Associates

9.100 Because of the familial nature of genetic information, it is possible that individuals will be discriminated against, not on the basis of information about themselves, but on the basis of information about their genetic relatives. Employers may seek to rely on the fact that genetic information about a member of a person’s family may sometimes provide relevant information about the person. An employer may, for example, refuse to employ an applicant because of a family history of breast cancer. In the anti-discrimination context, this act may be characterised either as (a) an act on the basis of the applicant’s ‘association’ with others, which is discussed below, or (b) an act on the basis of an ‘imputed’ disability, which has been discussed above.

71 Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002.

72 Attorney-General’s Department, *Consultation*, Canberra, 22 November 2002.

73 Centre for Law and Genetics, *Submission G255*, 21 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

9.101 Anti-discrimination legislation in Australia generally recognises that it is unlawful to discriminate against a person on the basis of his or her association with another person.⁷⁴ For example, s 15 of the DDA provides in part as follows:

(1) It is unlawful for an employer or a person acting or purporting to act on behalf of an employer to discriminate against a person on the ground of the other person's disability or a disability of any of that other person's associates:

- (a) in the arrangements made for the purpose of determining who should be offered employment; or
- (b) in determining who should be offered employment; or
- (c) in the terms or conditions on which employment is offered.

9.102 The 'associate' qualification is repeated in each provision setting out an area of activity in which discrimination on the ground of disability is prohibited.

9.103 Associate is defined in s 4 of the DDA as follows:

associate, in relation to a person, includes:

- (a) a spouse of the person; and
- (b) another person who is living with the person on a genuine domestic basis; and
- (c) a relative of the person; and
- (d) a carer of the person; and
- (e) another person who is in a business, sporting or recreational relationship with the person.

relative, in relation to a person, means a person who is related to the first-mentioned person by blood, marriage, affinity or adoption.

9.104 In relation to genetic status, the most relevant elements are para (c) of the definition of 'associate' (a relative of the person), and the definition of 'relative.' In combination, these provisions appear to be wide enough to cover those who are discriminated against on the basis of the genetic status of their relatives. This would extend to the situation where families comprise members who are not genetically related, such as adopted children, stepchildren, or children born through artificial reproductive technology using donor gametes.

9.105 Some state and territory legislation deals with the issue slightly differently by including 'association with a person' as a further ground of discrimination. For example, s 6 of the *Equal Opportunity Act 1995* (Vic) provides:

⁷⁴ *Anti-Discrimination Act 1977* (NSW) s 49B; *Equal Opportunity Act 1995* (Vic) s 6; *Anti-Discrimination Act 1991* (Qld) s 7; *Equal Opportunity Act 1984* (WA) s 66A; *Anti-Discrimination Act 1998* (Tas) s 16; *Anti-Discrimination Act 1992* (NT) s 19; *Discrimination Act 1991* (ACT) s 7(1).

The following are the attributes on the basis of which discrimination is prohibited in the areas of activity set out in Part 3 ...

- (b) impairment; ...
- (m) personal association (whether as a relative or otherwise) with a person who is identified by reference to any of the above attributes.

9.106 The HREOC Act and the WRA do not expressly address discrimination on the basis of association with another person.⁷⁵ The language of the HREOC Act and the WRA is potentially wide enough to cover discrimination on the basis of personal association but the issue is unclear.

9.107 Provisions on imputed disability will often cover cases of genetic discrimination where a person who does not have a disability is discriminated against on the basis of his or her association with a person who does have a disability. But this is not always the case. In *IW v City of Perth*⁷⁶ the High Court indicated that problems might arise where the relevant legislation does not cover discrimination on the basis of association. On the facts of that case it would not have been possible to rely on imputed disability because the 'person' alleging discrimination was an organisation, which could not itself suffer a disability.

9.108 In DP 66 the Inquiry proposed, therefore, that the regulations made under the HREOC Act should be amended expressly to include discrimination on the basis of association with a person who has an impairment or disability.

Submissions and consultations

9.109 Most submissions that addressed this issue supported the proposed amendment to the HREOC regulations. The Anti-Discrimination Board of NSW expressed the view that, while coverage under the DDA and most state and territory legislation was adequate:

Precisely because the genetic information obtained from one person may be indicative of the genetic make up of that person's blood relatives, it is essential that all State/Territory anti-discrimination legislation covers such circumstances.⁷⁷

9.110 The acting Disability Discrimination Commissioner, while supporting the change to the HREOC regulations, raised the following concern in relation to the DDA associate provisions:

As noted above I would support HREOCA coverage being brought into line with that under the DDA. However, in this context attention is required to how the DDA currently deals with associates.

The difficulty is that while the substantive sections dealing with unlawful discrimination address discrimination against associates as well as against people with a disability, the definitions of direct and indirect discrimination in sections 5 and 6 refer only to a disability of the aggrieved person.

⁷⁵ The same is true of the *Equal Opportunity Act 1984* (SA).

⁷⁶ *IW v City of Perth* (1997) 191 CLR 1.

⁷⁷ Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

At present (consistent with accepted rules of statutory construction) HREOC seeks to interpret and apply the DDA in a way which gives effect to the substantive provisions regarding associates rather than rendering them meaningless. It would be preferable however for the definitions of discrimination to expressly include associates rather than leaving this to interpretation.⁷⁸

Inquiry's views

9.111 In the Inquiry's view there is merit in amending the regulations made under the HREOC Act to include discrimination on the basis of association. This would bring the HREOC Act into line with the DDA in this regard.

9.112 The Inquiry notes the concern of the acting Disability Discrimination Commissioner in relation to the operation of the existing associate provisions in the DDA. In light of these concerns, the Inquiry would support a more comprehensive review of these provisions by the Commonwealth. However, given the Inquiry's Terms of Reference, the recommendation below is limited to addressing the gap in coverage under the HREOC Act.

Recommendation 9-4. The Commonwealth should amend the definition of 'impairment' in the regulations made under the HREOC Act to include discrimination on the basis of association with a person who has an impairment or disability.

Harmonisation of state and territory law

9.113 As noted above, federal, state and territory anti-discrimination legislation, while based on the same paradigm or framework, differs in significant ways including in relation to the way that 'disability' or 'impairment' is defined, and in relation to discrimination on the basis of association.

Submissions and consultations

9.114 Submissions received by the Inquiry generally expressed strong support for uniformity or greater harmonisation of federal, state and territory laws concerning discrimination in relation to human genetic information.⁷⁹ The Disability Discrimination Legal Service sounded a note of caution, however, in relation to

⁷⁸ Acting Disability Discrimination Commissioner - Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

⁷⁹ Including Anti-Discrimination Board of NSW, *Submission G194*, 27 November 2002; Anti-Discrimination Commission Queensland, *Submission G214*, 2 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Investment and Financial Services Association, *Submission G244*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

maintaining the ability of States and Territories to act independently⁸⁰ and the Queensland Government expressed concern that standards were not raised or lowered contrary to the policy of particular jurisdictions.⁸¹

9.115 On the other hand, the Human Genetics Society of Australasia noted that family members affected by genetic disorders may live in a number of States and Territories.⁸² The Australian Society for Medical Research expressed the view that greater national uniformity would benefit the public as well as the scientific community.⁸³ The Australian Institute of Actuaries noted that companies often operate across state borders and that it was confusing for such companies to be subject to different regimes in each jurisdiction.⁸⁴ The Inquiry considers that such inconsistencies may provide a disincentive for organisations to develop policies and programs that comply with anti-discrimination legislation because the applicable rules are unclear and may differ from one jurisdiction to another.

9.116 The Anti-Discrimination Board of NSW summarised the issue as follows:

Uniformity or, at a minimum, greater harmonisation of federal, State and Territory anti-discrimination legislation is crucial to an effective legislative regime to provide protection against genetic discrimination. It would ensure that people are afforded equal protection under the Australian law, regardless of which State or Territory people reside [in] and where the conduct occurs within Australia. Uniformity would reduce the complexity of jurisdictional decisions about whether to proceed under State/Territory or federal legislation for the would-be complainants. It also supports greater certainty about people's rights and responsibilities under anti-discrimination law, rather than such understanding being undermined by uncertainty which arises when there are inconsistencies between different federal, State and Territory laws. Uniformity of anti-discrimination legislation would enhance certainty by increasing the likelihood that case law from one jurisdiction is applicable in another and for precedent to be applied.⁸⁵

9.117 In addition, the acting Disability Discrimination Commissioner stated that:

In this area and more generally, HREOC supports Federal and State anti-discrimination laws taking a consistent approach wherever possible. For State laws to provide more restricted coverage than the DDA can serve only to mislead or confuse employers, service providers and others covered by the legislation regarding the extent of their obligations, since responsibilities under the DDA will apply in any case; and may cause procedural problems for complainants if they choose the wrong jurisdiction. Most States and Territories have in fact moved to harmonise their legislation with the DDA. This process should continue, through action by individual jurisdictions and preferably also through co-ordinated action through relevant Ministerial councils.⁸⁶

80 Disability Discrimination Legal Service, *Submission G146*, 28 March 2002.

81 Queensland Government, *Submission G274*, 18 December 2002.

82 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

83 Australian Society for Medical Research, *Submission G124*, 18 March 2002.

84 Institute of Actuaries of Australia, *Submission G105*, 7 March 2002.

85 Anti-Discrimination Board of NSW, *Submission G157*, 1 May 2002.

86 Acting Disability Discrimination Commissioner — Human Rights and Equal Opportunity Commission, *Submission G301*, 16 January 2003.

9.118 In consultation, HREOC noted that the heads of the various anti-discrimination bodies in Australia meet regularly to discuss issues such as harmonisation and that this was one mechanism through which this goal could be pursued.⁸⁷

Inquiry's views

9.119 The Inquiry is of the view that greater harmonisation across jurisdictions, for example, in relation to the definition of disability or impairment and the coverage of discrimination on the ground of association would be beneficial. Harmonisation is not intended to stifle the role of the States and Territories in innovation and experimentation, but to ensure that similar approaches are adopted where similar goals exist.

Recommendation 9-5. The States and Territories should consider harmonising their anti-discrimination legislation, and other relevant laws, in a manner consistent with the recommendations in this Report.

87 Human Rights and Equal Opportunity Commission, *Consultation*, Sydney, 18 November 2002.

Part C. Genetic Testing

10. Genetic Testing

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What is genetic testing?

10.1 Genetic information may come from many sources. These sources include a person's family medical history, a clinical examination that diagnoses a genetic disorder, or a scientific test.

10.2 Scientific tests that reveal genetic information are of varying types.

- Some genetic tests (here called DNA tests) directly analyse DNA or RNA. For example, testing of a genetic sequence may be undertaken by targeting a segment of DNA or RNA using a process known as polymerase chain reaction (PCR). This process, which was developed through the pioneering work of Kary Mullis and others at the Cetus Corporation in California in 1985 and led to the award of a Nobel Prize, enables the DNA from a single cell to be reproduced in large amounts (that is, amplified) for testing.¹
- Some tests do not analyse DNA or RNA material directly but test the biological products of particular genes. The measurement of certain proteins produced by genes, or certain metabolites, may reveal valuable information about gene function itself. For example, since the 1960s newborn screening programs have tested for phenylketonuria, galactosaemia and other genetic disorders. Some of these tests do not involve PCR analysis but still produce valuable genetic information.

1 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 19.

- Some routine biochemical tests of non-genetic substances may also reveal genetic information. For example, a positive test for high cholesterol or occult faecal blood may be the consequence of mutations in the genes conferring susceptibility to heart disease or colon cancer, respectively.² In an appropriate clinical setting, results of these biochemical tests may provide strong indicators of particular genetic disorders.
- Some genetic diagnoses are made on the basis of the morphological characteristics of certain cells, tissues or at postmortem examination. Examples include the microscopic appearance of red blood cells in thalassaemia; the characteristic electro-microscopic appearances of the skin in epidermolysis bullosa, or the constellation of signs identified at postmortem in a foetus with skeletal dysplasia.³
- Finally, some medical imaging processes reveal important genetic information. For example, nuchal translucency is an imaging procedure that is sometimes performed on a human foetus in utero. By measuring the presence of abnormal swelling under the skin at the back of the foetus's neck, clinicians can predict a likelihood that the baby will have the genetic condition Down Syndrome.

10.3 During the course of the Inquiry different views emerged as to which of the five types of tests described above should properly be regarded as genetic tests. There was general agreement that the first and second categories (namely, the direct testing of genetic material and the testing of the biological products of genes) are appropriately described in this way. However, the status of the other three categories was contested. Some people appeared content to describe these tests as genetic tests; others took the view that they are non-genetic tests that may reveal genetic information.

10.4 The difference in description may assume importance in situations in which particular regulatory consequences flow from the classification of a test as genetic or non-genetic. For example, the peak body representing life insurers in Australia, the Investment and Financial Services Association, has developed a genetic testing policy for the industry, which regulates the use of genetic tests in underwriting life insurance products. For the purpose of the policy, a 'genetic test' is defined to mean

the direct analysis of DNA, RNA, genes or chromosomes for the purpose of determining inherited predisposition to a particular disease or group of diseases, but excluding DNA, RNA, gene or chromosome tests for acquired disease.⁴

10.5 This narrow definition corresponds most closely with the first category of tests described above. The effect is to leave a broad class of other tests, which are not covered by the industry policy and may therefore be dealt with in the traditional, and more liberal, fashion, despite the fact that they may reveal genetic information.

2 J Beckwith and J Alper, 'Reconsidering Genetic Anti-discrimination Legislation' (1998) 26 *Journal of Law, Medicine and Ethics* 205, 207.

3 M Buckley, *Correspondence*, 15 November 2002.

4 See Investment and Financial Services Association, *IFSA Standard 11.00 'Genetic Testing Policy'* (2002), IFSA.

10.6 In many circumstances, however, this Inquiry is not concerned with the description of a particular test as genetic or otherwise. Rather it is concerned with the protection of genetic information, however it may be derived. For this reason, the Inquiry does not consider it necessary to propose a comprehensive definition of what a genetic test is. The Inquiry acknowledges that the direct testing of genetic samples and the testing of the biological products of genes form part of the core conception of genetic testing for the purpose of the Inquiry. Beyond this, much will depend on the context in which the question is asked. It is unlikely that the same answers will be given in the areas of medical research, clinical diagnosis, criminal investigation, or the many other circumstances in which genetic information may be sought and used.

Purposes of genetic tests

10.7 Genetic testing can be divided into three broad categories based on the purpose of the testing, namely, medical testing, identification testing and kinship testing. These categories are not fixed: genetic testing sought for one purpose can sometimes reveal unintended information, such as where medical testing reveals incidental information about parentage or lack of parentage.

10.8 There are numerous reasons for seeking a genetic test. Although the description given to a test varies according to context and user, the following are the most common types of genetic tests referred to in this Report.

- **Diagnostic testing** is performed to make or confirm a diagnosis of a specific disorder in a person who generally already has signs or symptoms of that disorder.
- **Predictive or presymptomatic testing** is performed on a person who generally has no signs or symptoms of a specific disorder at the time of testing, in order to determine whether or not that person has genetic variations that increase the likelihood that the person may, or will, develop the disorder in the future. Predictive testing is often performed in relation to genetic disorders that are not evident at birth but have their onset during adulthood. Predictive testing is also called presymptomatic testing where an individual's family medical history suggests that he or she may have the genetic disorder but symptoms of it are not yet manifest.
- **Genetic carrier testing** is performed on a person to determine whether or not that person has a genetic or chromosomal abnormality that does not generally affect the person's health but increases his or her chance of having children with the disorder in question.⁵ The outcome of such testing can influence future reproductive decisions.

5 Individuals sometimes experience mild health effects as a result of being carriers of the relevant mutation.

- **Screening testing** is performed on people who are not necessarily known to be at increased risk of a particular genetic disorder. Screening tests can be conducted on individuals, groups and entire populations, such as with postnatal screening using newborn screening cards. Cascade screening is a form of screening where once an affected individual is identified, potentially affected relatives are systematically identified and offered testing. In some cases screening testing is an extension of genetic carrier testing since it allows asymptomatic individuals in the community to be tested to see if they carry a genetic variation or mutation. This information may be helpful in family planning or in facilitating preventive measures to avoid disease.⁶
- **Pre-implantation and prenatal testing** is performed on a foetus in utero (or pre-implantation in the case of embryos used in artificial reproductive technology procedures). Prenatal genetic testing is typically performed where there are 'at risk' parents, such as parents who are carriers of mutated alleles for cystic fibrosis, Tay-Sachs disease, or β -thalassaemia. In the longer term, early detection through prenatal diagnosis may permit use of therapies such as blood transfusion, surgical correction or, possibly, gene therapy.
- **Research testing** involves the systematic analysis of genetic information to advance medical or scientific knowledge about how genes influence the health of individuals and populations. Genetic research testing may be conducted on identified or de-identified samples and generally requires the approval of a Human Research Ethics Committee, which oversee issues of consent and other ethical concerns.⁷
- **Identification testing or forensic testing** is currently performed on non-coding DNA, with respect to a number of agreed core loci, to construct a DNA profile. This profile is then matched against the profile from another DNA sample, for example, from a crime scene. This identification process is used in criminal investigations to exclude or identify a suspect, in searches for missing persons, and in the identification of deceased persons.⁸
- **Parentage and other kinship testing** is performed to determine whether two people are biologically related to each other. Parentage testing is the most common form of kinship testing and may be done for various purposes, including in relation to family law proceedings, immigration applications and disaster victim identification.⁹

6 For example, the knowledge that someone carried the genetic defect for haemochromatosis might allow the relatively simple option of blood letting to prevent complications from the condition.

7 See Pt D.

8 See Pt J.

9 See Ch 35–37, 42.

Who seeks genetic testing and why?

10.9 Genetic testing is sought in various contexts for different purposes. The uses of genetic testing are likely to expand over time as the testing processes become easier to undertake and their practical uses become clearer. These processes are likely to be facilitated by the knowledge that is becoming available as a result of the Human Genome Project.

10.10 The principal current users of genetic testing are identified below.

- **Medical practitioners** use genetic testing to diagnose patients for treatment as well as for predictive, presymptomatic, screening and prenatal purposes. Practitioners request the various types of genetic tests through request pathways, which may involve referral of the patient to a clinical geneticist and also pre-test and post-test counselling.
- **Medical researchers** use genetic testing to advance medical or scientific knowledge about how genes influence the health of individuals and populations. Genetic testing for research purposes may be conducted in concert with medical practitioners, who liaise with participating patients.
- **Individuals** generally cannot obtain direct access to clinical genetic testing by laboratories in Australia. Thus diagnostic, predictive, presymptomatic, genetic carrier, screening, pre-implantation and prenatal genetic testing must generally be sought through a medical practitioner. However, some laboratories offer kinship testing, particularly parentage testing, directly to individuals, and the range of testing available to the public is likely to expand in the future.
- **Police** use genetic testing in law enforcement primarily for the purpose of identification, such as to identify victims, deceased persons and suspects.
- **Lawyers and litigants** use genetic testing as evidence in criminal and civil cases. In criminal cases, genetic testing may be used to prosecute offenders, obtain acquittals, and to press for reversal of convictions on appeal. Litigants also use genetic testing in civil cases, for example, to establish parentage in family law or succession matters. In the future, genetic testing may also be used by litigants in negligence actions to establish or defend a claim.
- **Employers** may seek to use genetic testing to screen or monitor employees or job applicants. Although this type of testing is not common in Australia at present, overseas experience suggests that these uses are likely to expand in the future. Employers may seek to conduct genetic testing to reduce workers compensation claims, comply with occupational health and safety obligations, or increase productivity by screening out employees who are most likely to be absent from work due to illness. The testing may take the form of predictive or presymptomatic testing to identify whether an individual who is currently asymptomatic has a gene that increases the likelihood that he or she will develop

a disorder as a result of the workplace environment. Testing may also screen for genes or disorders that are unrelated to the workplace but which may render an individual undesirable to an employer.

- **Insurers** may use the results of genetic testing as a component of the underwriting process in applications for personal insurance, where health information is collected to assess the risk that applicants bring to the insurance pool. The testing may take the form of diagnostic, predictive or presymptomatic testing, particularly in relation to life insurance.
- **Government agencies** may use genetic testing to establish kinship or identification. For example, the Department of Immigration and Multicultural and Indigenous Affairs sometimes uses genetic testing to establish family linkage for the purposes of Australia's immigration programs; law enforcement agencies often use identification testing for the purposes of criminal investigation.

Who performs genetic testing?

10.11 Genetic testing is available from publicly or privately operated laboratories. These laboratories may be accredited or non-accredited. A national scheme of accreditation exists in Australia through the National Association of Testing Authorities, Australia (NATA). Accreditation is discussed further in Chapter 11.

Publicly operated genetic testing laboratories

10.12 Federal, state and territory governments operate genetic testing laboratories, which are usually attached to or affiliated with public hospitals or universities. In addition, some laboratories function within a particular government department that utilises genetic testing. Some publicly funded laboratories provide genetic testing services to both the government and private sectors. For example, the Australian Federal Police (AFP) has its own forensic services division, which performs DNA identification testing exclusively for the AFP, while in South Australia the Forensic Science Centre is government funded but provides testing services to government and the private sector.

10.13 Publicly funded genetic testing laboratories operate as service-orientated testing laboratories for government, community and health professionals, or as research-orientated testing laboratories, or both.

Privately operated genetic testing laboratories

10.14 Privately operated genetic testing laboratories are sometimes affiliated with private hospitals or universities, but many operate as independent profit-making entities. As with public laboratories, private genetic testing laboratories may offer service-orientated testing or research testing. Private laboratories sometimes also provide genetic testing services to government on an ad-hoc basis or under contract.

Access to genetic testing

10.15 Access to genetic testing in Australia is affected by a number of factors. These include the availability of genetic tests, the cost of testing (including the availability of public and private health insurance), the request pathways through which tests are ordered and laboratory protocols in relation to the performance of testing. These factors are discussed separately below.

Availability of genetic testing

10.16 The Human Genetics Society of Australasia (HGSA) maintains a register of medical genetic tests that are available in Australia and the laboratories that provide them. According to the HGSA, there are presently around 220 DNA diagnostic tests available from 44 laboratories across Australia.¹⁰

10.17 Some genetic tests offered overseas are not available in Australia. Likewise, some types of tests offered in Australia are not available, or not widely performed, in other countries. The availability of genetic testing in Australia is dependent not only on genetic testing technology, but on decisions about which tests are ethically acceptable, and on a cost-benefit analysis of a particular test. In addition, the availability of a genetic test in a particular laboratory may reflect the research interests of that laboratory. For example, a laboratory that undertakes research into a particular genetic disease might also offer, as part of its research work, a DNA diagnostic service for that disease.

10.18 The availability of genetic identification and kinship testing varies between states and between laboratories within a state. Identification and kinship testing is available in Australia from 21 NATA accredited laboratories and an unknown number of non-accredited laboratories for law enforcement, parentage, immigration and other purposes.

10.19 The availability of genetic testing is not limited to Australian laboratories. The marketing of genetic testing directly to the public through mail order and the Internet has facilitated access to genetic testing from overseas laboratories, sometimes at lower cost. The regulation of genetic testing provided directly to the public, including by service providers overseas, is discussed in Chapter 11.

Cost of genetic testing

10.20 Genetic testing is still a relatively slow and expensive process. However, technology is advancing rapidly. The development of automated 'DNA chip' technology may soon make it technically possible and financially practicable to conduct multiplex testing, in which screening is conducted for numerous genetic

10 J Brasch, *DNA Diagnosis of Genetic Disorders in Australasia*, Human Genetics Society of Australasia, <www.hgsa.com.au/labs.html>, 19 February 2003. Not all tests are available from all laboratories. The register does not include newborn screening, laboratories that test for cystic fibrosis or forensic testing laboratories used for identification testing.

mutations simultaneously in a single test procedure.¹¹ It may soon be the case that the genetic information available will outstrip the capacity of health systems to interpret it and counsel patients effectively.¹²

10.21 As with other health services, effective access to genetic testing depends on the cost to consumers of testing procedures and on the rebates provided by public and private health insurers. The cost of genetic testing procedures varies, from less than \$100 to more than \$1000, depending on a number of factors.¹³

- **Test methodology.** Low complexity tests (for example, single gene mutation) are less expensive than high complexity tests (for example, full gene sequencing).
- **Laboratory testing strategy.** Some laboratories test for a large number of mutations at once; others test in stepwise fashion, beginning with the most common mutations.
- **Number of individuals tested.** Several family members may need to be tested to obtain a meaningful test result.
- **Contractual agreements.** Private and public hospitals, insurers and laboratories negotiate contracts to set the price of testing and the amount of reimbursement.¹⁴
- **Specimen handling.** Some cell types require culturing or other special handling before testing.
- **Additional services.** Genetic consultation or counselling is usually recommended and sometimes required before genetic testing is performed. These fees should be considered in the total cost.
- **Intellectual property.** The enforceability of patents in relation to the human genome or genetic processes may substantially increase the costs of some tests. Patents provide temporary monopoly rights as a reward for innovation.

Medicare

10.22 Medicare is Australia's public health insurance scheme. Introduced in 1984, its stated objectives are to make health care affordable for all Australians, to give all Australians access to health care services with priority according to clinical need, and to provide a high quality of care.¹⁵

11 Also known as 'gene chips', 'biochips' and 'DNA microarrays'. See S Moore, 'Making Chips to Probe Genes', *IEEE Spectrum*, 1 March 2001, 54.

12 American Medical Association Council on Ethical and Judicial Affairs, 'Multiplex Genetic Testing' (1998) 28(4) *Hastings Center Report* 15.

13 See eg Children's Health System and University of Washington, *Gene Tests, Gene Clinics*, <www.genetests.org>, 20 February 2003.

14 See Medicare Benefits Schedule, Items 66794, 65132 and 65168.

15 See Medicare website: Commonwealth of Australia, *HIC*, <www.hic.gov.au/>, 20 February 2003.

10.23 Medicare currently funds a range of tests relevant to genetic disorders. These include cytogenetics,¹⁶ alpha fetoprotein and related biochemical markers, and ultrasound derived nuchal translucency. The Medicare Benefits Schedule (MBS) funds DNA genetic tests under four MBS items, which concern testing for haemochromatosis, Factor V Leiden, protein C or S deficiencies, and antithrombin 3 deficiency.¹⁷ DNA tests are Medicare funded under two categories:

- diagnostic testing to confirm a clinical observation (for example, in the case of haemochromatosis the patient must have raised iron levels); and
- screening of asymptomatic individuals where the patient is a first-degree genetic relative of another individual who is known to have the condition.

10.24 Under current arrangements, if medical practitioners wish to have a new test listed in the MBS, they must apply to the Medical Services Advisory Committee (MSAC). MSAC provides advice to the federal Minister for Health and Ageing about the strength of evidence relating to the safety, effectiveness and cost effectiveness of new and emerging medical services and technologies and under what circumstances public funding, including listing on the MBS, should be supported.

10.25 MSAC has an established process for assessing whether new medical technologies and procedures should be put on the MBS. For example, MSAC has recently assessed the genetic test for Fragile X syndrome, which had been proposed as an alternative to the currently funded chromosomal tests for this condition. On 20 August 2002, the Minister for Health and Ageing accepted MSAC's recommendation that public funding should be provided for the use of Fragile X testing.¹⁸

Private health insurance

10.26 Private health insurance currently provides very limited coverage for genetic testing. Private health insurers do not subsidise testing sought in outpatient services. They generally provide a subsidy only if genetic testing is required as part of a patient's treatment regime when admitted into hospital and only if MBS also provides a rebate for the test.

16 MBS cytogenetic testing for genetic abnormalities allows for chromosome studies in tissue, fluid or blood and relates to diverse clinical areas such as malignancy, prenatal diagnosis, infertility and constitutive chromosome abnormalities: Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

17 Department of Health and Ageing, *Medicare Benefits Schedule (MBS)*, Department of Health and Ageing, <www.health.gov.au/pubs/mbs/index.htm>, 3 February 2003.

18 Medicare Services Advisory Committee, *Genetic Test for Fragile X Syndrome: Assessment Report*, Department of Health and Ageing, <www.health.gov.au/msac/pdfs/msac1035.pdf>, 20 February 2003, 38.

Request pathways

10.27 A request pathway describes the steps that must be taken in order to obtain a genetic test, including referrals for testing and genetic counselling. The request pathway for genetic testing varies according to the type of testing sought, the laboratory from which the test is sought, and the health professional who requests it.

10.28 Medical genetic testing usually requires referral from a medical practitioner as well as pre-test and post-test counselling.¹⁹ The request pathway for kinship testing is less regulated. Generally it is available without referral or counselling—in some cases it is available without the consent of the person being tested. The request pathways for genetic testing form part of broader issues relating to the regulation of genetic testing, aspects of which are considered in Chapters 11 and 23.

Reliability of genetic testing

10.29 Some genetic tests are not entirely reliable for a number of reasons, both technical and non-technical. The stigmatisation and discrimination that may flow from genetic testing are central concerns for this Inquiry, and these concerns become acute when a test is unreliable. Issues related to the reliability of genetic information, including the interpretation of test results, are considered in Chapters 3, 11 and 23. Scientific reliability of genetic testing—the scientific or technical reliability of the test—is discussed below.

Scientific reliability

10.30 The scientific reliability of a genetic test may be affected by a number of factors including sample contamination, incorrect laboratory testing procedures, mislabelling, and transcription errors. Although there has been considerable attention paid in recent years to developing policies in relation to the ethical and lawful use of genetic information, there has been less discussion about the impact of erroneous information.

10.31 Every laboratory testing procedure, no matter how well-established, involves the possibility of error. This is equally true of genetic testing. For example, the PCR method of DNA amplification allows minute quantities of DNA to be replicated in a way that facilitates testing. Yet there is a danger that the sample may be contaminated with extraneous genetic material, such as from previously amplified products or from the operator, thus generating copies of irrelevant DNA. There are also occasional errors with the sequence fidelity of amplified products, resulting in reading errors.²⁰

10.32 The scientific reliability of a genetic test is measured by the ‘sensitivity’ and ‘specificity’ of the test. These are technical terms but, in essence, refer respectively to the statistical likelihood that a ‘true-positive’ will return a positive test result and that a

19 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: an Information Paper* (2000), NHMRC, Canberra.

20 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 20.

‘true-negative’ will test negative. Clinicians and patients desire 100% accuracy. However, few laboratory tests are currently more than 98% sensitive and specific.²¹

10.33 Moreover, every test result requires individual interpretation, with a further opportunity for error to be introduced. Because genetic tests are considered to be ‘scientific’, many non-experts may invest excessive confidence in their significance and predictive value.²² As a result, a small number of people who take genetic tests will receive inaccurate information about their genetic status. Whether this involves the trauma of a false positive or the spurious re-assurance of a false negative, either type of error is likely to have crucial implications for the individual, who might plan his or her life on the basis of the test result. In addition, because members of a family share genes and DNA, any error in a genetic test may have long term implications both for the person tested and his or her family. Predictive tests are particularly vulnerable to this difficulty because of the likelihood of a long delay before the error is recognised.

Fraud

10.34 Another problem with the reliability of genetic testing procedures is the possibility of fraud. The Inquiry has received a number of submissions suggesting that laboratory protocols do not sufficiently protect against intentional interference with laboratory samples tested.²³ For example, a lawyer described a client’s allegation that an opposing party in family law proceedings had bribed a laboratory or otherwise falsified parentage test results.²⁴

10.35 As discussed in Chapter 11, the scientific reliability of genetic testing is regulated by accreditation standards, which are administered by NATA and other bodies. However, the accreditation of a laboratory in accordance with the best technical and scientific standards is no guarantee against intentional deception by its employees. Although specific examples of possible laboratory fraud have been brought to the Inquiry’s attention, the Inquiry has no evidence of the incidence of fraudulent testing in Australia. Nevertheless, the possibility of fraud identified in the above submission indicates that this matter is one of continuing concern.

21 R Linsk, *Consultation*, Sydney, 20 February 2001.

22 In the forensic context, see Ch 44.

23 N Turner, *Submission G083*, 14 January 2002; *Confidential Submission G074ACON*, 10 January 2002.

24 N Turner, *Submission G099*, 22 February 2002. See further Ch 35.

11. Regulating Access to Genetic Testing

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Introduction

11.1 This chapter focuses on a range of issues concerning access to genetic testing.¹ To date, medical practitioners have been the primary 'gatekeepers' of access to genetic testing and the genetic information derived from it, at least for clinical purposes. Genetic testing for medical purposes usually requires a referral from a medical practitioner. Medical practitioners may request various types of genetic test, which may in turn require referral to a medical specialist, such as a clinical geneticist, or to a genetic counsellor.² However, genetic testing products and services also may be provided by laboratories and others directly to the public.

11.2 The steps that individuals must take in order to obtain a genetic test vary according to the type of testing sought, the laboratory concerned, and whether there is a request from a health professional. The nature of these steps and the consequent ability of individuals to gain independent access to genetic testing may be constrained by laboratory practices and protocols, including those relating to accreditation

1 In the context of parentage and other kinship testing, see also Ch 35.

2 Other issues relating to the ordering of genetic tests by medical practitioners are discussed in Ch 23.

requirements, and by ethical and professional standards applying to health professionals.

11.3 The chapter begins by describing the laboratory accreditation system, which seeks to ensure the technical proficiency of public and private genetic testing laboratories and the scientific reliability of the test results they produce. The chapter also considers ways in which accreditation standards can be enhanced to ensure high ethical standards in genetic testing.

11.4 The chapter then focuses on issues concerning the availability and use of genetic testing services provided directly to the public and, in particular, whether the supply and advertising of parentage and other kinship testing kits (whether for home or laboratory use) should be regulated by the Therapeutic Goods Administration.

Laboratory accreditation

11.5 Laboratory accreditation is the formal recognition of an organisation's competency to perform certain specific tests, classes or types of tests or instrument calibrations. In Australia, the technical competency of medical testing (of which genetic testing is a component) and forensic testing (of which parentage and other kinship testing is a component) is ensured by the accreditation scheme operated by the National Association of Testing Authorities, Australia (NATA).³ NATA is an independent, private, not-for-profit company, which operates as an association.

11.6 The Commonwealth government recognises NATA as the sole national accreditation body for establishing competent laboratory practice.⁴ According to a memorandum of understanding entered into in 1998, the Commonwealth has agreed to:

- use NATA accredited laboratories to meet its own testing needs wherever possible;
- encourage state governments and other instrumentalities to adopt a similar approach; and
- commit Commonwealth government laboratories to obtain and maintain NATA accreditation.⁵

3 There are currently 506 accredited laboratories in the field of medical testing and 30 laboratories in the field of forensic science. Of these, 55 laboratories are accredited for genetic testing, nine for forensic DNA typing, and nine for parentage testing: National Association of Testing Authorities, *Consultation*, Sydney, 19 November 2002.

4 National Association of Testing Authorities, *Memorandum of Understanding between the Commonwealth of Australia and the National Association of Testing Authorities, Australia*, <www.nata.asn.au/downloads/moa3-98.pdf>, 20 February 2003.

5 NATA website: National Association of Testing Authorities Australia, *Government Requirement for NATA*, <www.nata.asn.au/govt_reg/govreq2.htm>, 20 February 2003.

Pathology laboratory accreditation

11.7 The medical testing accreditation scheme is run jointly with the Royal College of Pathologists of Australasia (RCPA) based on policy guidance provided by the National Pathology Accreditation Advisory Council (NPAAC). The policy guidance takes the form of standards and guidelines which pathology laboratories must adopt in order to be accredited by NATA.⁶

11.8 Individual laboratories apply to NATA for accreditation in one or more categories of medical testing, and then undergo an inspection by NATA-selected assessors. Once accredited, laboratories become members of NATA.⁷ Ongoing accreditation requires periodic assessment by NATA assessors.

11.9 The NATA/RCPA accreditation scheme is also used by the Minister of Health and Ageing as a way of ensuring that laboratories wishing to obtain Medicare payment for medical services are suitably accredited.⁸ Laboratories wishing to access Medicare payments must comply with standards and guidelines developed or endorsed by NPAAC, gain accreditation from NATA, and be designated as an Approved Pathology Laboratory by the Health Insurance Commission.⁹

11.10 The current Australian pathology laboratory accreditation arrangements were reviewed by Corrs Chambers Westgarth Lawyers, at the request of the Department of Health and Ageing (the Commonwealth pathology services review). The July 2002 report of the review found that present arrangements were fundamentally sound and should be maintained.¹⁰

[C]urrent Australian pathology laboratory accreditation arrangements efficiently and effectively regulate the vast majority of pathology services provided in Australia. There is substantial qualitative evidence that the quality of pathology services has improved since the present arrangements were introduced.¹¹

6 NPAAC was established to make recommendations to the Commonwealth, States and Territories on matters relating to the accreditation of pathology laboratories and the maintenance of uniform standards of practice in pathology laboratories throughout Australia. See National Pathology Accreditation Advisory Council, *Functions*, <www.health.gov.au/npaac/functions.htm>, 20 February 2003.

7 Corrs Chambers Westgarth, *Evaluation of the Australian Pathology Laboratory Accreditation Arrangements* (2002), Department of Health and Ageing, Canberra, 3.

8 *Health Insurance Act 1973* (Cth) s 16A(2).

9 See Health Insurance (Accredited Pathology Laboratories — Approval) Principles 1999 (Cth). The Principles are determined by the Minister for Health and Ageing under the *Health Insurance Act 1973* (Cth) s 23DNA.

10 Corrs Chambers Westgarth, *Evaluation of the Australian Pathology Laboratory Accreditation Arrangements* (2002), Department of Health and Ageing, Canberra, Rec 2.1. Some problems were identified, including difficulties associated with efforts by the HIC to enforce compliance with NPAAC standards and the fact that the Commonwealth can only indirectly regulate pathology services through its administration of the Medicare Benefits Scheme, leaving unregulated some services that operate outside this framework. A number of strategies addressing all aspects of the accreditation system were recommended to strengthen the system.

11 *Ibid.*, 5.

11.11 The minimum standards for pathology laboratory practice in Australia are set out in NPAAC's *Standards for Pathology Laboratories*.¹² There are two other relevant NPAAC standards—*Quality Systems for Medical Laboratories* and *Requirements for Supervision of Pathology Laboratories*.¹³ A range of specific standards and guidelines supports these three key standards,¹⁴ including those relevant to genetic testing.¹⁵

11.12 NPAAC standards are supplemented by NATA field application documents specific to different areas of laboratory testing.¹⁶ Accreditation criteria for cytogenetic and molecular genetic testing specify the management and technical requirements that each laboratory must maintain, including:

- quality systems that should provide laboratory management with continuing confidence that results and conclusions are accurate and reliable;
- records to show that staff members have been properly trained and proof of qualifications and membership of professional societies;
- documented test and calibration methods and method validation;
- instructions, safety precautions and requirements for specimen collection; and
- test reports and calibration certificates.¹⁷

Forensic laboratory accreditation

11.13 Accreditation criteria for forensic identification testing cover the level of training of personnel, accommodation and environmental conditions in the laboratory, test and calibration methods and method of validation used, and quality assurance protocols and procedures.¹⁸

12 National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra.

13 National Pathology Accreditation Advisory Council, *Quality Systems for Medical Laboratories* (2001), Department of Health and Aged Care, Canberra; National Pathology Accreditation Advisory Council, *Requirements for Supervision of Pathology Laboratories* (1999), Department of Health and Aged Care, Canberra.

14 See Corrs Chambers Westgarth, *Evaluation of the Australian Pathology Laboratory Accreditation Arrangements* (2002), Department of Health and Ageing, Canberra, 6. These standards have been developed with reference to current Australian and other international standards including National Association of Testing Authorities (Australia), *AS ISO/IEC 17025: 1999 Australian Standard: General Requirements for the Competence of Testing and Calibration Laboratories* (1999), NATA, Sydney.

15 For example, National Pathology Accreditation Advisory Council, *Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection Techniques* (2000), Commonwealth of Australia, Canberra; National Pathology Accreditation Advisory Council, *Guidelines for Cytogenetics Laboratories* (2001), Department of Health and Aged Care, Canberra.

16 In relation to medical testing: National Association of Testing Authorities Australia, *ISO/IEC 17025 Application Document: Supplementary Requirements for Accreditation in the Field of Medical Testing* (2000), National Association of Testing Authorities, Australia.

17 Ibid.

18 National Association of Testing Authorities Australia, *ISO/IEC 17025 Application Document: Supplementary Requirements for Accreditation in the Field of Forensic Science* (2000), National Association of Testing Authorities, Australia.

11.14 Standards applicable to parentage testing provide, among other things, that laboratories must have documented policies for the interpretation of data for each method of DNA analysis,¹⁹ and that parentage test reports must comply with the *Family Law Regulations 1984* (Cth).²⁰

Non-accredited genetic testing

11.15 Non-accredited genetic testing occurs in Australia in two situations—when a non-accredited laboratory carries out genetic testing or when an accredited laboratory carries out genetic testing that does not comply with genetic testing accreditation criteria. The latter is possible because NATA permits accredited laboratories to conduct testing that does not comply with NATA requirements, provided that the laboratories do not claim to be accredited for the purposes of that particular test.²¹ Overseas laboratories that market genetic testing services via the Internet may also fail to be accredited by NATA or an equivalent international accreditation organisation.

11.16 Submissions and consultations highlighted a range of concerns about non-accredited testing. As discussed above, accreditation standards impose a wide range of general and specific management and technical requirements. Accredited laboratories are required, among other things, to employ quality management systems, comply with laboratory design and fitting standards, properly document test requests and specimens, and to be enrolled and participate in external proficiency testing programs.²²

11.17 The fundamental concern about non-accredited genetic testing is that there may be no independent assessment about the quality of the testing process, the proficiency of the personnel carrying out the test, or the reliability of the results. Under-performing laboratories outside the accreditation system may not be identifiable.²³

11.18 Other submissions expressed concerns that non-accredited testing may not comply with important ethical standards, particularly in relation to the adequacy of consent to testing, privacy and confidentiality, and ensuring that individuals are tested only in circumstances where they will be provided with proper information and advice

19 Ibid [5.4.1]. See B Atchison and N Redman, 'Interpreting DNA Evidence in Paternity Cases' (2000) 32 *Australian Journal of Forensic Sciences* 75.

20 National Association of Testing Authorities Australia, *ISO/IEC 17025 Application Document: Supplementary Requirements for Accreditation in the Field of Forensic Science* (2000), National Association of Testing Authorities, Australia, Sec 3, Pt B Supplementary Requirements for Accreditation: Parentage Testing [5.10.2(a)].

21 National Association of Testing Authorities Australia, *Correspondence*, 12 April 2002.

22 See National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra.

23 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Australian Medical Association, *Submission G212*, 29 November 2002; D Ackermann and others, *Submission G220*, 3 December 2002; National Legal Aid, *Submission G314*, 19 February 2003; Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Submission G281*, 24 December 2002.

in interpreting the test results, and access to genetic counselling, where necessary.²⁴ Accreditation standards were also seen as important in ensuring that some sensitive or complex tests are only ordered through medical practitioners.²⁵

11.19 As discussed in Chapter 35, in the interests of accurate, reliable and ethical parentage testing, the Inquiry recommends that NATA accreditation should be mandatory for all Australian laboratories that conduct DNA parentage testing.²⁶ However, concerns have also been expressed about non-accredited medical testing, including by companies offering genetic testing services directly to the public.

Recommendation of the Commonwealth pathology services review

11.20 The Commonwealth pathology services review recognised that most laboratories seek NATA accreditation as a means of accessing Medicare payments and that pathology laboratories that are not interested in reimbursement for services remain unregulated. The review noted that complementary state-based legislation may be appropriate to regulate these laboratories and enforce compliance with national pathology standards.²⁷

11.21 At present, Victoria is the only State with its own statutory accreditation scheme.²⁸ In Victoria, pathology services accreditation legislation, administered by the Pathology Services Accreditation Board on behalf of the State Minister for Health, governs the conduct of pathology testing.²⁹

11.22 As with the Commonwealth legislation, the assessment of Victorian pathology services uses the NATA/RCPA scheme and the NPAAC standards. However, enforcement of the Victorian scheme extends not only to pathology services that are eligible for Medicare payments, but also to those services that are either ineligible or do not seek access to such payments.³⁰

24 Centre for Genetics Education, *Submission G232*, 18 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002.

25 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

26 Recommendation 35–1. The Inquiry also recommends that forensic analysis of genetic samples should be conducted only by laboratories accredited by NATA in the field of forensic science (Recommendation 41–7).

27 Corrs Chambers Westgarth, *Evaluation of the Australian Pathology Laboratory Accreditation Arrangements* (2002), Department of Health and Ageing, Canberra, i.

28 See *Pathology Services Accreditation Legislation Review, Review of the Pathology Services Accreditation Act 1984: Discussion Paper* (2002), Department of Health and Human Services (Vic), Melbourne, 15.

29 *Pathology Services Accreditation Act 1984* (Vic). The Board's functions include accrediting and registering pathology services; making recommendations to the Minister on proposals for regulations pertaining to matters such as minimum qualifications required for the person in charge of each category of accredited pathology service and any person conducting tests, minimum standards required for the conduct of tests; and making recommendations to the Minister about which tests should be prescribed as exempted tests and non-regulated tests: s 9.

30 *Pathology Services Accreditation Legislation Review, Review of the Pathology Services Accreditation Act 1984: Discussion Paper* (2002), Department of Health and Human Services (Vic), Melbourne, 17.

11.23 The Commonwealth pathology services review concluded that state legislation to complement existing Commonwealth legislation could assure the public about the quality of the relatively small number of pathology services that are currently unregulated because they do not seek Medicare payment.³¹ The review recommended:

That the [Department of Health and Ageing], in partnership with State and Territory health authorities, undertakes a detailed evaluation of the need for, and potential costs and benefits of, additional legislation in all jurisdictions to complement the Australian pathology laboratory accreditation arrangements.³²

11.24 In August 2002, the Government expressed agreement with this recommendation and announced measures to enhance standards of pathology laboratory testing and identify laboratories that pose a risk to public health and safety.³³ The Commonwealth Department of Health and Ageing has confirmed that the establishment and implementation of a nationally consistent quality assurance and accreditation scheme is under consideration by the Australian Health Ministers' Advisory Council Advisory Group on Human Gene Patents and Genetic Testing.³⁴

New legislation to require accreditation

11.25 In DP 66, the Inquiry asked whether legislation should be enacted to require laboratories that conduct genetic testing to be accredited by NATA and to comply with accreditation standards in respect of all genetic testing.³⁵ In response, there was support for the principle that laboratories conducting medical genetic testing should be accredited and that additional regulation may be required to enforce accreditation requirements.³⁶

11.26 However, the New South Wales Health Department submitted that it would prefer to see improvements to existing accreditation processes and flexible policies rather than the introduction of new legislation.³⁷

31 Corrs Chambers Westgarth, *Evaluation of the Australian Pathology Laboratory Accreditation Arrangements* (2002), Department of Health and Ageing, Canberra, 5.

32 Ibid, Rec 2.2.

33 Senator the Hon Kay Patterson, 'Enhanced Pathology Laboratory Testing Standards to Protect Public Health and Safety', 29 August 2002.

34 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

35 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Question 5-1.

36 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Australian Medical Association, *Submission G212*, 29 November 2002; D Ackermann and others, *Submission G220*, 3 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Submission G281*, 24 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002. The HGSA did not consider that specific legislation was necessary to enforce accreditation requirements: Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; National Legal Aid, *Submission G314*, 19 February 2003.

37 NSW Health Department, *Submission G303*, 13 January 2003.

[A]ny policy will need to be flexible to enable genetic testing for often rare disorders for diagnostic purposes, where the test is still under development or part of a research study, to be undertaken by unaccredited research laboratories. Sometimes these laboratories are the only facility to offer a valuable test, and may be the patient's only hope.³⁸

11.27 It was also suggested that the enactment of legislation to prohibit genetic testing by non-accredited laboratories may have unforeseen consequences in relation to some forms of genetic testing.³⁹ The Department of Health Western Australia submitted that:

Careful consideration should be given to any statutory requirement that a provider of a genetic testing service must be accredited. Such criteria could be seen as anti-competitive and should only be introduced where it is justified on the basis of protecting consumers against identified harm.⁴⁰

11.28 The Cancer Council Victoria Cancer Genetics Advisory Council noted that new accreditation requirements could set a precedent for all forms of diagnostic testing with widespread implications across the entire pathology industry.

The cost and effort in achieving [NATA accreditation standards] would be prohibitive for many smaller laboratories which would cease testing. Many of these would be likely to be non-accredited but nonetheless highly capable research laboratories that perform diagnostic tests, often at no cost, for rare or esoteric genetic diseases (eg MEN1) that are not available elsewhere. Somehow these tests will need to be incorporated into an accredited laboratory setting without disrupting a valued clinical service and research activity.⁴¹

11.29 Most research laboratories are not NATA accredited, notwithstanding that they may perform diagnostic tests from time to time. This does not mean that research laboratories fail to meet stringent standards of technical competence and quality assurance, but rather that there has been no need for research laboratories to obtain accreditation, since they do not receive payment under Medicare. The Children's Cancer Institute Australia submitted:

We maintain that NATA accreditation should **not** be required during the development phase of genetic tests, including clinical trials for new tests being undertaken with research funding, and at no cost to either individuals or the health care system. Genetic tests are often developed in research laboratories, under appropriate approval by an institutional Human Research Ethics Committee, where such accreditation would not already be in place. So long as the gene test is in the research phase, it

38 Ibid.

39 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002.

40 Department of Health Western Australia, *Submission G271*, 23 December 2002.

41 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002. Similar concerns about the accreditation of research testing were also expressed in consultations: Children's Hospital at Westmead, *Consultation*, Sydney, 19 November 2002; Royal College of Pathologists of Australasia, *Consultation*, Sydney, 18 November 2002.

should not require NATA accreditation: the accreditation could be deemed to become essential at the point when the testing procedure attracts a fee or Medicare rebate.⁴²

11.30 The Human Genetics Society of Australasia (HGSA) stated that:

the consequence of requiring NATA accreditation for laboratories that provide clinical genetic testing is that many tests, currently provided by non-accredited research laboratories, would no longer be available, or there would need to be technology transfer to an accredited laboratory which can then perform the test. This issue will require addressing by the proposed HGCA in consultation with the NHMRC.⁴³

11.31 There is a recognised continuum between research and clinical practice.⁴⁴ It is not always clear when a genetic test performed by a pathologist is diagnostic, rather than for the purposes of research into causes of disease.⁴⁵ Genetic testing often starts in a research setting before entering the mainstream. It is important, therefore, to ensure that basic research is not impeded by imposing a requirement of NATA accreditation to the research context.⁴⁶ Another issue is whether or not an Australian laboratory that receives samples within Australia for dispatch and testing overseas needs to be accredited, even if no testing is performed on site.⁴⁷

11.32 Submissions and consultations confirmed that the NATA laboratory accreditation system is widely recognised as an effective means of ensuring minimum standards of technical proficiency in laboratory testing. The role of the pathology accreditation system has been highlighted by recent events involving pap smear testing,⁴⁸ and is seen as a critical factor in the effective operation of national screening programs for breast, cervical and bowel cancer.⁴⁹

11.33 Subject to the qualifications noted below, the Inquiry considers that accreditation requirements should be extended to all laboratories that conduct genetic tests for medical, diagnostic or treatment purposes. The Inquiry recognises, however, that further consideration of these issues should take place within a broader health policy context. In particular, the implications of any new accreditation legislation for

42 Children's Cancer Institute Australia, *Submission G221*, 29 November 2002.

43 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

44 Royal College of Pathologists of Australasia, *Consultation*, Sydney, 18 November 2002. See also Australian Academy of Science, *Submission G097*, 21 January 2002; Australian Society for Medical Research, *Submission G124*, 18 March 2002.

45 Royal College of Pathologists of Australasia, *Consultation*, Sydney, 18 November 2002.

46 Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

47 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

48 Senator the Hon Kay Patterson, 'Press Release: Victorian Women Can Have Confidence in Checks for Cervical Cancer', 7 March 2002.

49 Senator the Hon Kay Patterson, 'Enhanced Pathology Laboratory Testing Standards to Protect Public Health and Safety', 29 August 2002.

research testing, and testing performed in research laboratories, would have to be carefully considered and exemptions developed in appropriate cases.⁵⁰

Recommendation 11–1. In order to complement existing pathology laboratory accreditation arrangements, the Commonwealth, States and Territories should enact legislation to require laboratories to: (a) be accredited for any genetic test that they conduct for medical, diagnostic or treatment purposes; and (b) comply with the relevant accreditation standards. The legislation should make provision for exemptions in appropriate circumstances, such as for genetic tests performed by research laboratories.

Reform of accreditation standards

11.34 A related issue concerns the extent to which accreditation standards should address ethical concerns, as well as the technical proficiency of the testing process. Should accreditation standards seek, for example, to ensure that individuals who are subjected to genetic testing are adequately informed about any risks associated with the test and the implications of the test result and have the opportunity to be referred to genetic counselling? DP 66 noted that accreditation standards do not generally address issues of consent, although in the case of parentage testing for family law proceedings they may do so indirectly.⁵¹

11.35 Submissions and consultations raised concerns about whether accreditation standards provide adequate ethical safeguards. Many observed that the current accreditation standards do not address issues such as privacy, informed consent, access to counselling or chain of custody—rather they operate on ‘the purely technical level of laboratory procedures’.⁵² There was broad support in submissions for augmenting accreditation standards to deal better with consent and other ethical concerns.⁵³

50 For example, the *Pathology Services Accreditation Act 1984* (Vic) allows the Pathology Services Accreditation Board (PSAB) to exempt certain tests performed by medical practitioners from accreditation requirements and for pathologists (or other specialist medical practitioners) to perform tests which are not exempted, provided that the tests and the laboratory in which the tests are performed have been approved by the PSAB: *Pathology Services Accreditation Act 1984* (Vic) s 30(2)(a); s 30(4). The Victorian legislation defines a ‘pathology service’ as ‘a service in which human tissue, human fluids or human body products are subjected to analysis for the purposes of prevention, diagnosis or treatment of disease in human beings and includes any premises from which a service is conducted’: *Pathology Services Accreditation Act 1984* (Vic) s 3. This definition would include a research laboratory that performs medical testing on an *ad hoc* basis.

51 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [5.26].

52 C Andersen, *Submission G002*, 14 January 2002.

53 Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Genetic Technologies Corporation Pty Ltd, *Submission G245*, 19 December 2002; NSW Health

Relevant NPAAC Standards

11.36 NPAAC's *Standards for Pathology Laboratories*,⁵⁴ which came into effect on 1 January 2003, include general standards on laboratory ethics and informed consent.⁵⁵ In addition, NPAAC accreditation standards applying specifically to genetic medical testing contain sections relating to ethical concerns. As noted above, pathology laboratories must adopt NPAAC standards in order to be accredited by NATA.

11.37 NPAAC's *Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection Techniques* (the NPAAC Nucleic Acid Detection Standards) contain provisions concerning consent and counselling in genetic testing. These standards draw a distinction between two broad classes of medical genetic testing—'Class A: Diagnostic Genetic Tests' and 'Class B: Predictive, Carrier and Prenatal Genetic Tests'.⁵⁶

11.38 The NPAAC Nucleic Acid Detection Standards provide that Class A tests require verbal consent of the individual being tested (or legal guardian) and do not require specific pre-test counselling for genetic disease. Tests in this class are appropriate for access by the health professionals providing patient care.⁵⁷

11.39 In contrast, Class B tests would typically be the province of a specialist laboratory working in close association with a number of specialist referrers. The tests in this category ... require formal consent, pre- and post-test counselling, confidentiality procedures, and close dialogue between laboratory and clinical services.⁵⁸

Department, *Submission G303*, 13 January 2003; Department of Health Western Australia, *Submission G271*, 23 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; National Legal Aid, *Submission G314*, 19 February 2003.

54 National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra.

55 Ibid, 10 (Standard 5—Pre-analytical Stage). See also National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra, 5 (Standard 1—Laboratory Ethics).

56 National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra, 5. An NPAAC Advisory Document provides guidelines as to which molecular genetic tests should be categorised as Class A or Class B tests. The major discriminator is stated to be the 'reason for the performance of the test rather than test itself' eg a Fragile XA test could be a Class A or Class B test depending on whether it is offered for diagnosis in the developmentally delayed child or undertaken on a sample from a known carrier for prenatal diagnosis. Class A tests include predictive tests for conditions where a simple treatment exists, screening tests supported as a public health measure, and some carrier tests: National Pathology Accreditation Advisory Council, *NPAAC Advisory Document: Information for Classes of Genetic Testing* (2001) Department of Health and Aged Care.

57 National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra, 5.

58 National Pathology Accreditation Advisory Council, *Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection Techniques* (2000), Commonwealth of Australia, Canberra, 5.

11.40 Under the the NPAAC Nucleic Acid Detection Standards, laboratories have ethical responsibilities where requests for specialised testing (Class B tests) are

initiated by a health professional unlikely to be familiar with the complexities of the diagnosis, or where a person directly seeking genetics advice has initiated a request for testing without consultation with an appropriate health professional.⁵⁹

11.41 In these circumstances it is the responsibility of the laboratory director to ensure laboratory practice is consistent with accepted ethical guidelines.⁶⁰ Furthermore, where there are reasonable grounds for believing that informed consent may not have been given, appropriate care (which might include counselling) has not been available, or patient confidentiality may have been breached, the laboratory is obliged to contact the referring practitioner 'to ensure that informed consent is obtained prior to laboratory testing and provide the practitioner with information regarding how to access genetic services in the region'.⁶¹ The NPAAC Nucleic Acid Detection Standards also provide that laboratories

shall not provide genetic testing for any patient-initiated service, such as mail-order testing or shop-front testing, which does not occur in the context of a clinical service that provides for appropriate genetic counselling.⁶²

11.42 DP 66 proposed that NPAAC should inquire into whether accreditation standards should ensure that laboratories conduct genetic testing only on bodily samples collected with the consent of the individual to whom the sample relates or as approved by a Human Research Ethics Committee.⁶³

11.43 The Cancer Council of Victoria Cancer Genetics Advisory Committee agreed that NPAAC should amend its standards to make it a requirement for laboratories to conduct genetic tests only when consent has been obtained or waived.⁶⁴ However, the Committee noted that, at present, only predictive genetic tests currently require written consent, as well as pre- and post-test counselling. It stated that:

if recommendations are to be made to NPAAC to amend its standards, then further thought needs to be given whether this applies to all genetic tests and if not which tests.⁶⁵

59 Ibid, 6.

60 Such as those produced by the NHMRC or AHEC or by professional bodies with standing in the field, such as the Australian Medical Association, the Australian Cancer Network or the Human Genetics Society of Australasia: Ibid, 6.

61 Ibid, 6.

62 Ibid, 6.

63 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 5-1. NPAAC submitted that this Proposal was addressed in the NPAAC Nucleic Acid Detection Standards: National Pathology Accreditation Advisory Council, *Submission G184*, 12 September 2002.

64 Other submissions agreed that consent requirements are properly part of accreditation standards relating to genetic testing: Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002.

65 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

11.44 The Centre for Genetics Education noted that in the health care setting a patient may often not provide written consent to a diagnostic test but consent will be assumed by the fact of having consulted a health professional.

[I]t will be important in the areas of carrier and predictive testing however that the consent is informed and therefore it may be appropriate for the HGCA to be involved in the development of consent forms developed for national use ...⁶⁶

11.45 Submissions expressed particular concern about the possible scope of obligations placed on laboratories to ensure that proper consent processes have been followed.⁶⁷ HGSA's view was that the onus for demonstrating that appropriate informed consent has been obtained should 'rest with the managing clinician, rather than the laboratory'.

From the laboratory's point of view, it should be sufficient to have evidence, for example as a marked box on the test request form that the clinician considers that appropriate consent has been obtained.⁶⁸

Inquiry's views

11.46 While the primary focus of laboratory accreditation should be on technical proficiency and scientific reliability, the Inquiry considers that NPAAC should continue to include ethical considerations within its genetic testing accreditation standards. In this context, the RCPA submitted that:

international laboratory standards appear to be moving in the direction of inclusion of Ethics within Laboratory Standards, and the College is supportive of such moves. The peak body that would need to embrace the inclusion of an Ethics framework within Australian laboratory practice would be NPAAC, and the College would be supportive of any moves in this direction.⁶⁹

11.47 To this end, NPAAC should promulgate similar policies in other standards that are relevant to genetic testing laboratories, such as the NPAAC *Guidelines for Cytogenetics Laboratories*.⁷⁰

11.48 In Chapter 23 the Inquiry recommends that the Human Genetics Commission of Australia (HGCA) develop genetic testing and counselling practice guidelines (see Recommendation 23–3). These guidelines could identify specific genetic tests, or categories of tests, that require special consideration in obtaining informed consent or ensuring access to genetic counselling. In turn, NPAAC could ensure that their

66 Centre for Genetics Education, *Submission G232*, 18 December 2002.

67 Genetic Technologies Corporation Pty Ltd, *Submission G245*, 19 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

68 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

69 Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002. The RCPA noted that Draft International Standard ISO/DIS 15189, which may succeed ISO 17025 as the preferred standard supported by NPAAC, contains an appendix on ethics in laboratory medicine.

70 National Pathology Accreditation Advisory Council, *Guidelines for Cytogenetics Laboratories* (2001), Department of Health and Aged Care, Canberra. NPAAC has agreed that the issue could be addressed when the *Guidelines for Cytogenetics Laboratories* are reviewed in 2004: National Pathology Accreditation Advisory Council, *Submission G184*, 12 September 2002.

accreditation standards reflect the relevant HGCA guidelines. For example, should the HGCA recommend that a certain class of genetic test be conducted only after genetic counselling has been provided, NPAAC could ensure that accreditation standards reflect these obligations. The Inquiry also believes that it would be beneficial for such accreditation standards to be developed in consultation with the National Health and Medical Research Committee (NHMRC).

11.49 In addition, the Inquiry considers that NPAAC, in consultation with NATA and the RCPA, should consider how compliance with ethical accreditation standards should be assessed as part of the NATA/RCPA accreditation process. The means of verifying compliance with these considerations should be emphasised as part of training programs for NATA officers and peer assessors conducting assessments of laboratories.

Recommendation 11–2. While the primary focus of laboratory accreditation should remain on matters of technical proficiency and scientific reliability, the National Pathology Accreditation Advisory Council (NPAAC) should continue to develop ethical standards for medical genetic testing, in consultation with the Human Genetics Commission of Australia and the National Health and Medical Research Council.

Recommendation 11–3. NPAAC, in consultation with the National Association of Testing Authorities, Australia (NATA) and the Royal College of Pathologists of Australasia (RCPA), should examine how compliance with its accreditation standards in relation to consent, counselling and other ethical considerations in medical genetic testing should be assessed as part of the NATA/RCPA accreditation process.

Recommendation 11–4. NATA, in consultation with the RCPA, should develop training programs to equip its officers and peer assessors to verify compliance with NPAAC accreditation standards relating to consent, counselling and other ethical considerations.

Genetic testing services provided directly to the public

Direct to the public genetic testing

11.50 In this Report, direct to the public genetic testing refers to two different forms of genetic testing. One form is akin to home pregnancy testing, in which the test is performed and interpreted by the person at home. At present, this form of genetic testing is not available in Australia.

11.51 The second form is a test in which the person collects a bodily sample at home and sends it to a laboratory for analysis. Kits for testing may be made available through pharmacists or other retailers, by mail order or over the Internet. In general,

they may be expected to provide: instructions on how to collect and store bodily samples, implements to collect samples (for example, buccal swabs) and vials or other containers in which to store the samples for testing. The samples are forwarded through the mail to the company offering the services, either in Australia or overseas. The testing may or may not be done by an accredited laboratory.

11.52 DNA parentage testing is currently available in this second form by mail order and over the Internet. While the Inquiry is not aware of any Australian businesses that offer such a service outside the context of parentage testing, this is not the case overseas. For example, a United Kingdom biotechnology company, Sciona, has marketed a genetic testing service on the Internet and through The Body Shop retail chain. Sciona claimed that the testing service ‘offers a personalised dietary report generated by combining information from a lifestyle questionnaire and an understanding of your genes’.⁷¹ For £120, customers were sent a self-sampling kit and a lifestyle questionnaire, which were processed to produce a report that claimed to provide individualised recommendations on what to eat to ensure that body and diet work ‘in harmony’.⁷²

11.53 Sciona and The Body Shop were both criticised by GeneWatch UK,⁷³ which was concerned that the tests were unregulated and misleading. GeneWatch claimed that Sciona’s genetic tests misled subscribers into thinking that ‘good genes can cope with a bad diet or with smoking or excessive drinking’. It expressed its concern that subscribers were not informed about privacy or discrimination implications.⁷⁴ The Body Shop withdrew the kits from sale through its retail outlets.

11.54 A range of genetic testing kits is available over the Internet from providers in the United States. These include kits claimed to test for genes linked to the development of osteoporosis, heart disease, immune deficiency and obesity.⁷⁵ DNA identification kits have been marketed as a means for parents to collect and store DNA samples at home, which can later be used to trace missing persons.⁷⁶

11.55 It may be only a matter of time before there is broad public interest in genetic tests, including DNA identification and parentage testing kits, marketed directly to the public in Australia. The Department of Health and Ageing observed in relation to medical testing that:

71 See Sciona’s website: Sciona Ltd, *About Sciona*, <www.sciona.com/about/index.htm>, 22 July 2002.

72 See BBC News, *Row over Gene Testing Kit*, <www.news.bbc.co.uk/1/hi/sci/tech/1868767.stm>, 26 July 2002.

73 GeneWatch UK is ‘an independent organisation concerned with the ethics and risks of genetic engineering’: GeneWatch, *Home Page*, <www.genewatch.org>, 20 February 2003.

74 See GeneWatch, *Body Shop’s Genetic Tests Misleading and Unethical*, <www.genewatch.org/HumanGen/Press_Releases/pr22.htm>, 19 February 2003.

75 D Smith, ‘Home Genome Project’, *The Sydney Morning Herald*, 27 February 2003.

76 R Willing, ‘Child DNA Kits Become Businesses’ Favorite Freebie’, *USA Today*, 12 May 2002.

[t]he current genetic testing environment does not suggest that a wave of individuals will suddenly start to self-refer for testing, although the possibility cannot be discounted in light of previous experience with coronary artery calcium scoring which has been marketed directly to the public in the past.⁷⁷

11.56 In IP 26, the Inquiry asked whether genetic testing should be available only on the request of a medical practitioner.⁷⁸ Many submissions that addressed this question identified ethical concerns relating to direct to the public genetic testing and suggested the need for further regulation. The Australian Medical Association (AMA) submitted that:

genetic testing should be regulated to ensure that only registered medical practitioners are allowed to request that testing be performed. Genetic testing should not be performed in the absence of counselling. For this reason we are strongly opposed to the availability of 'home-based' testing kits due to the significant risk of physical or emotional harm that may result to individuals who submit to genetic testing without this counselling. There is also significant risk of genetic testing being performed on individuals without their consent if home testing kits are allowed. The AMA considers it necessary to enact legislation to ban this type of product.⁷⁹

11.57 The Department of Health and Ageing recognised the ethical issues raised by direct to the public genetic testing but submitted that there may be ways to address these other than by requiring that genetic testing be obtained only through medical practitioners.

In general, care must be taken not to implicitly assent to a proposition that because genetic testing is a medical service, medical practitioners should *ipso facto* mediate it. Where individuals do self-order tests, guidelines could assist testing services to meet their ethical obligations in relation to informed consent, e.g., directing consumers to information on pre- and post-test counselling and support services.⁸⁰

11.58 Similarly, the Office of the Federal Privacy Commissioner (OFPC) submitted that access to mail-order genetic testing cannot be effectively and comprehensively regulated so that testing is conducted only on the request of a medical practitioner and by an accredited laboratory.⁸¹ The OFPC noted that:

[i]f the sale of mail-order genetic tests and 'DIY' test kits by Australian companies continues, there should be widespread and comprehensive consumer education policies devised. There are valuable lessons to be learned from the lawful sale of potentially harmful commodities, such as tobacco and prescription drugs. Consideration may even be given to legislating, as a public health measure, for the provision at the point of sale of appropriate information about the tests and the implications of the results obtained. Such consumer education would have the added benefit of ensuring the informed consent of the individual, an important element of information privacy principles.⁸²

77 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

78 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, Question 8–2.

79 Australian Medical Association, *Submission G091*, 29 January 2002.

80 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

81 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

82 *Ibid.*

Overseas developments

11.59 The regulation of direct to the public genetic testing has been the subject of deliberations in other jurisdictions. In 1997, an Ontario report concluded that Canadian federal standards for approval of genetic testing should be carefully examined and monitored and that the federal government should ensure that ‘direct to consumer marketing of genetic testing should at minimum be clearly circumscribed if not entirely prohibited for certain forms of testing’.⁸³ A similar conclusion was reached by the US National Institutes of Health Task Force on Genetic Testing, which recommended that advertising or marketing of predictive genetic tests to the public should be discouraged.⁸⁴

11.60 The United Kingdom Advisory Committee on Genetic Testing (ACGT) prepared a Code of Practice in respect of genetic testing services supplied directly to the public.⁸⁵ The ACGT concluded that direct provision of testing should be limited in relation to testing for inherited dominant and X-linked disorders and adult-onset genetic disorders.⁸⁶ The Code of Practice is voluntary and there are no legal sanctions for failing to comply with it.

11.61 In July 2002, the United Kingdom Human Genetics Commission (HGC) released a consultation paper on the supply of genetic tests direct to the public.⁸⁷ The consultation paper noted that the technology of genetic testing has become simpler and more readily available, making rapid and relatively inexpensive genetic testing a reality, and that this development has been accompanied by a ‘growing understanding by the public of medical information’ and ‘a corresponding desire to make health decisions on the basis of information directly acquired and understood by the patient’.⁸⁸

11.62 The consultation paper noted that genetic testing services supplied direct to the public in the United Kingdom are regulated by a mixture of specific and non-specific regulation and voluntary compliance mechanisms, including the ACGT Code of Practice, laboratory standards, professional self-regulation, and consumer protection legislation. The HGC identified four options for future regulation of direct supply of genetic tests: no specific regulation; voluntary regulation; voluntary regulation with restrictions on the types of test that can be offered; and strict regulation.⁸⁹ The content

83 Province of Ontario, *Report to Premiers — Genetics, Testing and Gene Patenting: Charting New Territory in Healthcare*, <www.gov.on.ca/health/english/pub/ministry/geneticsrep02/report_e.pdf>, 20 February 2003, 85–86.

84 N Holtzman and M Watson, *Promoting Safe and Effective Genetic Testing in the United States*, National Institutes of Health, <www.nhgri.nih.gov/ELSI/TFGT_final/>, 19 February 2003.

85 Advisory Committee on Genetic Testing, *Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public* (1997), Department of Health, London.

86 *Ibid.*, 4: The ACGT recognised a limited role for the provision of genetic testing services direct to the public ‘centred around those tests which determine carrier status for inherited recessive disorders, where such status carries no significant direct health implications for the carrier individual’.

87 Human Genetics Commission, *Supply of Genetic Tests Direct to the Public: A Consultation Document*, <www.hgc.gov.uk/testingconsultation/index.htm>, 20 February 2003.

88 *Ibid.*, 5.

89 *Ibid.*, 21–24.

of the final report was discussed at an HGC plenary meeting in February 2003. An HGC statement indicated that it is likely the HGC will recommend a ‘mixture of new controls and safeguards’ according to the seriousness of the test:

For example, serious predictive genetic tests should generally be available only after medical consultation, while other tests may be suitable for wider access, e.g. diet-related tests. The commission is likely to recommend that the new Medicines and Healthcare Products Regulatory Agency (MHRA) is responsible for making these kinds of decision.⁹⁰

11.63 The European Group on Ethics in Science and New Technologies has also issued a statement warning that the mass marketing of genetic tests raises ethical, social and legal problems that require urgent attention. The Group expressed its concern about misleading and incomplete information being provided by marketers of tests, and about testing without consent (particularly in the case of parentage testing). The Group observed that advertising of genetic tests ‘tends to convert them into commodities’ and to give rise to a demand for genetic testing which may result in social disruption and personal conflict.⁹¹

Regulating direct to the public genetic testing

11.64 DP 66 identified two options for reform to regulate direct to the public genetic testing.⁹² These involved implementing one or both of the following:

- amending the *Therapeutic Goods Act 1989* (Cth) (*Therapeutic Goods Act*) and regulations to ensure that the supply and advertising of genetic tests directly to consumers is prohibited in Australia, except where specifically approved by the Therapeutic Goods Administration (TGA);
- making an appropriate body (such as the HGCA)⁹³ responsible for developing Codes of Practice and other advice on direct to the public genetic testing, including advice to the TGA on the regulation of genetic testing under its Act.

Therapeutic Goods Act and Regulations

11.65 The TGA is a Division of the Department of Health and Ageing with overall responsibility for administering the provisions of the *Therapeutic Goods Act*. The Act provides the legislative basis for a uniform national system of controls for therapeutic goods, including medicines, medical devices and other therapeutic goods such as ‘diagnostic goods for *in vitro* use’ (also called *in vitro* diagnostic devices, or IVDs).

90 Human Genetics Commission, ‘Statement About Progress of the Human Genetics Commission’s Consultation into the Supply of Genetic Tests Direct to the Public’, *Press Release* (London), 5 February 2003.

91 European Group on Ethics in Science and New Technologies, ‘Statement by the European Group on Ethics in Science and New Technologies on advertising genetic tests via the Internet’, *Ibid*(Brussels), 24 February 2003.

92 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [5.43].

93 See Ch 5.

11.66 These controls are primarily effected through regulation of the quality, safety, efficacy and timely availability of therapeutic goods; setting standards that the goods are required to comply with, and minimising any risk of misuse, inappropriate use, or unsafe use of the goods; and by regulating how the goods may be advertised. Unless specifically excluded or exempt, therapeutic goods may not be manufactured in Australia, supplied on the Australian market, or exported, unless included on the Australian Register of Therapeutic Goods (ARTG).

11.67 Therapeutic goods are defined in the Act as ‘goods that are represented ... for therapeutic use’.⁹⁴ Therapeutic use means use in or in connection with:

- (a) preventing, diagnosing, curing or alleviating a disease, ailment, defect or injury in persons or animals; or
- (b) influencing, inhibiting or modifying a physiological process in persons or animals; or
- (c) testing the susceptibility of persons or animals to a disease or ailment; or
- (d) influencing, controlling or preventing conception in persons; or
- (e) testing for pregnancy in persons; or
- (f) the replacement or modification of parts of the anatomy in persons or animals.⁹⁵

11.68 At present, therapeutic goods are classified as ‘registrable’ or ‘listable’ on a product-by-product basis. Registrable therapeutic goods are generally higher risk products and must undergo evaluation by the TGA prior to being included on the ARTG. When determining whether registrable goods should be included in the ARTG, the advice and recommendations of ministerially-appointed evaluation committees established under the regulations may be taken into account. The TGA may impose conditions on the registration or listing of goods, including conditions relating to their use and supply.⁹⁶

11.69 The *Therapeutic Goods Act* and the *Therapeutic Goods Regulations 1990* (Cth) (*Therapeutic Goods Regulations*) contain provisions that regulate the advertising of therapeutic goods. Advertisements for all therapeutic goods must comply with Part 2 of the *Therapeutic Goods Regulations* and with the Therapeutic Goods Advertising Code. A range of sanctions may be applied to non-conforming advertisements.⁹⁷

Current regulation of therapeutic goods used in genetic testing

11.70 Therapeutic goods used in genetic testing, whether sold as individual components or as a kit, are considered to be IVDs, which are defined as follows in the *Therapeutic Goods Regulations*:

diagnostic goods for in vitro use means any therapeutic device which is a reagent, reagent product, calibrator, control material, kit, instrument, apparatus, equipment or system, whether used alone or in combination (with other diagnostic goods for in vitro

94 *Therapeutic Goods Act 1989* (Cth) s 3.

95 *Ibid* s 3.

96 *Ibid* s 28.

97 *Therapeutic Goods Regulations 1990* (Cth) r 6, 7–8.

use), intended by the manufacturer to be used in vitro for the examination of specimens (including blood and tissue donations) derived from the human body, solely or principally for the purpose of giving information about a physiological or pathological state or a congenital abnormality or to determine safety and compatibility with a potential recipient.⁹⁸

11.71 Only a very limited number of IVDs are required to be included on the ARTG, including tests for HIV and Hepatitis C.⁹⁹ Most other IVDs—except goods for home use, goods that incorporate material of human origin, or goods supplied under the Pharmaceutical Benefits Scheme¹⁰⁰—are exempt from the registration and listing requirements of Part 3–2 of the *Therapeutic Goods Act*.

11.72 The *Therapeutic Goods Act* and *Regulations* cover ‘goods for home use’,¹⁰¹ which include any therapeutic device intended for use outside a laboratory setting or to be carried out by a lay person—for example a pregnancy test kit purchased in a pharmacy and designed to be carried out at home.

11.73 In the context of genetic testing, products used to provide genetic information may or may not be considered IVDs, depending on their intended use as stated by the manufacturer.¹⁰² For example, a buccal swab test kit containing implements for collecting and storing a buccal swab would be considered an IVD as it is supplied with the intention that it be used to examine a human specimen, while a pair of tweezers would not be considered to be an IVD unless they were marketed for the specific purpose of genetic sample (hair follicle) collection.¹⁰³

11.74 The situation is different in the case of goods used in DNA identification testing, including parentage testing. These goods do not fall within the definition of a therapeutic good because their intended purpose is not covered within the scope of ‘therapeutic use’ and therefore they are not currently regulated by the TGA at all.¹⁰⁴

New regulatory framework for IVDs

11.75 The TGA has initiated development of a new regulatory framework for IVDs and an expert advisory group has been established to assist the TGA in this regard. The development of a new regulatory framework is relevant to the future regulation of direct to the public genetic testing, as many genetic tests are considered to be IVDs. The expert advisory group has agreed that the new framework should include the following basic features:

98 Ibid r 2.

99 Ibid Sch 3, Item 7.

100 Ibid Sch 5, Item 7.

101 Ibid Sch 5, Item 7(b)(i). Tests for the screening and diagnosis of HIV and Hepatitis C are also not exempt from Pt 3.

102 Therapeutic Goods Administration, *Correspondence*, 19 July 2002.

103 Ibid.

104 Ibid.

- a risk-based classification system;¹⁰⁵
- essential principles for quality, safety and performance, with which all IVDs will have to comply prior to being supplied;
- options as to how compliance with the essential principles can be demonstrated, for example the implementation of a manufacturer quality system or design evaluation;
- the use of recognised standards as a way of demonstrating compliance with one or more essential principles;
- a comprehensive post market surveillance and adverse incident reporting program; and
- the continued use of the ARTG as the central point of control for the legal supply of medical devices in Australia.¹⁰⁶

11.76 The new regulatory framework for IVDs will not distinguish between products intended for use in a laboratory and those intended for use in the home. It is expected that this may affect the availability of direct to the public genetic testing products.¹⁰⁷

11.77 Other issues being considered as part of the development of new IVDs regulation include how to take account of distinctions between tests performed and interpreted by the person at home and tests where samples are collected at home but analysed in a laboratory. Also being considered is the possible expansion of the definition of ‘therapeutic use’ in s 3 of the *Therapeutic Goods Act* to include parentage and other kinship testing.¹⁰⁸

Code of practice for direct to the public genetic testing

11.78 A voluntary code of practice could be developed to complement increased regulation by the TGA of direct to the public genetic testing. Such a code could include minimum technical standards for companies supplying products and minimum ethical standards for laboratories supplying the testing service.

11.79 For example, in the United Kingdom, the ACGT Code of Practice on genetic testing services supplied direct to the public prescribed minimum requirements in relation to testing laboratories, equipment and reagents, confidentiality and storage of samples and records, tests that may be supplied, to whom they may be supplied,

105 In terms of risk classification, genetic tests are likely to fall into the mid- to high risk categories due to the potential impact on individuals being tested: Therapeutic Goods Administration, *Consultation*, Canberra, 21 November 2002.

106 Therapeutic Goods Administration, *Correspondence*, 30 January 2003. See also Therapeutic Goods Administration, *Australian Medical Devices Guidelines: An Overview of the New Medical Devices Regulatory System* (2002) Department of Health and Ageing, 5.

107 Therapeutic Goods Administration, *Correspondence*, 30 January 2003.

108 Therapeutic Goods Administration, *Consultation*, Canberra, 21 November 2002.

customer information, genetic consultation, and the involvement of general medical practitioners.¹⁰⁹ More recently, the United Kingdom's Department of Health has developed a voluntary code applying to organisations advertising and providing genetic paternity testing services directly to the public (see Chapter 35).¹¹⁰

11.80 The HGCA would be an appropriate body to develop a code of practice in Australia, in consultation with interested business and health consumer groups. In addition, if parentage and other DNA identification tests were to be regulated by the TGA, the HGCA could provide the TGA, or its statutory advisory committees,¹¹¹ with advice on whether certain categories of testing should be approved and on what terms.¹¹²

Inquiry's views

11.81 Submissions generally supported more stringent regulation by the TGA of direct to the public genetic testing,¹¹³ including for parentage and other identification testing.¹¹⁴ The availability of direct to the public genetic testing raises a range of concerns. These include the following:

- error and fraud are more likely where the collection of a genetic sample is conducted without the supervision of a medical practitioner or the testing laboratory;

109 Advisory Committee on Genetic Testing, *Code of Practice and Guidance on Human Genetic Testing Services Supplied Direct to the Public* (1997), Department of Health, London.

110 Group on Genetic Paternity Testing Services, *Code of Practice and Guidance on Genetic Paternity Testing Services*, (2001) Department of Health, <www.doh.gov.uk/genetics/paternity.htm>, 19 February 2003.

111 The Medical Devices Evaluation Committee, the Therapeutic Goods Committee and the National Coordinating Committee on Therapeutic Goods.

112 Therapeutic Goods Administration, *Consultation*, Canberra, 28 May 2002.

113 Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; D Ackermann and others, *Submission G220*, 3 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; A Gesche, *Submission G264*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Law Institute of Victoria, *Submission G275*, 19 December 2002; National Legal Aid, *Submission G314*, 19 February 2003. The Department of Health Western Australia highlighted the need to identify clearly the harm that can flow from unregulated home testing in designing appropriate new regulation: Department of Health Western Australia, *Submission G271*, 23 December 2002. The Department also referred to the use of Ministerial product safety or information standards under s 65E of the *Trade Practices Act 1974* (Cth) as an alternative or additional regulatory mechanism.

114 N Stott Despoja, *Submission G198*, 27 November 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

- sample contamination may occur because the sample may not have been stored correctly or mailed to the laboratory under optimal conditions, or because the chain of custody of the sample cannot be verified;
- informed consent may not have been obtained from the individual sampled (or from their parent or guardian); and
- individuals are less likely to be provided with appropriate information about the implications of the genetic test or to be assisted in follow-up decision making, and are unlikely to be referred for genetic counselling, when compared with a test that is arranged through a medical practitioner.

11.82 The Inquiry considers that there are strong arguments for regulating the supply, directly to the public, of products used in some forms of genetic testing. The conduct of genetic tests within the home, and even the collection of a sample at home for analysis in a laboratory, raises issues of quality assurance, whether informed consent has been granted, and the real possibility of harm where individuals are not provided with appropriate information about the implications of genetic test results.

11.83 Additional controls for direct to the public genetic testing could include specific labelling or other requirements for genetic IVDs, or the development of criteria that would be used to determine IVDs for which direct consumer access could be approved. In particular, there may need to be unique labelling requirements for some genetic testing products. Labels or product information sheets may need to include information about the desirability of pre- or post-test counselling, the need to seek medical assistance in confirming, interpreting or understanding test results, and so on. The approval process for some genetic IVDs may need to reflect the implications of self-diagnosis for the health system—where individuals are likely to subsequently arrange a medical consultation to verify or explain test results.¹¹⁵ These are all matters suitable for regulation by the TGA.

11.84 The Inquiry has also concluded that IVDs used in DNA identification testing, including parentage testing, should be regulated by the TGA, in a similar way to IVDs. This would allow the TGA to refuse approval where, for example, the goods are not safe for the purposes for which they are to be used, the presentation of the goods is unacceptable, the goods do not conform to applicable standards, or the goods do not comply with prescribed quality or safety criteria.¹¹⁶

Recommendation 11–5. The Commonwealth should amend the *Therapeutic Goods Act 1989* (Cth) (*Therapeutic Goods Act*) and regulations made under that Act to enable the Therapeutic Goods Administration (TGA) to regulate more effectively in vitro diagnostic devices used in genetic testing provided directly to the public.

115 Therapeutic Goods Administration, *Consultation*, Canberra, 21 November 2002.

116 See *Therapeutic Goods Act 1989* (Cth) s 26.

Recommendation 11–6. The Commonwealth should amend the *Therapeutic Goods Act* and regulations made under that Act to enable the TGA to regulate DNA identification test kits used in genetic testing provided directly to the public, including for parentage and other kinship testing.

Recommendation 11–7. The HGCA should develop codes of practice and advice relating to technical and ethical standards for genetic testing services provided directly to the public, including advice to the TGA or its statutory advisory committees.

Regulating access to offshore testing

11.85 DP 66 noted that individuals in Australia can access overseas genetic testing services with relative ease using the Internet and the postal service. The problems involved with regulating such access were referred to in a number of submissions.¹¹⁷

11.86 While it may be possible to regulate the supply and advertising of genetic testing products directly to the Australian public through the *Therapeutic Goods Act* and regulations, effective regulation of genetic testing services provided overseas by foreign companies and advertised through the Internet is more difficult. Moreover, ensuring accreditation of all genetic testing performed in Australia will not necessarily overcome concerns about access by individuals to non-accredited genetic testing conducted overseas.

11.87 DP 66 noted that federal legislation has been enacted to restrict access to certain forms of Internet content such as offensive material and interactive gambling.¹¹⁸ These laws and associated industry codes could serve as models for regulating the advertising on the Internet of genetic testing services.¹¹⁹ For example, the online content scheme provides that any person may complain to the Australian Broadcasting Authority (ABA) if they believe Australians can access prohibited or potentially

117 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002; Medical Practitioners Board of Victoria, *Submission G155*, 10 April 2002; *Confidential Submission G022CON*, 3 December 2001; Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Genetic Technologies Corporation Pty Ltd, *Submission G245*, 19 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Queensland Government, *Submission G274*, 18 December 2002.

118 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [5.66]–[5.69].

119 See *Broadcasting Services Act 1992* (Cth) s 216B; Sch 5; Internet Industry Association, *Internet Industry Codes of Practice for Industry Self Regulation in Areas of Internet Content Pursuant to the Requirements of the Broadcasting Services Act 1992 as amended (9 May 2002)*, Australian Broadcasting Authority, <www.aba.gov.au/internet/industry/codes/content/printer.htm>, 20 February 2003; *Interactive Gambling Act 2001* (Cth); Internet Industry Association, *Internet Industry Interactive Gambling Industry Code: A Code for Industry Co-Regulation in the Area of Internet Gambling Pursuant to the Requirements of the Interactive Gambling Act 2001 (9 May 2002)*, Australian Broadcasting Authority, <www.aba.gov.au/internet/gambling/pdf/itf/gamblingcode.pdf>, 20 February 2003.

prohibited online content using the Internet.¹²⁰ If the content is hosted overseas the ABA notifies the content to the suppliers of filter software in accordance with procedures outlined in the Internet Industry Association Codes of Practice.¹²¹

11.88 The *Interactive Gambling Act 2001* (Cth) reflects a similar attempt to regulate Internet content hosted overseas. Complaints about interactive gambling content hosted outside Australia are investigated by the ABA and, if the content is found to be prohibited, the ABA will notify the content to the makers of Internet content filter software.¹²²

11.89 There are major technical difficulties in preventing access in Australia to Internet material hosted overseas because no blocking technique is completely effective.¹²³ Nevertheless, in relation to online offensive material, the government has concluded that, where it is technically and commercially feasible to prevent access in Australia, this should be done. The fact that such regulation may not be completely effective was considered ‘no reason not to try to align online content regulation with regulation applying to conventional media’.¹²⁴

11.90 Although the regulation of offensive material and interactive gambling on the Internet provide some comparisons, the Inquiry has concluded that it would be premature to implement a similar regime intended to restrict Internet advertising of products or services used in direct to the public genetic testing.

11.91 The advertising of genetic IVDs is already subject to the restrictions contained in the *Therapeutic Goods Act*, the *Therapeutic Goods Regulations* and the *Therapeutic Goods Advertising Code*. These restrictions also apply to Internet advertising. For example, it is an offence to advertise a genetic IVD on the Internet¹²⁵ unless the device is listed.¹²⁶ However, while advertising of therapeutic goods on the Internet is regulated when the Internet service provider is based in Australia, the TGA

120 Under the legislation, prohibited material is defined on the basis of the current national Classification Code, as RC and X-rated material, and R-rated material hosted in Australia that is not protected by adult verification procedures.

121 Department of Communications Information Technology and the Arts, *Broadcasting and Online Regulation — Online Content*, DCITA, <www.dcita.gov.au/Article/0,,0_1-2_10-3_481-4_106158,00.html>, 20 February 2003. A number of filter products are listed in the IIA Codes (scheduled filters). One of the criteria for inclusion of filters on the schedule is ability to be updated to reflect notifications from the ABA. Internet service providers are required to provide, on a cost-recovery basis, one or more of the scheduled filters for the use of their subscribers.

122 Ibid.

123 See eg P Greenfield, P Rickwood and H Tran, *Effectiveness of Internet Filtering Software Products* (2001) CSIRO.

124 Department of Communications Information Technology and the Arts, *Frequently Asked Questions*, DCITA, <www.dcita.gov.au/Article/0,,0_1-2_10-3_481-4_13871,00.html#Filtering>, 11 October 2002.

125 Under the Act, an ‘advertisement’ in relation to therapeutic goods, includes any statement, pictorial representation or design, however made, that is intended, whether directly or indirectly, to promote the use or supply of the goods: *Therapeutic Goods Act 1989* (Cth) s 3(1).

126 *Therapeutic Goods Regulations 1990* (Cth) r 6(1)(f). Unless the advertisement is directed exclusively to health professionals: *Therapeutic Goods Regulations 1990* (Cth) r 4.

is unable to regulate advertisements of any nature when the Internet service provider is based overseas.¹²⁷

11.92 There has been some recognition that advertising therapeutic goods (and medicines in particular) on the Internet may need specific regulation. For example, in 2001, the final report of the National Competition Review of Drugs, Poisons and Controlled Substances Legislation stated:

A number of stakeholders raised the issue of advertising [medicines] via the Internet. The Review noted that the current advertising restrictions apply to all forms of advertising, including the Internet. However, the capacity of the Commonwealth, State and Territory governments to regulate Internet advertising originating from overseas sources is limited. This is an international problem and one which the Commonwealth government and the governments of other countries are attempting to resolve. The Review was also concerned about the potential dangers facing consumers who purchase medicines from Internet suppliers.¹²⁸

11.93 The public health and safety implications of Internet advertising of medicines can be expected to be even more significant than those relating to the Internet advertising of direct to the public genetic testing. To date, no regime has been implemented specifically to prevent access to Internet material that is hosted overseas and advertises therapeutic goods.

11.94 The Queensland Government noted that federal, state and territory trade practices legislation may already regulate Internet advertising sufficiently to address concerns relating to direct to the public genetic testing and submitted that:

Other methods of safeguarding the interests of consumers may be more effective than imposing a legislative regime that requires surveillance and enforcement. Other methods may include an education campaign through doctors' surgeries, legal offices, schools and hospitals to assist consumers to make informed decisions and permitting accredited laboratories to advertise their NATA accreditation.¹²⁹

11.95 With these considerations in mind, the Inquiry has concluded that any consideration of options for regulating advertising on the Internet of genetic testing provided directly to the public should take place as part of addressing concerns about the Internet advertising of therapeutic goods generally, rather than in the context of the present Inquiry.

127 The TGA may liaise with consumer affairs bodies in the relevant country regarding inappropriate advertising material which is either mailed to Australian addresses or is placed onto the Internet: Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

128 R Galbally, *Review of Drugs, Poisons and Controlled Substances Legislation Final Report*, Commonwealth of Australia, <www.health.gov.au/tga/docs/pdf/rdpfinb.pdf>, 20 February 2003.

129 Queensland Government, *Submission G274*, 18 December 2002.

12. A New Criminal Offence

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Introduction

12.1 This chapter considers whether, in view of the potential for individuals to access genetic testing services, there should be additional legal protection against the taking and testing of genetic samples without the knowledge and consent of the individual from whom the samples are derived.

12.2 The Inquiry has concluded that additional legal protection is required and that this should take the form of a new criminal offence. In addressing the issue, this chapter considers the harm sought to be remedied by the new offence, the inadequacies in the current law, and the reasons that criminal (as distinct from civil) penalties are justified. The legal elements of the new offence are also examined.

12.3 Biomedical technology currently enables genetic testing to be performed on minute bodily samples. The polymerase chain reaction method of genetic testing enables DNA from a single cell to be amplified many times to produce quantities of genetic material suitable for testing. Further, the ongoing development of automated 'DNA chip' technology will make it possible to obtain information about numerous genetic mutations simultaneously in a single test procedure.¹

12.4 Human genetic samples may be found nearly everywhere. Genetic information may be derived from easily accessible bodily samples such as hair follicles (humans shed hundreds of hairs every day), saliva left on a glass or cigarette, cheek

¹ Also known as 'gene chips', 'biochips' and 'DNA microarrays'. See S Moore, 'Making Chips to Probe Genes', *IEEE Spectrum*, 1 March 2001, 54.

cells left on a toothbrush, sloughed cells deposited on an item of clothing, or mucus in a tissue.²

12.5 In combination, the power of biomedical technology and the ubiquity of human genetic samples leaves open the potential for bodily samples to be taken and tested without the knowledge or consent of the individual to whom they relate. In this chapter, this is referred to as non-consensual genetic testing.

12.6 Discussion of ‘genetic trophy hunters’ has become increasingly common in the media. Fear of genetic trophy hunters has been cited as the reason that bodyguards of former United States President, Bill Clinton, collected a pint glass after he had drunk from it in a British pub, to ensure that his DNA could not be obtained.³ Newspapers have also reported a ‘plot’ to obtain a sample of Prince Harry’s DNA—a girl would be used to set a ‘honey trap’ and pluck a strand of hair from his head.⁴ The media also reported extensively on claims that American movie mogul, Kirk Kerkorian, obtained DNA from dental floss in Steve Bing’s garbage bin in an attempt to prove that Bing had fathered a child by Kerkorian’s ex-wife. Bing in turn sued Kerkorian for trespass.⁵

12.7 A link between ease of access to genetic testing and the potential for improper use of these services was made in a number of submissions. For example, Kathy Liddell stated:

Genetic information might be collected surreptitiously (eg from a sample of skin, hair roots or saliva) for unauthorised purposes. The risk of this is increased by the availability of over-the-internet and over-the-counter genetic services. Although this conduct would generally breach the *Privacy Act* or the common law, there is an argument that criminal penalties should apply.⁶

12.8 The Office of the Federal Privacy Commissioner stated that it was concerned about

the problems involved in a person’s obtaining genetic information about an individual for the wrong reasons without the individual’s knowledge or consent, whether by mail-order or through DIY testing. This issue raises, among others, proof of *bona fides*, proof of the identity of the person requesting the test and possible criminal sanctions for the unlawful use of genetic information without the knowledge and consent of the individual. The [Privacy Act] provides general protection in many of these circumstances but developments in this area should be monitored closely.⁷

12.9 The Department of Health and Ageing agreed that ‘over-the-counter’ testing created the potential for genetic testing of an individual without the individual’s knowledge or consent.

2 On the ubiquity of human genetic samples, see Ch 3.

3 G Hinsliff, ‘Bid to Outlaw DNA Trophy Hunters’, *The Observer* (London), 3 March 2002.

4 Palace Silent over Plot to Obtain DNA from Prince Harry’, *Canberra Times*, 16 December 2002.

5 Paternity Tests Have Exploded with a Bing’, *The Sun-Herald*, 26 May 2002.

6 K Liddell, *Submission G141*, 23 March 2002.

7 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

Current DNA profiling technologies enable accurate results from very small samples such as a hair and these can be easily obtained without a person being aware of it. Regulation of this practice raises difficulties because testing services are available internationally (often through internet marketing). Nonetheless, commercial and unaccredited laboratories exist in Australia and regulation of 'DNA theft' is an important issue which must be addressed.⁸

The harm of non-consensual genetic testing

12.10 In determining whether the non-consensual collection and use of bodily samples for the purpose of genetic testing should be proscribed by law and, if so, how, it is necessary to examine the harm that may arise from this conduct.

12.11 The harm caused by non-consensual genetic testing may be categorised in various ways, depending on whether one looks at the collection of the genetic sample, the testing of the sample to derive genetic information from it, or the possible uses of the information so derived.

12.12 The collection of a sample may, in some circumstances, involve a physical harm or a trespass to the person (a battery), as when a person is held down and a bodily sample is taken by force. Collection may also result in emotional harm. Emotional harm may result from situations where, from the perspective of the individual concerned, intimate bodily samples (such as menstrual blood or semen) are taken, or kinship or identity is questioned.

12.13 The most obvious harm arising from testing of the sample is the intrusion on basic human dignity and autonomy. The harm may be also characterised as involving a breach of information privacy.⁹ Genetic testing may result in the disclosure of sensitive personal information of many kinds. Testing can reveal information about the present and future health of an individual, an individual's identity, or his or her parentage or kinship. The fact that harm may be caused by the non-consensual disclosure of these kinds of information is recognised by laws that proscribe disclosure in other contexts, including legal and statutory duties of confidentiality, and information and health privacy legislation.

12.14 The possible uses of the information derived from non-consensual testing may also give rise to harm, including harm caused:

- by the use of genetic information by employers, insurers and others for discriminatory purposes;
- to individuals who involuntarily learn about their long-term health prognosis and other physical and behavioural characteristics, in breach of their 'right not to know';¹⁰

⁸ Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

⁹ As discussed in Ch 7, 'information privacy' can be defined as the right of an individual to control the collection, use and disclosure of information relating to them (personal information).

¹⁰ The right not to know is discussed in Ch 7.

- by media publicity about an individual's genetic characteristics, especially where that individual is a celebrity or otherwise newsworthy;
- by the use of genetic information by police in criminal proceedings or by litigants in civil proceedings; and
- by disruption to family relationships and harmony where parentage or other kinship information is disclosed.

12.15 The Inquiry is of the view that non-consensual genetic testing may compromise significant interests, and that these interests merit protection under the law. To do so would reflect the ethical consideration of respect for individual freedom and autonomy, which is the basis for protecting privacy and providing remedies for the harm caused by breaches of privacy.

Application of existing Australian law

12.16 Given that the conduct of non-consensual genetic testing has a clear potential to cause harm, it is necessary to ask to what extent existing Australian law offers protection from such harms.

12.17 The legal position in the United Kingdom has been considered by the Human Genetics Commission (HGC). In its May 2002 report, the HGC concluded that there are scenarios where current legal remedies may not offer sufficient protection against breach of an individual's genetic privacy. For example:

- X takes Y's beer glass and obtains an analysis of his DNA. He or she then sells to a newspaper the information that Y has a particular genetic condition.
- X de-encrypts anonymised genetic information about Y from a research study for some wrongful purpose.
- X obtains a sample from Child A, for whom he has no parental responsibility, in order to ascertain whether he is the father of the child.¹¹

12.18 In Australia, some legal protection against the non-consensual collection and use of bodily samples exists under current law, for example, through the common law tort of trespass to the person, the criminal law of assault, property law, and privacy legislation. However, this protection is limited.

12.19 Any touching of a person's body without consent may constitute a trespass. In theory, the person may seek a civil remedy through the courts against the trespasser. In practice, civil remedies in tort are costly to obtain. Non-consensual touching may also constitute an assault in criminal law. However, where the assault is minor or seen as little more than a technical breach of the law, police and prosecutors may be reluctant to take action.

11 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 60.

12.20 In any case, battery and assault are not relevant to the collection of ‘discarded’ genetic material, such as hair from combs, saliva from a glass, cheek cells from a toothbrush, or mucus from a tissue.

12.21 The taking of such genetic samples is unlikely to constitute theft. While current law recognises possessory rights and limited ownership interests in preserved samples of tissue, no proprietary rights are vested in the individual from whom samples are taken, such as might enable that individual to bring an action against others who deal with the samples.¹²

12.22 The *Privacy Act 1988* (Cth) (*Privacy Act*) and similar state and territory legislation provide only limited protection against the collection and testing of genetic samples without consent in the kinds of circumstances identified by the HGC. This is for several reasons. First, except in New South Wales,¹³ information and health privacy legislation does not currently apply to genetic samples, as opposed to the genetic information derived from them. The collection of a genetic sample without consent (for example, from a beer glass) does not breach the federal *Privacy Act*¹⁴—although it might do so if the Inquiry’s recommendations in relation to extending the *Privacy Act* to cover genetic samples are implemented.¹⁵

12.23 Second, leaving aside the distinction between a genetic sample and genetic information derived from it, the coverage of the *Privacy Act* is limited because acts done, or practices engaged in, by individuals are exempt if done or engaged in ‘other than in the course of a business carried on by the individual’.¹⁶ Further, the National Privacy Principles (NPPs) do not apply to the collection, use or disclosure of personal information by an individual ‘only for the purposes of, or in connection with, his or her personal, family or household affairs’.¹⁷ Arguably, many of the circumstances in which genetic testing might take place surreptitiously will have purposes that relate to an individual’s personal, family or household affairs. These might include purposes relating to family health, personal identity, or ‘peace of mind’ parentage testing.

12.24 In the circumstances identified by the HGC in the three examples above, the acts of person X generally will not be regulated by the *Privacy Act*, unless done in the course of a business. Even then, if X is a journalist and the collection and testing of the sample is done in the course of journalism (for example, for the purpose of a news story relating to a celebrity paternity dispute), the acts may be exempt under other provisions of the *Privacy Act* dealing specifically with media organisations and journalism.¹⁸

12 See Ch 20.

13 *Privacy and Personal Information Protection Act 1998* (NSW) s 4.

14 The Inquiry proposes that the *Privacy Act* should be amended to define ‘personal information’ as including bodily samples from an individual whose identity is apparent or can reasonably be ascertained from the sample. See Ch 8.

15 See Ch 8.

16 *Privacy Act 1988* (Cth) s 7B(1).

17 *Ibid* s 16E.

18 *Ibid* s 7B(4).

12.25 In order to derive information from a genetic sample, an individual would ordinarily need to submit the genetic sample to a laboratory for testing. It is relevant, therefore, to consider the obligations of the laboratory under privacy legislation. At least in the case of a private sector organisation, the collection, use and disclosure of personal information by the laboratory will be subject to the NPPs. However, the laboratory may not know the identity of the individual from whom the sample was derived. If so, the laboratory will not be dealing with ‘personal information’ covered by the *Privacy Act*, even though the individual who submitted the sample for testing knows from whom it came.¹⁹ Even assuming the laboratory, by analysing and reporting the results of testing, is collecting and disclosing personal information in terms of the *Privacy Act* and the NPPs, the legal protection extended to the individual from whom the sample was derived appears limited.²⁰

12.26 The Inquiry has concluded that the current law is inadequate to meet the harms arising from non-consensual genetic testing and that a legislative response is required to address this situation.

Options for regulating non-consensual genetic testing

12.27 If unauthorised non-consensual genetic testing is to be proscribed by law, how should the practice be regulated? In particular, should the relevant conduct be subject to criminal or civil remedies,²¹ or both (as is the case, for example, with battery)?²²

12.28 Decisions about the form and level of penalty to be applied to proscribed conduct should depend on the purpose of the penalty, as well as the area of activity, the type of wrongdoer and the nature of the wrongdoing. The purposes of penalties may include punishment or retribution, social condemnation, deterrence, protection of third parties or the public at large, and payment of reparation or compensation.²³

12.29 The main purposes of criminal law are traditionally considered to be deterrence and punishment. The aim of social condemnation, or stigma, traditionally applies more to criminal than civil penalties.²⁴ Civil sanctions generally have the practical function of discouraging undesirable behaviour by imposing a financial cost on it,²⁵ such as by imposing an obligation on a wrongdoer to compensate the injured party for loss.

19 Ibid s 6(1). See Ch 7 on the definition of ‘personal information’ under the *Privacy Act*.

20 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Ch 5, fn 78.

21 A ‘civil penalty’ is one imposed by courts applying civil rather than criminal court processes: Australian Law Reform Commission, *Principled Regulation: Federal Civil and Administrative Penalties in Australia*, Report 95 (2002), ALRC, Sydney [2.45].

22 A battery may constitute both a tort or civil wrong (trespass to the person) and a criminal offence (assault).

23 Australian Law Reform Commission, *Principled Regulation: Federal Civil and Administrative Penalties in Australia*, Report 95 (2002), ALRC, Sydney [3.5].

24 Ibid [3.5]–[3.6].

25 See J Coffee, ‘Paradigms Lost; The Blurring of the Criminal and Civil Law Models — And What Can Be Done about It?’ (1992) 101 *Yale Law Journal* 1875.

12.30 The criminal law covers a vast array of activities and offences. These range from murder and assault to offensive language and jay-walking. In the federal sphere, criminal law includes customs infringements and breaches of consumer protection laws.²⁶ Criminal law is not only concerned with serious offences. There are, for example, scores of low-level record-keeping and information offences, which are treated criminally in many regulatory regimes.²⁷ In fact, outside areas of regulatory law it is relatively rare for the conduct of individuals to be made subject to non-criminal penalties.²⁸

12.31 One guide to the appropriateness, or otherwise, of subjecting non-consensual genetic testing to criminal penalty is the criminalisation of analogous conduct. While it is difficult to identify close analogies, existing crimes legislation (in addition to stealing offences) contains offences relating to:

- unauthorised supply of forensic material for a DNA database,²⁹
- unauthorised access to data held in computers,³⁰
- ‘peeping or prying’ near buildings,³¹
- stalking or intimidation;³² and
- interference with human remains.³³

12.32 Analogous offences are also contained in other legislation. For example there are offences in the Human Tissue Acts concerning removing tissue from persons, living or dead, without consent or authority.³⁴ The unauthorised disclosure of health information obtained by public sector health administrators and employees in the course of their employment is also subject to criminal penalty.³⁵

12.33 Alternatively, new civil remedies might be created to deter non-consensual genetic testing. This approach could be taken instead of, or in addition to, the creation of any new criminal offence. One such approach would be to amend the *Privacy Act* to ensure that the conduct involved in non-consensual genetic testing constitutes an

26 See D Brown and others, *Criminal Laws: Materials and Commentary on Criminal Laws and Process of New South Wales* (2001) The Federation Press, Sydney.

27 A Ashworth, ‘Is the Criminal Law a Lost Cause?’ (2000) 116 *Law Quarterly Review* 225, 243.

28 Regulatory law concerns the way that governments regulate private sector activity or otherwise intervene in the operation of different areas of society outside of traditional criminal law. Criminal regulatory offences include a number of traditional crimes such as fraud or obtaining benefits by deception, but also offences that are not so obviously criminal in their nature, such as failing to provide certain types of information or failing to meet a certain licensing standard: Australian Law Reform Commission, *Principled Regulation: Federal Civil and Administrative Penalties in Australia*, Report 95 (2002), ALRC, Sydney [2.8].

29 *Crimes Act 1914* (Cth) s 23YDAD.

30 *Crimes Act 1900* (NSW) s 308H; *Summary Offences Act 1966* (Vic) s 9A.

31 *Crimes Act 1900* (NSW) s 547C.

32 *Ibid* s 562AB; *Crimes Act 1958* (Vic) s 21A.

33 *Crimes Act 1900* (NSW) s 81C.

34 See eg *Human Tissue Act 1983* (NSW) s 36 and cognate legislation in other jurisdictions.

35 See eg *Health Administration Act 1982* (NSW) s 22; *Health Services Act 1988* (Vic) s 141.

interference with privacy in a broader range of circumstances, enabling the individuals concerned to seek compensation under the Act. This could be backed up by legislation preventing the use of non-consensual genetic test results in court proceedings.³⁶

A new criminal offence

12.34 Serious privacy and ethical concerns arise from non-consensual testing. The range of genetic information that can be derived from a bodily sample is wide and may have great importance and sensitivity for individuals, their families and their genetic relatives.

12.35 In the United Kingdom, the need for a new criminal offence was recently acknowledged by the HGC, which recommended that:

consideration be given to the creation of a criminal offence of the non-consensual or deceitful obtaining and/or analysis of personal genetic information for non-medical purposes.³⁷

12.36 Many submissions to the Inquiry supported the idea that some kinds of non-consensual genetic testing should be criminalised.³⁸ For example, the Victorian Health Services Commissioner stated:

Privacy legislation has an important part to play, but may not be sufficient on its own. Genetic testing should never be allowed without consent, unless there are very special circumstances.³⁹

12.37 The New South Wales Legal Aid Commission stated:

The main concern is the protection of the subject's privacy, and their privacy is breached by the fact of another person having obtained the genetic information without consent, even if they do not use or disclose the information. This is particularly the case as it is most likely that a person seeking to obtain genetic

³⁶ L Skene, *Consultation*, Sydney, 27 June 2002.

³⁷ Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 62.

³⁸ K Liddell, *Submission G141*, 23 March 2002; Sydney IVF Limited, *Submission G062*, 14 January 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Commonwealth Attorney-General's Department, *Submission G228*, 12 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Genetic Technologies Corporation Pty Ltd, *Submission G245*, 19 December 2002; Sydney IVF Limited, *Submission G246*, 19 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; A Gesche, *Submission G264*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; National Legal Aid, *Submission G314*, 19 February 2003. The HGSA submitted that 'consent to medical genetic testing should not be brought within the scope of the criminal law': Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

³⁹ Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

information about a subject without their consent will be a family member or someone else close to the subject. In most cases the information will be of little interest to people outside the subject's immediate circle, but access to the information by a friend or relative has the potential to affect the subject's personal relationships.⁴⁰

12.38 The Centre for Law and Genetics expressed strong support for the proposed criminal offence. In the view of the Centre, the proposal 'addresses most of the potential abuses of DNA'.⁴¹ The Commonwealth Attorney-General's Department confirmed that the development of a model offence relating to non-consensual genetic testing is feasible.⁴²

12.39 However, there was significant opposition to the new criminal offence from some quarters. For example, opposition was voiced by organisations and individuals who also opposed the Inquiry's proposed reforms in relation to the regulation of parentage and other kinship testing⁴³ on the basis that such reforms are a barrier to the 'right' of fathers to know about the parentage of their children.⁴⁴ For example, the Men's Confraternity WA stated:

The suggestion by the [Inquiry] that a father be prosecuted and have a criminal conviction recorded against him if he has a DNA test conducted on his child, if doubts arise as to his paternity, is in our view an outrage and borders on the obscene. By refusing a man the right to access his child's DNA, he is being denied one of his basic human rights.⁴⁵

12.40 Jonathon Baxter noted that the proposed new criminal offence would make it a criminal offence 'in many cases for a father to determine his own paternity, without first applying to the family court'. He asked:

Given the family court's long history of bias against fathers, particularly in relation to custody matters; given the vindictive nature of many ex-wives who would no doubt use such an application by the father to damage his relationship with his children; and finally, given the publicly stated, extreme views of [Chief Justice] Nicholson on the subject ... how can you possibly make such a proposal?⁴⁶

12.41 Another submission stated that:

to make it a criminal offence for a man to disprove an allegation of paternity is to deny that man his right to prove himself innocent of an allegation against him.⁴⁷

40 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

41 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

42 Commonwealth Attorney-General's Department, *Submission G228*, 12 December 2002.

43 J Dezordi, *Submission G180*, 4 September 2002; Men's Rights Agency, *Submission G213*, 29 November 2002; Men's Confraternity WA Inc, *Submission G234*, 17 December 2002; J Baxter, *Submission G280*, 27 December 2002; A Unger, *Submission G192*, 18 November 2002; R Kane, *Submission G179*, 4 September 2002.

44 See Ch 35.

45 Men's Confraternity WA Inc, *Submission G234*, 17 December 2002.

46 J Baxter, *Submission G280*, 27 December 2002.

47 A Unger, *Submission G192*, 18 November 2002.

12.42 As discussed in Chapter 35, the Inquiry recommends that, in general, parentage testing of children should be performed only with the written consent of all persons with parental responsibility for the child, or pursuant to a court order.⁴⁸ It can be assumed that those organisations and individuals who oppose these recommendations would also oppose the new criminal offence—at least in so far as it relates to non-consensual parentage testing by parents or putative parents—even if this was not expressly stated in submissions.

12.43 A number of recommendations contained in this Report address concerns about non-consensual testing. The recommendations in Chapter 11 in relation to accreditation standards and the regulation of direct to the public genetic testing are also intended, in part, to address concerns about non-consensual testing. As noted above, Chapter 35 focuses on the need to protect both adults and children from non-consensual parentage testing. However, these measures may not be sufficient.

Inquiry's views

12.44 The Inquiry considers that there are compelling arguments in favour of creating a new criminal offence. Given the wide range of situations in which harm may be caused by non-consensual testing, the offence should cover a broad range of testing, rather than be confined to particular categories of testing, such as parentage or kinship testing.

12.45 In the Inquiry's view, criminalising non-consensual genetic testing would not be disproportionate to the potential mischief to be avoided. In particular, given the difficulties in regulating access to, and the conduct of, genetic testing laboratories overseas, the threat of criminal prosecution of individuals in Australia may be the only effective deterrent. Useful analogies may be drawn with other acts that are already subject to criminal sanction.

12.46 The Inquiry does not consider that the *Privacy Act* should be the primary vehicle by which non-consensual genetic testing should be prohibited. It is appropriate that, in some circumstances, the use and disclosure of genetic information derived from non-consensual genetic testing constitute an interference with privacy under that Act. However, the focus of the *Privacy Act* is, and should remain, on regulating the practices of government and business rather than individuals in their private capacities. Further, where the *Privacy Act* is breached, the enforcement mechanisms may be seen as providing an inadequate sanction for non-consensual genetic testing.⁴⁹

48 See Recommendation 35–9.

49 Enforcement of the Act is generally through resolution of individual complaints lodged with the Privacy Commissioner. When the Privacy Commissioner determines that a person's privacy has been interfered with, the Commissioner can impose a number of penalties, including a declaration that the organisation should not repeat or continue the offending conduct, a request that the organisation redress the loss or damage incurred, or a request that the organisation pay compensation for any loss or damage incurred. The decisions of the Privacy Commissioner are enforceable by the Federal Court or the Federal Magistrates Court. See *Privacy Act 1988* (Cth) Div 2–3. The Commonwealth Department of Health and Ageing noted that the Australian Health Ministers' Advisory Council (AHMAC) National Health Privacy Code (see Ch 7) may include penalties for non-consensual testing: Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

The elements of the offence

12.47 It is easier to recommend the creation of a new criminal offence in principle than to determine how it should be drafted and what exceptions should be recognised. These difficulties were noted by the HGC, which observed in respect of its recommendation that:

[t]here are a number of aspects that merit further close examination. For example, it will be important to ensure that any new offence does not interfere with appropriate and lawful non-consensual use of genetic material or genetic information by the police or courts. We would also not wish to see any new offence that might inhibit the use of genetic testing in medical and research settings.⁵⁰

12.48 The Inquiry's views on the approach that should be taken are discussed below. The elements of the new criminal offence which need to be determined are: what constitutes the offending conduct, who are the potential offenders, what should be the required fault element of the offence, what exceptions should be recognised, and what penalties should apply?

The offending conduct

12.49 The offending conduct (the *actus reus*) might encompass the taking of the sample or deceit in the taking of the sample, the submitting of the sample for testing, testing the sample, using or disclosing the results of testing, or a combination of these acts.

12.50 The New South Wales Legal Aid Commission submitted that

the conduct constituting the offence should be the taking of the sample for the purpose of genetic testing, or submitting the sample for testing without consent, rather than improper use or disclosure of information obtained from the sample.⁵¹

12.51 The Department of Human Services South Australia stated that the offending conduct should specifically include the 'publication' or 'use' of an identified individual's results without consent from the individual from whom the sample was obtained.⁵²

12.52 The Inquiry's view is that the offence should focus on submitting the sample for testing and testing the sample. 'Genetic testing' will need to be defined for these purposes. Criminalising the taking of the sample is inappropriate given the ubiquity of genetic samples. The harm is not simply in the taking of the sample (leaving aside situations where there is a battery). For example, if an individual chooses to collect a lock of hair found on a hairdresser's floor, it is difficult to argue that any more harm is

50 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 62.

51 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

52 Department of Human Services South Australia, *Submission G288*, 23 December 2002. National Legal Aid expressed similar views: National Legal Aid, *Submission G314*, 19 February 2003.

done to the person to whom the sample relates than taking a photograph of them without their consent—which is not an offence unless followed by an unlawful use.⁵³

12.53 It has been suggested that the focus of regulation should be on the unauthorised uses of the information derived from testing, especially for financial advantage.⁵⁴ However, there would be significant difficulties involved in defining the unauthorised uses, given the range of information that may be derived from samples and the spectrum of possible uses. Some of these uses would, in any case, be proscribed by existing law, such as where the publication of genetic information is defamatory.

12.54 The Inquiry considers that harm is done to individuals who are tested without their knowledge and consent, even if the testing is done only to satisfy another individual's curiosity and there is no further use of the information obtained. Once the information comes into existence, the potential for improper use and disclosure will exist. By way of analogy, it is an offence to 'hack' into another person's computer,⁵⁵ or to 'tap' their telephone,⁵⁶ without lawful authority, even if no interesting information is obtained in the process.

The offenders

12.55 The primary targets of the criminal offence should be individuals or bodies corporate who submit samples for testing, with the requisite fault element. It may also be desirable to target individuals or bodies corporate who conduct the testing, such as laboratories and their employees, agents or officers. Where the offender is a body corporate, the physical elements of the offence will be attributed to the body corporate where committed by an employee, agent or officer acting within the actual or apparent scope of his or her employment, or within his or her actual or apparent authority.⁵⁷

The fault element

12.56 The concept of criminality involves the notion of individual culpability.⁵⁸ Consistently with the approach taken to the drafting of other criminal offences such as stalking, the fault elements should be those of intention or reckless indifference. The Inquiry does not intend that mere error or inadvertence in the conduct of genetic testing should be criminalised.

53 L Skene, *Consultation*, Sydney, 27 June 2002.

54 *Ibid.*

55 *Crimes Act 1900* (NSW) s 308H; *Summary Offences Act 1966* (Vic) s 9A.

56 *Telecommunications (Interception) Act 1979* (Cth) s 7.

57 See *Criminal Code Act 1995* (Cth) ss 12.1, 12.2.

58 Australian Law Reform Commission, *Principled Regulation: Federal Civil and Administrative Penalties in Australia*, Report 95 (2002), ALRC, Sydney [2.9].

12.57 In relation to individuals or bodies corporate who submit samples for testing, the Inquiry suggests that the fault elements of the offence should be that the offending individual or body corporate⁵⁹ intends that the sample be tested and knows that the individual from whom the sample has been taken has not consented to testing or is reckless as to that fact,⁶⁰ and there is no lawful authority for the test.

12.58 In relation to laboratories, their employees, agents or officers, the fault elements should be that the offending body corporate or individual knows that the individual from whom the sample has been taken has not consented to testing, or is reckless as to that fact, and there is no lawful authority for the test.

12.59 In this context, the Cancer Council of Victoria Cancer Genetics Advisory Committee noted that human errors occur even in well—run and reputable laboratories and submitted that:

It would be an injustice if a laboratory scientist or director were prosecuted for unknowingly performing a genetic test without first checking that it was accompanied by a signed consent form. The intention of such legislation would be to stop reckless testing and not to penalise individuals or laboratories for occasional errors.⁶¹

12.60 Genetic Technologies recognised that there may be a need for criminal sanctions to protect individuals from non-consensual testing.⁶² However, it submitted that laboratories should not be subject to offence provisions.

It is our view that it is the individual commissioning the test who takes responsibility for it. It would be significantly detrimental to the parentage testing industry to make a laboratory liable to complicity in the commission of a crime. Parentage testing is a private matter. Furthermore, there will always be means of obtaining samples in a non-consensual way for those determined to do so.⁶³

12.61 The Inquiry disagrees with this view, in so far as it implies that laboratories have no role to play in ensuring that appropriate consent is obtained. As discussed in Chapter 11, laboratory accreditation standards relating to clinical genetic testing already place some obligations on laboratories in this regard. The Inquiry has recommended that responsible bodies should continue to develop accreditation standards relating to ethical concerns.

12.62 In the Inquiry's view, to require that laboratories, and those that manage them, not be 'recklessly indifferent' as to the circumstances in which they receive samples for testing does not impose an unduly onerous obligation. The standard of scrutiny expected of laboratories will depend on the source of the request for testing. For example, where a laboratory receives a request for a routine medical test from a

59 Where the offender is a body corporate, the fault elements of the offence will be attributed to the body corporate where the body corporate 'expressly, tacitly or impliedly authorised or permitted the commission of the offence': *Criminal Code Act 1995* (Cth) s 12.3(1).

60 A person has knowledge of a circumstance or a result if he or she is aware that it exists or will exist in the ordinary course of events: *Ibid*, s 5.3.

61 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

62 Genetic Technologies Corporation Pty Ltd, *Submission G245*, 19 December 2002.

63 *Ibid*.

medical practitioner, the laboratory need not ‘look behind’ the request. More scrutiny may be needed where testing, such as parentage testing, is provided directly to the public. In any case, laboratories that take appropriate steps to ensure that relevant consent forms have been signed by the appropriate persons (in circumstances that raise no suspicion about their validity) would not commit any offence.

Defining the ambit of the offence

12.63 In framing the new offence, care will need to be taken to ensure that acceptable genetic testing practices do not fall within its ambit. Consideration will need to be given to what constitutes ‘consent’. For example, where the sample has been taken from a child, consent may need to be framed in terms of the proposals set out in Chapter 35, which deal with consent to the collection and testing of a child’s genetic sample for the purpose of determining parentage. Where samples are being tested for medical research purposes, it should be sufficient that consent has been waived by a Human Research Ethics Committee.⁶⁴

12.64 Legitimate genetic testing practices that should not be caught by the new criminal offence include genetic testing:

- ordered by medical practitioners in order to assist in the treatment of patients (for example, relying on implied consent);
- for medical research purposes, especially where a Human Research Ethics Committee, in granting ethical approval for a research proposal, has waived consent requirements (see Chapter 15);
- for parentage testing involving children—with appropriate parental consent or pursuant to a court order (see Chapter 35);
- for law enforcement purposes pursuant to legislative authority or a court order (see Chapter 39).

12.65 In this context, the Human Genetics Society of Australasia noted that, while it supported legal sanctions in relation to non-consensual paternity testing, it was very concerned about any suggestion that there might be legal sanctions for non-consensual genetic testing carried out by a doctor in the course of health care.

Not only is it difficult to define what is, and what is not, genetic testing but there are also various forms of consent and various settings in which genetic testing is carried out. While accepting the need for consent to genetic testing as a general principle, the HGSA considers that consent to medical genetic testing should not be brought within the scope of the criminal law.⁶⁵

64 See Ch 15.

65 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

12.66 The Inquiry considers that the new criminal offence should incorporate reference to a ‘lawful authority’ defence. The *Criminal Code Act 1995* (Cth) provides that a person is not criminally responsible for an offence if the conduct constituting the offence is justified or excused by or under a law.⁶⁶

12.67 Other legislation may provide a useful guide to exceptions that may be required. For example, the Victorian anti-stalking provision does not apply to conduct engaged in by a person performing official duties for the purposes of the enforcement of the criminal law, the administration of any Act, the enforcement of a law imposing a pecuniary penalty, the execution of a warrant, or the protection of the public revenue.⁶⁷

12.68 Similarly, the use and disclosure principle in the *Privacy Act* (NPP 2) contains exceptions relating to use and disclosure for research relevant to public health or public safety; serious threats to an individual’s life, health or safety; serious threats to public health or public safety; use by law enforcement agencies in investigation, prosecution and court proceedings; and other uses and disclosures ‘required or authorised by or under other law’.

The penalty

12.69 The Inquiry has no concluded view on the range or level of penalty most appropriate for breach of the proposed new criminal offence. However, the available penalties should reflect the wide spectrum of circumstances in which non-consensual genetic testing may take place. The penalty where a media organisation uses the test results for financial benefit should differ from that imposed on an individual who is concerned about the health of a relative. The penalties for testing for entirely prurient reasons should differ from those where there is some valid reason for wanting to obtain a genetic test result.

12.70 By way of comparison with broadly analogous offences in New South Wales, the maximum penalties for:

- unauthorised access to data held in computers is two years imprisonment;⁶⁸
- peeping or prying is three months imprisonment or a fine of two penalty units (currently \$220);⁶⁹ and
- stalking or intimidation is five years or a fine of 50 penalty units (currently \$5500).⁷⁰

66 *Criminal Code Act 1995* (Cth) s 10.5.

67 *Crimes Act 1958* (Vic) s 21A(4).

68 *Crimes Act 1900* (NSW) s 308H; *Summary Offences Act 1966* (Vic) s 9A.

69 *Crimes Act 1900* (NSW) s 547C.

70 *Ibid* s 562AB; *Crimes Act 1958* (Vic) s 21A.

12.71 The Department of Health Western Australia, which gave in principle support for the development of a new model criminal offence, considered that it may be sufficient to impose pecuniary penalties and to provide for courts to issue injunctions restraining repetition.⁷¹

12.72 The possible application of proceeds of crime legislation to money or other property derived from the commission of the new criminal offence should also be considered.⁷² Such legislation may, for example, allow profits obtained by a 'genetic trophy hunter' to be confiscated.

12.73 The Inquiry considers that the Standing Committee of Attorneys-General is the appropriate body to initiate, through an appropriate committee,⁷³ the drafting of a model criminal offence relating to non-consensual genetic testing, intended for enactment into Commonwealth, state and territory law.⁷⁴

Recommendation 12–1. The Standing Committee of Attorneys-General should develop a model criminal offence relating to non-consensual genetic testing, for enactment into Commonwealth, state and territory law. Criminal liability should attach to any individual or corporation that, without lawful authority, submits a sample for genetic testing, or conducts genetic testing on a sample, knowing (or recklessly indifferent to the fact) that the individual from whom the sample has been taken did not consent to such testing.

71 Department of Health Western Australia, *Submission G271*, 23 December 2002.

72 See *Proceeds of Crime Act 2002* (Cth).

73 For example, the forensic procedures provisions of the *Crimes Act 1914* (Cth) (Pt 1D) were developed for the Standing Committee of Attorneys-General by the Model Criminal Code Officers Committee.

74 The Commonwealth Attorney-General's Department suggested that a new criminal offence could be developed (in the context of improvements to the Model Forensic Procedures Bill) by the Joint Standing Committee of Attorneys-General and the Australasian Police Ministers Council Working Group: Commonwealth Attorney-General's Department, *Submission G228*, 12 December 2002.

Part D. Human Genetic Research

13. The Regulation of Human Genetic Research

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Introduction

13.1 This chapter introduces Part D, which considers ethical, privacy and related issues concerning the use of genetic samples and information in the conduct of research involving humans (referred to as human genetic research).

13.2 This chapter briefly summarises the present regulatory framework for the ethical conduct of research. This framework is centred on the National Health and Medical Research Council's (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement)¹ and on review of research proposals by Human Research Ethics Committees (HRECs).²

13.3 Some of the complexities involved in regulating human genetic research were summarised by William Lowrance in a recent report for the Nuffield Trust:

Debates in the coming era over genetic information are likely to severely test the legal concepts of privacy and autonomy, and also the fundamentals of bioethics, which are already stretched thin. Informational privacy will have to be distinguished from decisional autonomy. Progress will have to be made in sorting-out the rights of

1 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

2 The role of HRECs is discussed in more detail in Ch 17.

relatives of data-subjects. And the public health aspects of genetics will become more prominent and have to be dealt with.³

13.4 The adequacy or otherwise of the existing regulatory framework has been the subject of much comment in the course of the Inquiry. The nature of this comment, and the issues raised in consultations and submissions, are reflected in this chapter and those that follow.

What is human genetic research?

13.5 Human genetic research is broadly defined in the National Statement as a process which

enhances our understanding of how genes and environmental factors interact to influence the health of individuals and populations and in doing so, generates knowledge with the potential to improve individual and community health.⁴

13.6 Some human genetic research can be conducted without the need for genetic information that can be related to specific persons or communities. For example, research on molecular processes can explore genetic processes and the production of enzymes or proteins. Similarly, research can use anonymous population data to examine patterns of human inheritance of disease or conditions.

13.7 Unless otherwise indicated, this Report uses human genetic research to refer to any research that uses genetic samples or genetic information, whether or not those samples or information are identified, potentially identifiable, or de-identified.⁵ Such research must involve *human* genetics and does not include the use of human tissue samples in order to study, for example, the genetics of infectious agents such as viruses.

13.8 When discussing the protection of privacy interests in genetic samples and information, the chapters focus on human genetic research that needs to use or will develop information that is either identified or potentially identifiable. Privacy interests are not generally threatened where the identity of an individual is not apparent or cannot reasonably be ascertained from information.⁶

13.9 However, ethical concerns may be raised by the collection or use of genetic samples and information in human research, even where the samples or information are de-identified. Basic ethical principles such as integrity and respect for persons⁷ should

3 W Lowrance, *Learning from Experience: Privacy and the Secondary Use of Data in Health Research* (2002), The Nuffield Trust, London, 59.

4 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, Ch 16.

5 As these terms are used in the National Statement: *Ibid* 9.

6 See *Privacy Act 1988* (Cth) definition of 'personal information': s 6.

7 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [1.1]–[1.2].

be observed even where samples or information are to be de-identified⁸ or where the research involves a collectivity. For example, one consequence of de-identification is that clinically relevant information produced by research cannot be related to participants or their families.⁹ On the other hand, some genetic disorders are so rare that families might be identified from the disclosure of results in de-identified form.¹⁰

The importance of human genetic research

We all share the basic human genome, although there are individual variations which distinguish us from other people. ... This sharing of our genetic constitution not only gives rise to opportunities to help others but it also highlights our common interest in the fruits of medically-based genetic research.¹¹

13.10 The 1999 Wills Report, which resulted from a major strategic review of health and medical research in Australia, referred to the need to take advantage of advances in biotechnology to improve the health of the Australian population, to build the economy and to create jobs.¹²

13.11 Human genetic research generates knowledge with the potential to improve individual and community health. Research can also reveal information about an individual's susceptibility to disease and hence about the individual's future health. Such information may be of interest and benefit to research participants especially if preventive strategies exist.¹³

13.12 The completion of the first mapping of the human genome under the Human Genome Project has opened huge potential for research into the ways in which genes relate to human conditions, capacities, diseases, impairments and susceptibilities. The scope of genetic research may be illustrated by reference to the current activities of just one major Australian genetic research organisation. The Murdoch Childrens Research Institute is conducting research into, among other things, the underlying genetic causes of neuromuscular disorders, inherited hearing loss, attention deficit hyperactivity disorder, cancers, ataxias and addiction.¹⁴

13.13 Although exact figures are not available, it is likely that human genetic research will become an increasingly important component of medical research generally. One indication of this is that clinical trials of new drugs increasingly include

8 The concept of de-identification and its relevance to issues of consent to participation in research is discussed in Ch 15. Issues concerning the de-identification of genetic samples and the coverage of the *Privacy Act 1988* (Cth) are discussed in Ch 8.

9 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.6].

10 *Ibid* [16.5].

11 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 38.

12 Health and Medical Research Strategic Review, *The Virtuous Cycle, Working Together for Health and Medical Research* (1999), Commonwealth of Australia, Canberra, 1.

13 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [6.5].

14 Murdoch Children's Research Institute, *Gene Identification and Expression*, <www.murdoch.rch.unimelb.edu.au/pages/lab/gene_identification/overview.html>, 25 June 2002.

genetic sub-studies.¹⁵ It has long been known that drug toxicity and efficacy have a genetic component¹⁶ and these sub-studies are intended to investigate genetic causes of individual variations in clinical response to drugs.

Balancing interests

Researchers need to access valuable human tissue samples in a manner which takes account of the precious nature of the resource, the responsibility of clinicians and pathologists to prioritise the diagnostic process, the rights of the individual donors, and the rapid rate of scientific and clinical advances.¹⁷

13.14 The recommendations in this Report are intended to foster human genetic research, while providing reassurance to the community that such research is subject to proper ethical scrutiny and legal control. The recommendations balance the interests of researchers—who need access to human genetic samples and information from many sources—and the needs of individuals and their relatives—whose autonomy must be respected and whose privacy must be protected. The ability to conduct human genetic research may be prejudiced if potential volunteers fear that participation in research will generate information that they may subsequently be required to disclose to insurers, employers or others.

13.15 The interests of the genetic relatives of participants also need to be considered. Research results may be significant to the health of genetic relatives, including those who have not participated in the research and who may not have been aware that the research was being done. These family members may have an interest in the genetic samples or information of the research participant because testing those samples or acquiring that information may create new options for life decisions, including those with potential to improve health.

13.16 However, some family members may prefer not to be given information that may provide knowledge of future health or health risks. In addition, other family members who are not genetic relatives, such as partners and spouses, may have an interest because of concerns about the health of actual or potential offspring. The information generated by such research may also be of relevance to people in the community who are unrelated to participants or their families but whose family histories or health conditions have a similar genetic basis. Not all of this information will be of benefit to families. Indeed, in certain situations the implications of the information may be highly negative, such as incidental evidence of non-paternity.

15 One estimate is that approximately one in five clinical trials have an associated genetic sub-study and this proportion is increasing: HREC Chairs and Officers, *Consultation*, Sydney, 20 June 2002.

16 Advisory Committee on Health Research, *Genomics and World Health* (2002), World Health Organization, Geneva, 71.

17 Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002.

Present regulatory framework for research ethics

The NHMRC

13.17 The NHMRC is the statutory authority that governs the principles and procedures applicable to medical research and ethical matters relating to health. The *National Health and Medical Research Council Act 1992* (Cth) (NHMRC Act) establishes the NHMRC as a statutory corporation and prescribes its membership.¹⁸

13.18 The functions of the NHMRC are to inquire into, issue guidelines on, and advise the Commonwealth, the States and the community on matters relating to the improvement of health; prevention, diagnosis and treatment of disease; provision of health care; public health and medical research; and ethical issues relating to health.¹⁹

The National Statement

13.19 In relation to medical research, the NHMRC Act requires the NHMRC to issue guidelines for the conduct of medical research on humans, which are to be issued precisely as developed by the Australian Health Ethics Committee,²⁰ one of its principal committees. The present National Statement was issued by the NHMRC on 28 June 1999 in exercise of these statutory obligations.²¹ It is the successor document to the NHMRC *Statement on Human Experimentation*, first issued in 1966.

13.20 The National Statement is endorsed by the Australian Vice-Chancellors' Committee, the Australian Research Council, the Australian Academy of the Humanities, the Australian Academy of Science, and the Academy of the Social Sciences in Australia. Briefly, the National Statement.²²

- contains ethical principles relevant to all research involving humans;
- requires that particular matters are to be addressed when research involves children and young people, persons with an intellectual or mental impairment, persons highly dependent on medical care, those in dependent or unequal relationships, collectivities, and Aboriginal and Torres Strait Islander people;
- requires that specific matters be addressed in the consideration and approval of research involving radiation, assisted reproductive technology, clinical trials, epidemiology, human tissue samples, genetics, or deception in the conduct of research; and
- sets out the formation, membership and functions of HRECs.

18 *National Health and Medical Research Council Act 1992* (Cth) ss 6, 20–21.

19 *Ibid* s 7.

20 *Ibid* s 8.

21 The National Statement does not include updated references to the new private sector provisions of the *Privacy Act* (and the NPPs), particularly in relation to consent and collection issues.

22 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

Human Research Ethics Committees

13.21 The primary function of an HREC is to protect the welfare and rights of participants in research. An often overlooked secondary purpose of the National Statement and thus of HRECs is to ‘facilitate research that is or will be of benefit to the researcher’s community or to humankind’.²³

13.22 The National Statement provides that research proposals involving human participants must be reviewed and approved by an HREC. The National Statement sets out requirements that must be followed by institutions or organisations in establishing HRECs, by researchers in submitting research proposals to HRECs, and by HRECs in considering and reaching decisions regarding those proposals and in monitoring the conduct of approved research.²⁴ The National Statement places clear responsibilities upon institutions and researchers as well as providing the framework upon which the Australian system of research ethics review by HRECs is based.²⁵

Other regulatory mechanisms

13.23 In addition to the role of the NHMRC, HRECs and the National Statement, the ethical and legal framework for conducting research in Australia includes the following:²⁶

- relevant statutory restrictions on dealing with personal information (which may include genetic information), including those under the *Privacy Act 1988* (Cth) and related guidelines;²⁷
- the common law duty to exercise reasonable care, which is owed by researchers, research organisations and HRECs to participants in research;²⁸
- standards for the scientific validity of research, notably the *Statement and Guidelines on Research Practice* issued by the NHMRC and the Australian Vice-Chancellor’s Committee,²⁹ and

23 Ibid Preamble.

24 Ibid [2].

25 Selected issues concerning the membership and functions of HRECs are discussed in more detail in Ch 17.

26 This regulatory framework was described in more detail in Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, Ch 6. The National Statement is supplemented by commentary in the National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), NHMRC, Canberra.

27 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council.

28 In accordance with the common law relating to the tort of negligence. The Law Institute of Victoria noted that the ‘proximity’ of the relationship is no longer seen as the unifying criterion of duties of care: see the decision of the High Court in *Perre v Apand Pty Ltd* (1999) 198 CLR 180, 209. Researchers, research organisations and HRECs may be liable to third parties, including families, relatives and other parties who suffer reasonably foreseeable loss or damage: Law Institute of Victoria, *Submission G275*, 19 December 2002.

29 National Health and Medical Research Council and Australian Vice Chancellors’ Committee, *Statement and Guidelines on Research Practice* (1997), NHMRC, Canberra.

- NHMRC guidelines applying to specific types of research, such as the NHMRC *Guidelines for Ethical Review of Research Proposals for Human Somatic Cell Gene Therapy and Related Therapies*.³⁰

Reform of the regulatory framework for human genetic research

13.24 The Inquiry received a wide range of views and comments about the adequacy of the regulatory framework for human genetic research. Views ranged from those that were highly supportive of the existing framework through to those suggesting a broad agenda for change. In many cases the changes proposed were broadly applicable to all human research and not limited to addressing issues raised by human genetic research specifically.³¹

13.25 A common theme in consultations with individuals and organisations involved in human genetic research was concern about the present and possible future costs of compliance with regulatory requirements. It was noted that research funding rarely includes any allocation for compliance costs—such as the cost of operating HRECs, obtaining ethics approval for research proposals, complying with ethical requirements for the de-identification of samples, and satisfying monitoring and reporting obligations.³²

13.26 The same groups, and HREC members, frequently observed that the National Statement has only been in operation since 1999 and that there may not have been enough time to evaluate whether its guidance is effective in protecting privacy and ensuring ethical conduct in human genetic research. Particular concerns were expressed about the need to avoid any retrospective reforms that might have adverse practical implications for existing long-term research projects.³³

13.27 The NHMRC's Research Committee emphasised that the Inquiry, when considering the introduction of new regulatory mechanisms, should assess

whether there is evidence that abuses have occurred in association with Australian data collections in the area under investigation. This is particularly necessary where the regulations envisaged are likely to act as a barrier to research. It should be born in mind that in genetic research, in particular, primary beneficiaries of the results of the research are likely to be the subjects (including their families) from whom data is collected. Barriers which delay study commencement or progress in this highly competitive setting—often involving at least some commercial funding—may prevent

30 National Health and Medical Research Council, *Guidelines for Ethical Review of Research Proposals for Human Somatic Cell Gene Therapy and Related Therapies* (1999), National Health and Medical Research Council, Canberra.

31 The Department of Human Services South Australia observed that many of the concerns about regulating genetic research are common to many other areas of medical research and suggested that broader review may be required: Department of Human Services South Australia, *Submission G288*, 23 December 2002.

32 Queensland Institute of Medical Research, *Consultation*, Brisbane, 13 December 2001.

33 *Ibid.*

the studies being conducted on Australian population samples, with a resulting loss to the individuals as well as to Australian research.³⁴

13.28 Several other submissions identified the commercialisation of human genetic research as an important background development.³⁵ For example, the Centre for Law and Genetics noted that

it is a feature of human genetic research that commercial arrangements include the hand over of tissue or blood samples. In this respect the samples and information they contain have become commercial commodities.³⁶

13.29 Many submissions suggested significant changes to the regulatory framework for protecting privacy and ensuring ethical conduct in human genetic research. For example, Privacy NSW suggested a range of reforms to improve the 'transparency and accountability of ethics review in respect of genetic research involving humans'. These included the establishment of an independent, national ethics review committee; enhancing the obligations of HRECs to report their decisions; a single, independent and transparent complaints mechanism to deal with decisions made by any HREC; strong civil and criminal sanctions where privacy is breached; and effective audit of an HREC's activities, including monitoring throughout the research period.³⁷

13.30 Professor Nick Saunders and Associate Professor Paul Komesaroff highlighted several issues of particular interest to the research and ethics communities. These included concerns relating to the question of waiver of consent, the monitoring of research by HRECs, de-identification of genetic samples, withdrawal of participants from research projects, provision of advice to participants and the storage of data.³⁸

13.31 Specific reforms suggested in submissions and consultations are discussed in the following chapters. The National Statement is scheduled for formal review by the NHMRC during the 2003–2005 triennium, consistently with NHMRC policy of revising guidelines that have been in place for five years. A number of recommendations made in this Report are directed to the NHMRC, for implementation as part of its review of the National Statement.

34 National Health and Medical Research Council Research Committee, *Submission G262*, 20 December 2002.

35 M Dunne, *Submission G041*, 17 December 2001; Australian Academy of Science, *Submission G097*, 21 January 2002; A Rose, *Submission G116*, 18 March 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002; N Saunders and P Komesaroff, *Submission G084*, 9 January 2002; J Fleming, *Submission G241*, 20 December 2002; Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003.

36 The Centre for Law and Genetics noted that there is currently 'no systematic data collection or analysis of the changing features of research in an increasingly commercialised environment': Centre for Law and Genetics, *Submission G048*, 14 January 2002.

37 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

38 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

Enforcing compliance with the National Statement (Chapter 14)

13.32 The enduring value of the National Statement was highlighted in a number of submissions—even those that suggested changes to its scope or content. For example, the Office of the Federal Privacy Commissioner observed that, like the Declaration of Helsinki and other international ethical guidelines, the National Statement has persuasive authority, has been reviewed at appropriate intervals, and has proven responsive to past challenges in medical research.³⁹ Saunders and Komesaroff observed that the National Statement ‘commands profound moral authority in the community’.⁴⁰

13.33 However, the application of the requirements for institutions and their HRECs to be registered with the NHMRC and to follow the processes set down in the National Statement is incomplete. As emphasised in many submissions, there is currently no obligation on private research bodies to adhere to the provisions of the National Statement.⁴¹ Submissions and consultations suggested that, in view of the increasing commercialisation of human genetic research, the mechanisms by which compliance with the National Statement is enforced should be strengthened.

13.34 Chapter 14 examines this issue and recommends that the NHMRC review the mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.

Human genetic research and consent (Chapter 15)

13.35 The concept of consent is fundamental to the legal and ethical regulation of human research. The National Statement generally requires consent to the use of human tissue samples, genetic material and genetic information in medical research, other than in limited and defined circumstances.

13.36 Submissions and consultations raised concerns about various aspects of consent and, in particular, whether adequate privacy protection is afforded by the current provisions under which HRECs may waive consent requirements in granting ethical approval for research proposals. A related issue concerns the extent to which researchers are able to obtain consent from potential research participants for the use of their genetic samples or information for unspecified future research.

13.37 Chapter 15 examines issues of consent in human genetic research. The Inquiry recommends new reporting obligations for HRECs with respect to proposals for which waiver of consent has been granted under the National Statement. The Inquiry also recommends that training and other support for HRECs be augmented in relation to decision-making on waiver of consent. In the Inquiry’s view, the National

39 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

40 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

41 Although, in research using experimental therapeutic goods, conformity to the National Statement is required by legislation: *Therapeutic Goods Act 1989* (Cth) s 19; *Therapeutic Goods Regulations 1990* (Cth) r 12AD.

Statement should also be amended to provide clear guidance about obtaining consent to unspecified future human genetic research.

Encouraging best practice in human genetic research (Chapter 16)

13.38 Submissions and consultations suggested that it may be useful for the NHMRC to develop information and advice, including examples and practical guidance, for the preparation of human genetic research protocols and consent forms for human genetic research.

13.39 The intention of these documents would be to give further guidance to researchers and HRECs on what the NHMRC considers to be best practice in the conduct of human genetic research. The content of these documents is discussed in Chapter 16. Particular concerns relate to the coding and de-identification of genetic samples and information, and to the disclosure to research participants of the potential commercialisation of research outcomes.

Strengthening ethical review by HRECs (Chapter 17)

13.40 Submissions and consultations suggested a range of possible reforms aimed at strengthening the current system of ethical review of human genetic research and the role of HRECs within that system.

13.41 A range of options is discussed in Chapter 17, including possible reforms relating to:

- the structure of ethics review;
- the quality of ethics review, including the membership of HRECs, their expertise and sources of advice;
- monitoring the conduct of research approved by HRECs;
- the needs of HRECs, HREC members and researchers, in terms of resources, education and training;
- the accountability of HRECs, including reporting on review of research proposals by HRECs; and
- the accreditation of HRECs.

13.42 In Chapter 17 the Inquiry recommends that the NHMRC develop and implement procedures to promote consistency, efficiency, transparency and accountability in HREC review of human genetic research, through a systematic quality improvement program.

14. Enforcing Compliance with the National Statement

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Introduction

14.1 As discussed in Chapter 13, the present regulatory framework for the ethical conduct of human research is centred on the National Health and Medical Research Council's (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement)¹ and on review of research proposals by Human Research Ethics Committees (HRECs).

14.2 This chapter examines the regulatory status of the National Statement and the consequences of non-compliance for researchers or research organisations. The chapter then discusses whether this status is adequate to protect participants involved in genetic research wherever conducted in Australia. The chapter also considers whether the current regulatory framework is adequate to protect genetic samples and information or whether different accountability and enforcement mechanisms are required.

¹ National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

Consequences of non-compliance with the National Statement

14.3 The National Statement provides that the NHMRC, through the Australian Health Ethics Committee (AHEC), will audit the activities of HRECs to ensure compliance with the National Statement.² The National Statement requires institutions and their HRECs to present an annual compliance report on their ethics approval procedures.³ The requirements for HRECs to register with the NHMRC, report annually and follow the processes in the National Statement (and other guidelines) are not mandated by legislation.

14.4 AHEC presents an annual report of HREC activities to the Council of the NHMRC. The Council in turn reports to the Commonwealth Parliament. The AHEC report includes a statement on the compliance reports received from HRECs in that year. AHEC may name any non-compliant HREC and its institution, although this power has not yet been required to be exercised.

14.5 Despite the lack of statutory powers of enforcement, there has been a high level of voluntary compliance by HRECs with the requirements of the National Statement. The Centre for Law and Genetics observed that

the general acceptance by all research institutions of the National Statement has, in effect, if not in law, created a uniform research regime.⁴

14.6 The power to withdraw funding is the most important and direct mechanism by which the NHMRC may induce compliance with the National Statement. An assessment of the scope and effectiveness of potential withdrawal of NHMRC funding as an incentive to comply requires an understanding of how medical research in general, and human genetic research in particular, is funded in Australia. The funding of medical research and the withdrawal of funding are discussed below.

14.7 Non-compliance with the National Statement may have other consequences for researchers and research organisations. For example, the Human Genetics Society of Australasia (HGSA) noted that other sanctions include:

disciplinary action by the employer, rejection of articles by journals in the absence of ethics committee approval, peer group censure and public disclosure of unethical conduct.⁵

14.8 Some additional factors that encourage compliance with the National Statement are described in more detail below.

2 Ibid [2.46].

3 These reporting requirements are discussed in more detail in Ch 17.

4 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

5 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

Withdrawal of funding

14.9 The NHMRC is one of the major providers of medical research funding to institutions such as hospitals, universities and research institutes. The NHMRC also makes recommendations to the Commonwealth on expenditure for research and training in medicine and public health, including recommendations on the application of the Medical Research Endowment Fund.⁶ The National Statement has been endorsed by the Australian Research Council and other bodies that have a role in research, such as the Australian Vice-Chancellors' Committee and the learned Academies.

14.10 It is a condition of an institution's continuing eligibility to receive NHMRC funding that all research involving humans conducted in the institution must be approved by an HREC that has been established under, and functions according to, guidelines issued by the NHMRC. Compliance with the National Statement is also a condition of NHMRC grants of research funds. In the event that reporting by research organisations and their HRECs reveals that they have not complied with the National Statement, implementation of NHMRC policy may result in that institution ceasing to be eligible to receive funding for research. The effectiveness of this mechanism has not needed to be tested in Australia, but American experience suggests that withdrawal, or threat of withdrawal, of funding by the peak research funding body has salutary effects upon institutions.⁷

NHMRC funding

14.11 In assessing the scope and effectiveness of potential withdrawal of NHMRC funding as an incentive to comply with the National Statement, it is important to note that the NHMRC is the largest single provider of peer-reviewed health research funds. In 2002, the NHMRC's research expenditure was \$277.1 million.⁸ In 2001, funding through NHMRC accounted for 34% of public sector funding, 27% of total government funding and 18% of total expenditure on health and medical research in Australia.⁹

14.12 While it may appear, from the 18% figure, that much health and medical research lies outside NHMRC funding, in practice, the influence of NHRMC funding decisions extends beyond projects that receive direct funding. Researchers and research organisations commonly derive funds from a variety of sources. It is common for research to be supported by private sector funding but conducted at publicly funded

6 The Medical Research Endowment Fund is established by the *National Health and Medical Research Council Act 1992* (Cth) s 49.

7 See R Steinbrook, 'Protecting Research Subjects — The Crisis at Johns Hopkins' (2002) 346 *New England Journal of Medicine* 716. The frequency with which these powers are used is an important element in their effectiveness. See the published letters sent by the US Office for Human Research Protections to research organisations: Office for Human Research Protections, *Compliance Oversight*, US Department of Health and Human Services, <ohrp.osophs.dhhs.gov/compovr.htm>, 14 March 2003.

8 National Health and Medical Research Council, *Investing in Australia's Health* (2002) National Health and Medical Research Council, 45.

9 National Health and Medical Research Council, *Correspondence*, 13 February 2003.

institutions that require compliance with the National Statement and other NHMRC guidelines.

14.13 Further, any withdrawal of funding by the NHMRC, once made public, would be likely to be followed by action by other funding bodies,¹⁰ such as those responsible for the allocation of non-NHMRC Commonwealth government research funds or state government funds for hospital research activities.

Private sector funding

14.14 The 1999 Wills Report, which resulted from a major strategic review of health and medical research in Australia, provided the following breakdown of total expenditure on health and medical research.¹¹ No similar breakdown is available for human genetic research specifically.

Table 14-1 Expenditure on Health and Medical Research by Sector

Commonwealth government and higher education	47.5%
State government	24.5%
Private non-profit	15.9%
Business enterprise	12.1%
TOTAL	100%

14.15 If these figures are assumed to apply to human genetic research as they do to health and medical research generally, between 25% and 30% of research funding may be provided by the private sector and therefore lie beyond the direct reach of the NHMRC.

14.16 However, the distinction between public and private research should not be overstated. Public and private research funding may converge in the same research projects. For example, one feature of medical and other research in Australia is the Cooperative Research Centre Program. Cooperative Research Centres (CRCs) are collaborative research ventures involving researchers from Commonwealth and state government funded organisations (such as universities and university-based research institutes), private non-profit organisations and business enterprise.¹²

10 National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002.

11 Health and Medical Research Strategic Review, *The Virtuous Cycle, Working Together for Health and Medical Research* (1999), Commonwealth of Australia, Canberra, 166, Exhibit 5.0-5. These figures were based on ABS 1996 data and NHMRC 1994 data.

12 The total 1999-2000 public and private sector funding for the Gene CRC was \$40.6 million of which Commonwealth CRC program funding comprised \$13.1 million. The core participants, including the private sector partner (Cerylid), provided the balance of the resources: CRC for Discovery of Genes for Common Human Diseases, *CRC for Discovery of Genes for Common Human Diseases*, Gene CRC, <www.crc.gov.au/centres/medical/genes.htm>, 3 May 2002.

Other consequences of non-compliance

14.17 The withdrawal of NHMRC funding remains a powerful incentive for public sector researchers and research institutions to comply with the National Statement. In addition, compliance is required or encouraged by other factors, including privacy laws, therapeutic goods legislation, concern about professional standing and publication, and the threat of potential litigation.

Privacy law

14.18 As discussed in Chapter 15, the *Privacy Act 1988* (Cth) (*Privacy Act*) and other privacy legislation imposes legal requirements to obtain consent to the collection, use or disclosure of personal information, including genetic information. These requirements may be waived where collection of the information is for research purposes and is carried out in accordance with guidelines issued by the NHMRC and approved by the federal Privacy Commissioner under s 95 or s 95A of the *Privacy Act* (the s 95 and s 95A Guidelines).

14.19 The s 95 and s 95A Guidelines require consideration, by an HREC, of whether the public interest in the research outweighs to a substantial degree the public interest in compliance with privacy principles.¹³ In some circumstances, a determination by an HREC to waive consent, made other than in compliance with the National Statement, may constitute a breach of the *Privacy Act*.

14.20 This may lead to researchers or research organisations being named in the NHMRC's annual report or in a report to a Commonwealth agency or the federal Privacy Commissioner.¹⁴ Naming can be expected to affect adversely the reputation of the named researchers or research organisations, their employment prospects and capacity to attract future research funding.

14.21 Further, where the conduct of a private sector researcher or research organisation is in breach of the *Privacy Act*, affected individuals may complain to, and have their complaints investigated by, the federal Privacy Commissioner or by an adjudicator under an approved privacy code.¹⁵ The Privacy Commissioner may make determinations declaring that conduct constituting an interference with the privacy of an individual should cease and that compensation for loss or damage be paid.¹⁶ Such determinations, and similar determinations of adjudicators, are enforceable in proceedings before the Federal Court or Federal Magistrates Court.¹⁷

13 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra 3.2(b); National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council, D.4.

14 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [4.3].

15 See *Privacy Act 1988* (Cth) Pt V.

16 *Ibid* s 52.

17 *Ibid* s 55A.

Therapeutic Goods Regulations

14.22 The *Therapeutic Goods Regulations 1990* (Cth) provide that unregistered therapeutic goods may be used in clinical trials only on certain conditions.¹⁸ These conditions include a requirement that the use is in accordance with the National Statement as published by the NHMRC from time to time.¹⁹ Non-compliance with the National Statement in effect renders unlawful the continued use of unregistered therapeutic goods.²⁰

Professional standing and publication

14.23 Non-compliance with accepted ethical standards, including the National Statement, may have consequences for how individual researchers are viewed by their peers.

Professional peer pressures and the need for acceptance of research outcomes in the international scientific community are powerful drivers in this area.²¹

14.24 Compliance with the National Statement and other ethical standards may be a pre-requisite for publication of the results of research and the professional benefits that derive from publication.²² The World Medical Association's *Ethical Principles for Medical Research Involving Human Subjects* (the Helsinki Declaration) states that reports of experimentation not in accordance with the ethical principles laid down in the Declaration should not be accepted for publication.²³ The International Committee of Medical Journal Editors requires authors, when reporting experiments on human subjects, to 'indicate whether the procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional or regional) and with the Helsinki Declaration'.²⁴

The National Statement and litigation

14.25 The principles of ethical conduct and the process for ethical scrutiny of research proposals set out in the National Statement constitute accepted standards of research practice. The conduct of a researcher or research organisation may be compared with these standards in any litigation arising from a research project in which, for example, a question arises as to whether reasonable care was taken. In this

18 *Therapeutic Goods Act 1989* (Cth) s 19; *Therapeutic Goods Regulations 1990* (Cth) r 12AD.

19 *Therapeutic Goods Regulations 1990* (Cth) r 12AD(c).

20 *Therapeutic Goods Act 1989* (Cth) ss 19–22.

21 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

22 Although it is debateable whether, in the case of path-breaking new research results, publishers will be so scrupulous in this regard.

23 World Medical Association, *Ethical Principles for Medical Research Involving Human Subjects* (2000), Helsinki, Principle 27.

24 International Committee of Medical Journal Editors, *Uniform Requirements for Manuscripts Submitted to Biomedical Journals*, <www.icmje.org/index.html>, 3 May 2002.

context, an analogy may be drawn with clinical practice guidelines,²⁵ including those issued by the NHMRC itself.

14.26 Courts may take standards for the conduct of human research set out in the National Statement as evidence of accepted professional standards of practice. It has been said that the evidentiary value of clinical practice guidelines depends on their purpose, development, ratification, dissemination, use, and whether they are current.²⁶ On this basis, a court might find the National Statement influential or even decisive evidence.

Regulation of private sector human genetic research

[G]enetic research depends on the medical-industrial complex. The gene has become a kind of commodity as vital to the economies of developed countries as oil or uranium. ... Developed countries are fiercely competing with each other to take the lead; universities are running the risk of becoming the handmaidens of private industry; and corporations are locked in battle to be the first to perfect the molecule that will enable them to forge ahead of the competition and line the pockets of their shareholders.²⁷

14.27 Consultations and submissions highlighted the continuing convergence of public and private research. Many private sector organisations receive public funding and there is increasing collaboration through public and private partnerships—collaboration recommended and encouraged by the Wills Report.²⁸

14.28 Investment in research by private commercial interests is likely to have at least two relevant consequences. First, institutions conducting privately funded research may not be dependent on remaining eligible for public research funding. Second, the commercial interests of researchers and research organisations may lead to conflicts of interest that can compromise the validity and safety of research.

14.29 In 2001, the House of Representatives Standing Committee on Legal and Constitutional Affairs reported on its inquiry into the scientific, ethical and regulatory aspects of human cloning and stem cell research. The Standing Committee observed that:

25 See J Pelly and others, 'Clinical Practice Guidelines Before the Law: Sword or Shield?' (1998) 169 *Medical Journal of Australia* 330, 330–333; B Bennett, 'Setting the Standard? Clinical Practice Guidelines and Medical Negligence Litigation' (1997) 20 *University of New South Wales Law Journal* 707.

26 F Tito and L Newby, *Medico-Legal Implications of Clinical Practice Guidelines* (1998) National Breast Cancer Centre.

27 H Doucet, 'The Role of Ethics in the Development of Genetics' (2001) 2(3) *Isuma: Canadian Journal of Policy Research* 40, 43.

28 Health and Medical Research Strategic Review, *The Virtuous Cycle, Working Together for Health and Medical Research* (1999), Commonwealth of Australia, Canberra, Ch 8, App 1 (Government Response to the Recommendations and Actions in the Report of the National Health and Medical Research Strategic Review—NHMRC to Action).

The growth and spread of cloning research and the substantial involvement of the private sector in it renders it very difficult for a body such as the NHMRC or AHEC to monitor this area of risk. The leverage of the NHMRC is very much tied to its capacity to grant or withhold funding and hence its real capacity to influence the private sector must be problematic as AHEC itself acknowledged. In such an environment sanctions such as the loss of research funding may have minimal influence.²⁹

14.30 The Committee concluded that the current regulatory environment for medical research was ‘deeply unsatisfactory’ and was the product of an era when the majority of research funding was provided by government and most research occurred within tertiary institutions that were publicly funded.³⁰ The Committee stated that consistent regulation should be applied to both publicly and privately funded research.

If the current framework continues ... it is likely to lead to the evolution of a system increasingly similar to that in the United States ... There the public sector is regulated and the private sector, where much of the research is undertaken, is subject to limited regulation.³¹

14.31 Human genetic research involves significant private sector funding, public/private partnerships and commercial pressures on public sector research, such as that conducted in universities. The Standing Committee’s observations may be as relevant to the regulation of human genetic research as to human cloning and stem cell research.

Regulation of medical research overseas

14.32 Despite significant differences, the approach taken to the regulation of the conduct of medical research in Australia is broadly similar to that in Canada, the United Kingdom and the United States. In each jurisdiction, national ethical standards have been established by government regulation or by government bodies. These national norms are applied to research proposals by multi-disciplinary ethics committees. In each jurisdiction, public funding serves as the primary means by which compliance with ethical standards is enforced, whether directly or indirectly.

14.33 The centrepiece of regulation in Canada is the *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans* (Tri-Council Policy Statement).³² Under the Tri-Council Policy Statement all research that involves living human subjects requires review and approval by a Research Ethics Board.³³ The Canadian research councils require, as a minimum condition of funding, that researchers and

29 House of Representatives Standing Committee on Legal and Constitutional Affairs, *Human Cloning: Scientific, Ethical and Regulatory Aspects of Human Cloning and Stem Cell Research* (2001), Parliament of the Commonwealth of Australia, Canberra, 162–164.

30 Ibid, 167.

31 Ibid.

32 Natural Sciences and Engineering Research Council of Canada and Social Sciences and Humanities Research Council of Canada, *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans*, Canadian Institutes of Health Research, 8 May 2002.

33 Ibid, art 1.1.

their institutions apply the ethical principles and articles of the Tri-Council Policy Statement.³⁴

14.34 In the United Kingdom, the *Research Governance Framework for Health and Social Care*³⁵ applies to all research relating to the responsibilities of the Secretary of State for Health.³⁶ Research within the National Health Service (NHS) must have the prior approval of an NHS Research Ethics Committee (REC).³⁷ Ethics review by RECs is also required for research funded by the Medical Research Council³⁸ and General Medical Council.³⁹

14.35 In the United States, the centrepiece of regulation of the conduct of medical research is known as the 'Common Rule'.⁴⁰ The Common Rule applies only to research that is supported in some way by the federal government. Importantly, this includes research funded by the National Institutes of Health, the focal point for federally funded medical research.

The need for reform

14.36 The Inquiry recognises that the National Statement continues to have an important and valued influence on the way in which research involving humans is conducted in Australia. The National Statement has played an important role in regulating research, even in the absence of an explicit legal basis.⁴¹ The National Statement has been said to command 'profound moral authority in the community, independently of the theoretical sanction of the loss of NHMRC funding support'.⁴² Indeed, many HRECs have been created in institutions that do not receive NHMRC funding.

14.37 Some submissions suggested that legislation to extend the scope or enforceability of the National Statement was not necessary.⁴³ However, most submissions to the Inquiry supported strengthening the mechanisms by which

34 Ibid, art 1.1(a).

35 Department of Health (UK), *Research Governance Framework for Health and Social Care*, <www.nserc.ca/programs/ethics/english/policy.htm>, 7 May 2002.

36 Ibid, 3.

37 Ibid, 33.

38 Medical Research Council, *Medical Research Council Guidelines for Good Clinical Practice in Clinical Trials*, <www.mrc.ac.uk/pdf-ctg.pdf>, 8 May 2002.

39 General Medical Council, *Research: The Role and Responsibilities of Doctors*, <www.gmc-uk.org/standards/standards_frameset.htm>, 8 May 2002.

40 *Federal Policy for the Protection of Human Subjects (Basic DHHS Policy for Protection of Human Research Subjects)* 45 CFR Pt 46 Subpt A.

41 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

42 Ibid.

43 For example, Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; D Cavaye, *Submission G110*, 14 March 2002.

compliance with the National Statement is enforced—to require all researchers and research organisations to comply.⁴⁴

14.38 In particular, submissions questioned the effectiveness of the existing legal status of ethics review requirements, in view of increasing private sector involvement in the funding of research. For example, Jennifer Fleming stated:

The increasing commercialisation of human genetic research and clinical research overall requires a strengthening of the mechanisms to ensure compliance irrespective of whether the research is conducted by public or private research bodies or in collaboration with overseas sponsors.⁴⁵

14.39 Similarly, Privacy NSW submitted:

The commercial context in which the vast majority of research occurs undermines the traditional justification for the largely self-regulatory framework underpinning research involving humans. With reductions in public funding, even universities and public hospitals now compete for private industry involvement and funding of their activities.⁴⁶

14.40 The New South Wales Genetics Service Advisory Committee also referred to the need to monitor the ethical performance of research that is not dependent on public funding or carried out through the public health system. The Committee stated that regulators should have power to review and oversee research undertaken by private organisations, and it supported sanctions where there has been unethical conduct.⁴⁷

14.41 The Inquiry recognises the importance of private sector investment in Australian human genetic research. As the Australian Academy of Science has observed:

44 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Australian Academy of Science, *Submission G097*, 21 January 2002; Queensland University of Technology, *Submission G109*, 14 March 2002; Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Caroline Chisholm Centre for Health Ethics, *Submission G061*, 26 December 2001; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Health Consumers' Council, *Submission G174*, 18 September 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Australian Medical Association, *Submission G212*, 29 November 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; J Fleming, *Submission G241*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Australian Institute of Aboriginal and Torres Strait Islander Studies, *Submission G286*, 16 December 2002; Department of Human Services Victoria — Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

45 J Fleming, *Submission G241*, 20 December 2002.

46 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

47 New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002.

No significant political or academic group argues against the underlying principle of commercial involvement in research, because ending this involvement would require a corresponding injection of several hundred million dollars per annum into biomedical research by the Australian government.⁴⁸

14.42 At present, most privately funded human genetic research in Australia is associated in some way with a university, public hospital or public agency (such as the CSIRO). Similarly, many individual researchers have ongoing employment or professional relationships with public bodies, or membership of professional associations that expect (and in some cases require) compliance with the National Statement and other NHMRC guidelines.

14.43 The Inquiry has not been made aware of any privately funded genetic research in Australia that does not comply with the requirements of the National Statement. Indeed, some private sector organisations involved in research have voluntarily adopted the National Statement as an organisational policy⁴⁹ and some, including private hospitals, submit research proposals for ethics review by HRECs registered with AHEC.

14.44 Submissions acknowledged that there is no evidence that private sector research is being conducted other than in accordance with accepted ethical standards. Nevertheless, mechanisms to ensure private sector compliance were supported. For example, the Australian Medical Association stated that:

Although private sector research should not be stereotyped as being conducted in an 'ethical vacuum', there is certainly more scope for unethical conduct in that sector, and this certainly means that greater scrutiny of the private sector is required.⁵⁰

14.45 The potential for withdrawal of funding, the inability to publish research, and peer pressure from within the scientific and medical community, are powerful incentives for researchers and research organisations to comply with the National Statement. However, these incentives may be attenuated in relation to private sector research.

14.46 The Inquiry has concluded that there should be more formal requirements for private sector genetic research involving humans to comply with the National Statement and, in particular, with requirements for ethical review by an HREC constituted and operating in accordance with the National Statement.

48 Australian Academy of Science, *Submission G097*, 21 January 2002.

49 For example, Autogen Limited, an Australian biotechnology research and development company has established its own ethical standards for research. These standards require that ethics approval for Autogen research projects will be obtained through 'a recognised Human Ethics Committee in Australia and a Human Ethics Committee in any country from which samples are to be collected' and will comply with ethical principles identified in the National Statement: Autogen Limited, *Ethics Policy*, <www.autogenlimited.com.au/ethics.html>, 13 May 2002.

50 Australian Medical Association, *Submission G212*, 29 November 2002.

Options for reform

14.47 The options for reform to enforce compliance with the National Statement include:

- the enactment of legislation to prohibit the conduct of any human genetic research, other than in compliance with the National Statement;
- the establishment of a national licensing or registration scheme covering human genetic research in Australia; and
- new self-regulatory arrangements applying to research organisations conducting human genetic research.

14.48 These regulatory options, and the views expressed in relation to them in submissions and consultations, are discussed further below.

Legislating for compliance with the National Statement

14.49 In DP 66, the Inquiry proposed that the *National Health and Medical Research Council Act 1992* (Cth) (NHMRC Act) be amended to prohibit the conduct of any human genetic research, other than in compliance with the National Statement.⁵¹

14.50 There is a precedent for enforcing compliance with the National Statement through legislation. As noted above, the *Therapeutic Goods Regulations 1990* (Cth) provide that unregistered therapeutic goods may be used in clinical trials only on certain conditions,⁵² including a requirement that the use is in accordance with the National Statement.⁵³ DP 66 asked whether such a regulatory response would be appropriate for regulating the conduct of human genetic research.

14.51 The National Statement comprises broad ethical principles and short statements of considerations relevant in specific research contexts. It offers guidance on, rather than prescription of, ethically sound research design and practice.⁵⁴ Whether a research organisation has conducted research in compliance with the National Statement as a whole will generally not be conducive to definitive answer.⁵⁵

14.52 Another difficult issue is the type of penalty that might follow failure to comply with the National Statement. Legislation could be drafted to make both individual researchers and the executive officers of a non-complying institution liable to a criminal penalty, along with the institution itself. The HGSA noted that

51 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 11–1.

52 *Therapeutic Goods Act 1989* (Cth) s 19; *Therapeutic Goods Regulations 1990* (Cth) r 12AD.

53 *Therapeutic Goods Regulations 1990* (Cth) r 12AD(c).

54 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 1.

55 However, it would be relatively easy to establish whether an organisation has established and operates an HREC of the prescribed composition.

a system of fines could be introduced, directed at employing institutions. This would stimulate organisations to invest appropriate resources in ethical oversight of human research.⁵⁶

14.53 Submissions generally supported the policy underlying the proposal to amend the NHMRC Act, namely, that the conduct of all human genetic research in Australia should comply with the National Statement, whether in the public or private sector. With few exceptions, submissions also supported the idea that, at least in relation to human genetic research, legislation should explicitly require compliance with the National Statement.⁵⁷ For example, the Australian Academy of Science submitted that all research with human subjects, not only that conducted with NHMRC support or in public hospitals and universities, should be legally required to go before an HREC for approval.⁵⁸ The Queensland University of Technology submitted that the National Statement be given further legislative force.⁵⁹

14.54 The Caroline Chisholm Centre for Health Ethics stated that:

The relevant NHMRC guidelines are well constructed and offer quite good advice on the ethically appropriate ways to handle genetic information. However, they lack the force or authority that regulation of this area is likely to require.⁶⁰

14.55 The Australian Red Cross Ethics Committee submitted that:

the ethical oversight of all human genetic research should be strengthened, by prohibiting any genetic research that does not comply with NHMRC standards.⁶¹

14.56 The Department of Human Services South Australia stated that:

Where any research body fails to comply with national standards regulating genetic research they are damaging not only their own reputation and that of the scientific community in general, but eroding public confidence in a system designed to represent the public interest when such decisions are made. Failure to comply with the national standard should result in any organisation that conducts genetic research being prohibited from conducting any further research.⁶²

56 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

57 Health Consumers' Council, *Submission G174*, 18 September 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Australian Medical Association, *Submission G212*, 29 November 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; J Fleming, *Submission G241*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Australian Institute of Aboriginal and Torres Strait Islander Studies, *Submission G286*, 16 December 2002; Department of Human Services Victoria - Metropolitan Health & Aged Care Services Division, *Submission G289*, 24 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

58 Australian Academy of Science, *Submission G097*, 21 January 2002.

59 Queensland University of Technology, *Submission G109*, 14 March 2002.

60 Caroline Chisholm Centre for Health Ethics, *Submission G061*, 26 December 2001.

61 Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003.

62 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

14.57 However, some submissions questioned whether amendments to the NHMRC Act are the appropriate mechanism by which to require compliance with standards for the ethical conduct of research. Others highlighted more general problems involved with legislating for compliance.

14.58 The Queensland Government expressed reservations about the proposal to amend the NHMRC Act and cautioned that

further careful consideration should be given to the proposal. In addition, it should be recognised that the guidelines in their current form are not appropriate to be enforceable. ... Careful consideration of the impact of enforceable re-drafted standards would be necessary to ensure that there were no unintended consequences which may limit the progress of research conducted in an acceptable manner.⁶³

14.59 Concerns were also expressed about the implications for the NHMRC, should the NHMRC Act be amended to require the NHMRC to enforce compliance.

If the National Statement became an enforceable document, and the NHMRC a regulatory body, this would represent a significant shift in its role. ... It is possible that the existing valued functions of the NHMRC might be adversely affected should it become a regulatory body. Extensive consultation with stakeholders would be required to inform detailed consideration of this proposal.⁶⁴

14.60 More generally, submissions recognised practical difficulties in framing legislation to require compliance. The Commonwealth Department of Health and Ageing stated:

While the Department acknowledges the issues regarding the current system, the establishment of a framework for sanctions would also face complications. Full regulation of the research industry would also require a vast amount of resources.⁶⁵

14.61 The New South Wales Health Department gave in principle support for the proposal to amend the NHMRC Act, but noted that this option 'may be difficult in practice'.

It is supported for genetic research because of its potential to intrude on privacy and affect the well-being not only of the individual, but also family, and the fact that there is such a strong private sector involvement. A lot of research goes on in the public sector that is already very well regulated by current legislation. An incremental approach as the need develops may be preferable.⁶⁶

Licensing of human genetic research

14.62 Another option raised in DP 66 was the establishment of a licensing or registration scheme covering human genetic research in Australia. However, there was no significant support in submissions or consultations for the introduction of licensing

63 Queensland Government, *Submission G274*, 18 December 2002.

64 Queensland Government, *Submission G274*, 18 December 2002. It should be noted that the *Research Involving Embryos Act 2002* (Cth) establishes a licensing committee under the NHMRC, with a regulatory role.

65 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

66 NSW Health Department, *Submission G303*, 13 January 2003.

or registration—provided that regulation of human genetic research could be strengthened through reforms to the ethics review system and through other means to enhance compliance with the National Statement.

14.63 A licensing regime for the conduct of human genetic research could require an undertaking to comply with the National Statement as a condition of a licence to conduct research. A substantial breach of the National Statement could result in conditions being placed on the licence, or suspension or cancellation of the licence.

14.64 Relevant examples of licensing regimes are those established under the *Gene Technology Act 2000* (Cth) (Gene Technology Act) in relation to certain dealings with genetically modified organisms, and for research involving embryonic stem cells under the *Research Involving Human Embryos Act 2002* (Cth).

Registration of human genetic research

14.65 While licensing generally involves close and ongoing supervision of the regulated activity, a more ‘light touch’ regulatory approach is the establishment of a registration scheme. Under such a scheme there would be a legislative requirement to register research with a regulatory agency, accompanied by penalties for the conduct of unregistered human genetic research. The provisions of the Gene Technology Act and the *Gene Technology Regulations 2001* (Cth) concerning ‘notifiable low risk dealings’ are examples of such an approach.⁶⁷

14.66 Alternatively, a registration scheme for human genetic research could focus on HRECs, rather than on the conduct of research itself. The National Statement currently requires the NHMRC, through AHEC, to audit the activities of HRECs to ensure compliance with the National Statement.⁶⁸ While not required by the NHMRC Act, HRECs are registered with AHEC and report annually on their membership and other aspects of their operations.

14.67 One option for reform would be to enact legislation that requires human genetic research to be approved by an HREC constituted in accordance with the National Statement, and for HRECs to be registered with AHEC. Non-compliance with these requirements could be punishable by fine or civil penalty.

14.68 A potential advantage of a registration scheme for human genetic research would be the development, for the first time, of a comprehensive database of all human genetic research being conducted in Australia. Depending on the nature of regulation, this database might include information about organisations conducting human genetic research, their HRECs or information about specific human genetic research projects.⁶⁹

67 *Gene Technology Regulations 2001* (Cth) rr 12–13.

68 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.46].

69 However, there are other ways in which such a database might be developed. Centralised collection of data on human research taking place in Australia could be achieved by the NHMRC placing an obligation on HRECs to provide such data to it, without recourse to new legislation. A proposal to establish a centralised clinical trials database has been under discussion within the NHMRC for a number of years.

Industry self-regulation

14.69 An alternative to legislation might involve new self-regulatory arrangements, under which organisations involved in the conduct of human genetic research might agree to comply with the National Statement or with a code of research ethics that incorporates similar principles of ethical review of research.

14.70 Self-regulatory arrangements sometimes provide for the establishment of industry-specific bodies with varying regulatory functions and powers. These bodies may deal with matters such as developing industry standards and codes of practice, and setting standards of accreditation for members.⁷⁰

14.71 The Inquiry understands that two established industry associations, AusBiotech Limited and Medicines Australia represent virtually all significant private sector players that fund or support human genetic research in Australia. The activities of these bodies might be the appropriate starting point for the development of a self-regulatory scheme.

14.72 However, the ethical conduct of human genetic research may not be an appropriate area for self-regulation, given the potential consequences for research participants and others. There was no support for new self-regulatory approaches in the submissions received by the Inquiry.

Inquiry's views

14.73 The Inquiry has concluded that, given the existing and likely future extent of private sector research, especially that which is not carried out in conjunction with public sector researchers or research institutions, new mechanisms are required to achieve compliance with the National Statement.

14.74 However, the Inquiry does not recommend the immediate enactment of new legislation to enforce compliance with the National Statement or to subject human genetic research to a new licensing or registration regime.⁷¹

14.75 There are many issues involved in the design and implementation of an effective regulatory scheme for human genetic research and these need to be addressed in more detail. These include:

70 Examples are the various industry ombudsman offices, professional and trade associations, and industry councils and associations. There is a large number of such schemes: see eg the federal government's self-regulation website has a directory of 45 national self-regulatory schemes: Department of Treasury, *Directory of National Self-Regulatory Schemes*, <www.selfregulation.gov.au>, 20 February 2003.

71 Leaving aside existing or augmented requirements for HRECs to be registered with AHEC. An exception to this position involves the establishment and operation of standing human genetic research databases. The registration of research databases is discussed in Ch 18.

- How should human genetic research be defined for the purposes of the regulatory scheme?⁷² This is important to the coverage of the regulatory scheme and has to take into account other regulatory regimes in areas such as human cloning and stem cell research.
- Who would be the target of regulation? Regulation could apply to individual researchers, research organisations or their HRECs, specific human genetic research projects or to specific collections of genetic samples or information.
- What agency should have responsibility for operating the regulatory scheme? Regulatory responsibility could be given to an existing agency, such as the NHMRC, but this might signal a significant shift in its functions and have resource implications.
- What investigative, enforcement or other functions and powers should be given to the agency with responsibility for operating the new regulatory scheme?
- How can an effective regulatory scheme be designed and implemented so as to preserve the intrinsic nature of ethical obligations and not engender a literal or legalistic attitude toward compliance?

14.76 Many other issues arise, including questions about how new regulation would be funded, the compliance costs for research organisations and how the scheme may affect the international competitiveness of Australian human genetic research.

14.77 Constitutional limits on federal legislative power are an important consideration. The most obvious head of federal legislative power appears to be the corporations power,⁷³ but amendments to the NHMRC Act may be insufficient to regulate effectively all human genetic research. Corresponding legislation may have to be enacted by the Commonwealth, States and Territories.

14.78 These problems are not insurmountable and options for further regulation should be thoroughly reviewed. The Inquiry believes that the NHMRC is the appropriate body to advise government on options for regulating human genetic research conducted in the private sector. Accordingly, the Inquiry recommends that the NHMRC, as part of its review of the National Statement in the 2003–2005 triennium, should review the mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.

72 Human genetic research is defined in the National Statement as a process which enhances ‘understanding of how genes and environmental factors interact to influence the health of individuals and populations and in doing so, generates knowledge with the potential to improve individual and community health’: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, Ch 16.

73 *Australian Constitution* s 51(xx).

Recommendation 14–1. The National Health and Medical Research Council, as part of its review of the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) in the 2003–2005 triennium, should review the mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.

15. Human Genetic Research and Consent

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Introduction

15.1 The concept of consent is fundamental to the legal and ethical conduct of research and to the protection of privacy. This chapter examines issues related to consent in research using genetic samples or information.

15.2 The National Health and Medical Research Council's (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement)¹ generally requires consent to the use of human tissue samples, genetic material and genetic information in medical research. The general consent requirement may be waived by a Human Research Ethics Committee (HREC) in certain restricted circumstances.

15.3 In addition, the *Privacy Act 1988* (Cth) (*Privacy Act*) imposes legal requirements to obtain consent for the collection, use or disclosure of personal information, including genetic information. These requirements may also be waived by

¹ National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

an HREC where collection of the information is carried out in accordance with guidelines under s 95 or s 95A of the *Privacy Act* (the s 95 and s 95A Guidelines).²

15.4 A central concern of this chapter is the extent to which waiver of consent under the National Statement or under the s 95 and s 95A Guidelines may undermine privacy protection. A related issue concerns the extent to which researchers are able, under the National Statement, to obtain consent from research participants for the use of their genetic samples or information for unspecified future research.

The National Statement and consent

15.5 Consent is a basic principle of ethical research. The National Statement states that the consent of participants in research must be obtained, except in some types of research using de-identified data³ and in specified circumstances approved by an HREC.⁴ Consent must be based on information about the purpose, methods, demands, risks, discomforts and outcomes of the research and it must be voluntary and not impaired by any coercion, inducement or influence. It must be given by the participant, where competent, or by a person acting with lawful authority on behalf of a participant who lacks competence.⁵ A person must be able to refuse to participate without giving reasons⁶ or to withdraw consent to further involvement in research.⁷ The circumstances in which consent may be waived in research using genetic samples or information are discussed in more detail below.

Consent and epidemiological research

15.6 The National Statement provides that the consent of participants should generally be obtained for the use of identified or potentially identifiable data for epidemiological research.⁸ An HREC may approve access to identified or potentially identifiable data without consent where the HREC is satisfied that obtaining consent is likely to cause unnecessary anxiety to those whose consent would be sought or would prejudice the scientific value of the research and there will be no disadvantage to the participants. An alternate ground for approving such access is that it is impossible to obtain consent in practice, due to the quantity, age or accessibility of the records. Access to data without consent, on either ground, may be approved only where the

2 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [3.2(b)]; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council.

3 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [1.11].

4 These circumstances include certain specified research involving persons highly dependent on medical care Ibid [6.9]; using information for epidemiological research [14.4]; using human tissue samples [15.8]; or using genetic material or genetic information [16.13].

5 Ibid [1.7].

6 Ibid [1.8]–[1.9].

7 Ibid [1.12].

8 Ibid [14.3]. Epidemiological research is concerned with the description of health and welfare in populations through the collection of data related to health and the frequency, distribution and determinants of disease in populations, with the goal of improving health.

HREC determines that the public interest in the research outweighs to a substantial degree the public interest in privacy.⁹

Consent and the use of human tissue samples

15.7 The National Statement provides that consent should generally be required for collection of human tissue for research purposes.¹⁰ Consent should be voluntary, specific to the purpose for which the tissue is to be used and follow the provision of full information about the research, including advice as to whether any remaining tissue samples are to be stored following completion of the research.¹¹

15.8 It is important to distinguish between two situations in which stored human tissue is used for research.

- Tissue samples may be collected primarily for use in research and stored in collections maintained by researchers and research organisations. Chapter 18 considers the issues related to human genetic research databases¹² (often referred to as ‘tissue banks’).
- Tissue samples may be collected primarily for therapeutic and diagnostic purposes and stored in collections maintained by hospitals or pathology laboratories, and then used in research. These human tissue collections are discussed in Chapter 19.

15.9 The National Statement provides that where it is proposed that human tissue samples previously collected and stored with consent for research be used for a different research purpose, separate consent for the different research should be obtained.¹³ It also states that consent should be obtained for the use of human tissue samples that have been collected and stored after clinical procedures or held in ‘archives or banks’, in any research which may lead to harm or injustice or be of benefit to the donor.¹⁴ An HREC may waive the requirement for individual consent to the use of human tissue samples in research in restricted circumstances after taking into account certain matters.¹⁵

Consent and the use of genetic material or genetic information

15.10 The National Statement separately provides that consent should generally be required for the use of stored genetic material or genetic information.¹⁶ Those from whom consent is sought must be informed about a range of prescribed matters, including whether it is intended to store their genetic material and information for as

9 Ibid [14.4].

10 Ibid [15.4].

11 Ibid [15.5].

12 As that term is defined in Ch 18.

13 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [15.6].

14 Ibid [15.7].

15 Ibid [15.8].

16 Ibid [16.12].

yet unspecified future research;¹⁷ whether their genetic material is to be disposed of on completion of research;¹⁸ and that participants may withdraw from the research and either request disposal of their genetic material and information or that it be retained in de-identified form.¹⁹

15.11 When human genetic research reveals information important to the future health of an identified or potentially identifiable participant or his or her offspring, the research protocol must provide for the same consent, counselling and confidentiality protection as would apply in a clinical setting.²⁰ If participants are asked to consent to the use of their genetic material or information in future research, information and counselling about possible consequences should be provided.²¹ The National Statement notes that in general, genetic material and information will be used for future research in de-identified form and feedback will not be possible.²²

15.12 An HREC may waive the requirement for consent to participation in human genetic research. In reaching that decision, the HREC may consider factors that mirror those applicable to waiving consent to the use of human tissue samples.²³

15.13 In addition, the National Statement anticipates that institutions or organisations may wish to conduct research on genetic material and information collected for non-research purposes. It states that such institutions or organisations should develop and disseminate a general policy that informs patients that such material and information may be used for future research following HREC approval, and give patients an opportunity to opt out of participation in such research.²⁴

The Privacy Act and consent

15.14 Under National Privacy Principle (NPP) 10.3 of the *Privacy Act*, health information may be *collected* without consent for research purposes if obtaining consent is impracticable, de-identified information would not be suitable, and the collection is in accordance with guidelines issued by the NHMRC and approved by the Privacy Commissioner under s 95A of the Act (the s 95A Guidelines).²⁵

15.15 Under NPP 2.1(d), health information may be *used or disclosed* without consent for research purposes if obtaining consent is impracticable, the use or disclosure is conducted in accordance with the s 95A Guidelines and, in the case of

17 Ibid [16.10(j)]. Conducted in accordance with [16.12] and [16.16].

18 Ibid [16.10(k)].

19 Ibid [16.10(l)].

20 Ibid [16.15].

21 Ibid [16.16].

22 Ibid [16.16].

23 Ibid [16.13].

24 Ibid [16.14].

25 *Privacy Act 1988* (Cth) NPP 10.3(a)–(d)(iii). Information may also be collected as required by law or in accordance with ‘rules established by competent health or medical bodies that deal with obligations of professional confidentiality’: NPP 10.3(d)(i)–(ii).

disclosure, it is reasonably believed that the recipient will not disclose the information.²⁶

15.16 When a proposal for medical research would involve a breach of the Information Privacy Principles (IPPs), consent requirements may be waived in accordance with the s 95 Guidelines. The s 95 Guidelines apply to medical research that involves use or disclosure of personal information, typically without the consent of the person to whom it relates, where the information is held by a Commonwealth agency.

15.17 The Privacy Commissioner may approve s 95 Guidelines (in relation to the IPPs) and s 95A Guidelines (in relation to the NPPs) only where he or she is satisfied that the 'public interest' in research or in the use and disclosure of health information in accordance with the guidelines 'substantially outweighs the public interest' in maintaining adherence to the IPPs or the NPPs.²⁷

15.18 The s 95 and s 95A Guidelines provide that an HREC should not approve research that would otherwise breach the *Privacy Act* unless it considers that the public interest in the research outweighs to a substantial degree the public interest in privacy. Each set of guidelines provides a similar framework for weighing these interests.²⁸ HRECs are directed to consider certain specified matters, including the value and public importance of the research, the likely benefits to the participants, whether the research design can be modified, the financial costs of not proceeding with the research, the type of personal information being sought, the risk of harm to individuals and the extent of possible breach of privacy.²⁹

15.19 Non-compliance with the s 95 Guidelines may lead to researchers being named in the NHMRC's annual report or in a report to a Commonwealth agency or the federal Privacy Commissioner.³⁰ Where the conduct of an organisation or agency is in breach of the *Privacy Act*, affected individuals may complain to, and have their complaints investigated by an adjudicator under an approved privacy code or by the federal Privacy Commissioner.³¹

15.20 The s 95 and s 95A Guidelines do not apply to the collection, use and disclosure of health or other personal information in research by individuals or organisations that are not covered by the *Privacy Act*. For example, the *Privacy Act* does not apply to state public sector entities, including public teaching hospitals and

26 Ibid NPP 2.1(d).

27 Ibid ss 95(2), 95A(3).

28 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [3.3]; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council, D.5.

29 See National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [3.3(a)]–[3.3(h)]; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council [D.5(a)]–[D.5(k)].

30 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [4.3].

31 See *Privacy Act 1988* (Cth) Pt V.

associated research bodies, where such bodies are established for a public purpose under a law of a State.³² However, these organisations may be covered by state legislation.³³

15.21 As discussed in more detail in Chapter 21, a disclosure that is permitted by the *Privacy Act* may nonetheless breach the common law. While the *Privacy Act* permits the disclosure of patient health information to medical researchers without consent,³⁴ disclosure may nevertheless breach a common law duty of confidentiality. The determination of an HREC that personal information can ethically be disclosed does not necessarily provide protection against an action for breach of confidence.³⁵

Waiver of consent

Operation of the National Statement

15.22 The National Statement recognises that there may be circumstances in which the use of identifiable information in research may be justified without complying with the requirement of individual consent from all participants. This can occur in epidemiological research but is also possible for human tissue research and genetic research.

15.23 The National Statement provides that HRECs may ‘sometimes’ waive consent after taking into account a number of factors. The matters that may be taken into account are:

- the nature of any existing consent relating to the collection and storage of the sample;
- the justification presented for seeking waiver of consent including the extent to which it is impossible or difficult or intrusive to obtain specific consent;
- the proposed arrangements to protect privacy including the extent to which it is possible to de-identify the sample;
- the extent to which the proposed research poses a risk to the privacy or well being of the individual;

32 Ibid ss 6C(1), 6C(3)(c).

33 For example, *Health Records Act 2001* (Vic) Health Privacy Principle 1.1(e)(iii); 2.2(g)(iii). These provisions allow for the collection, use and disclosure of health information for research without the consent of the individuals concerned in accordance with guidelines issued or approved by the Health Services Commissioner under s 22.

34 *Privacy Act 1988* (Cth) NPP 2.1(d).

35 It has been suggested that the law should be clarified to ensure that the use of personal information in medical research in circumstances where it would constitute a breach of confidence shall be lawful, provided the research has received ethics approval: Law Reform Commission of Western Australia, *Report on Confidentiality of Medical Records and Medical Research*, 65 (1990), Law Reform Commission of Western Australia, Perth, Pt II [6.1]; Rec 1. See also C Thomson, ‘Records, Research and Access: What Interests Should Outweigh Privacy and Confidentiality? Some Australian Answers’ (1993) 1 *Journal of Law and Medicine* 95; R Magnusson, ‘Confidentiality and Consent in Medical Research’ (1995) 17(4) *Sydney Law Review* 549, 549.

- whether the research proposal is an extension of, or closely related to, a previously approved research project;
- the possibility of commercial exploitation of derivatives of the sample; and
- relevant statutory provisions.³⁶

Waiver of consent and tissues obtained for clinical purposes

15.24 Waiver of consent issues often arise where researchers propose to access and use material collected for therapeutic or diagnostic purposes, such as pathology samples.³⁷ The individuals from whom such samples have been taken may not have consented to the use of these samples in any research at all, let alone in unspecified research. Human tissue collections are discussed in detail in Chapter 19.

15.25 It is easier to argue that consent should be waived for new research on tissue originally collected for research purposes than for research on tissue originally collected for other purposes. In the latter case, there can be no doubt that research is an unrelated secondary purpose rather than a purpose that is related³⁸ (or directly related in the case of health information) to the primary purpose and within the reasonable expectations of the individual concerned.³⁹ On the other hand, requirements to seek consent to the use of archival tissue will often be difficult in practice because it may no longer be possible to contact the individuals to whom the samples relate.

15.26 Where tissue derived from clinical practices is used for the purposes of quality assurance, rather than for research, consent requirements may be avoided. The Office of the Federal Privacy Commissioner's *Guidelines on Privacy in the Private Health Sector* state that an organisation's quality assurance or clinical audit activities may constitute directly related secondary purposes, for which consent may not be required.⁴⁰ The Australian Health Ethics Committee (AHEC) has recently released

36 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [15.8].

37 It should be understood that the vast majority of stored clinical samples are never used for research but are retained, in accordance with industry standards and guidelines, for quality assurance, medico-legal and other reasons.

38 Research may be a related purpose where it is connected with the clinical care of the patient: '[t]here is no dividing line between clinical care and research and the clinician/scientist has a duty to his/her patient to conduct every possible test to find the cause and best treatment for a disease': Australian Academy of Science, *Submission G097*, 21 January 2002.

39 That is, in terms of *Privacy Act 1988* (Cth) NPP 2.1. It has been suggested that more research is needed on community attitudes to the use of genetic information without specific consent: L Skene, 'The Genetics Debate: Why Doctors Must be Heard' (Paper presented at 2001 UMMS Lecture, 15 November 2001). The article refers to the unpublished results of a survey conducted by Patterson and Gillam at ethics committee seminars in which 29% of 177 respondents stated that they wanted to be contacted for consent to the use of their stored tissue in research, 40% if the research was on a disease that affected them and 52% if there was a possibility of commercialisation.

40 Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 14–15.

guidance for institutions, HRECs and health professionals on when quality assurance in health care requires ethics review.⁴¹

15.27 Several submissions expressed concern about the use of clinical tissue. One submission, from a woman whose family is affected by Huntington's disease, stated:

The collection and use of human tissue samples certainly should be regulated. Tissue samples could be used for research, but the samples should be de-identified first. People who have tests done for therapeutic reasons may not be aware that they have 'interesting' DNA. ... When these tissue samples are used for genetic research, then that is when the researchers have crossed the 'consent' line.⁴²

15.28 The Androgen Insensitivity Syndrome Support Group Australia (AIS Support Group) submitted that HRECs should never provide a waiver of consent under the National Statement⁴³ but recognised that such a prohibition would present considerable difficulties in some circumstances and, in particular, in relation to the use of samples initially obtained for therapeutic or diagnostic purposes.⁴⁴

15.29 Privacy NSW raised the specific concern that the National Statement 'effectively dispenses with the need for fully informed consent' where a person's genetic information is not initially collected for research purposes but is later used for research.⁴⁵

De-identification and consent

15.30 As discussed in Chapter 16, the National Statement makes distinctions between identified, potentially identifiable and de-identified personal information or material.⁴⁶ The concept of de-identification is relevant in several ways to issues of consent to participation in research, and to the privacy and ethical implications of consent requirements.⁴⁷

15.31 The National Statement provides that when consent is being sought from individuals for the collection of genetic material or information, they should be informed whether their genetic material and information will be used in an identified, potentially identifiable or de-identified form.⁴⁸

41 Australian Health Ethics Committee, *When does Quality Assurance in Health Care Require Independent Ethical Review?* (2003), AHEC, Canberra.

42 Confidential Submission G051CON, 14 January 2002.

43 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

44 Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002.

45 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

46 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 9.

47 Issues concerning the de-identification of genetic samples and the coverage of the *Privacy Act 1988* (Cth) are discussed in Ch 8.

48 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.10(b)].

15.32 Coded data is not ‘de-identified’ for the purposes of the National Statement because coding, by its very nature, is reversible.⁴⁹ Unlike the *Privacy Act*, the provisions of the National Statement continue to apply to de-identified material and information. However, where information is completely de-identified it may be impossible to seek consent to its further use from individuals.⁵⁰

15.33 The extent to which coded data can be considered to be de-identified is not entirely clear under either the *Privacy Act* or the National Statement. For example, where an independent person (for example, a gene trustee) holds the code and it is not possible for the researchers to ascertain the identity of the individual concerned (without the assistance of the independent person), is the information de-identified so that the *Privacy Act* does not govern it, or does it depend on the circumstances and, in particular, the practices or policies of the gene trustee?

15.34 The Victorian Department of Human Services Genetics Advisory Committee considered that specific consent should not be necessary for ‘de-identified research’.⁵¹ Other submissions also suggested that specific consent to the use of clinical samples in research should not be required where the samples are de-identified.⁵² These views are consistent with conclusions of the United Kingdom Human Genetics Commission, which has stated that ‘it is acceptable to seek general consent in cases where there is to be irreversible or reversible anonymisation of data and samples’.⁵³

Reform of the waiver of consent provisions

15.35 DP 66 asked whether changes are needed to the National Statement or the s 95 or s 95A Guidelines, in relation to waiver of consent by HRECs.⁵⁴ In this context, it is important to appreciate that the s 95 and s 95A Guidelines apply only to Commonwealth agencies and private sector organisations, respectively. In contrast, the waiver of consent provisions of the National Statement are intended to apply to all human research conducted in Australia, notwithstanding that the National Statement lacks legislative force (see Chapter 14).

49 Although one of the factors that may be taken into consideration by an HREC is determining whether consent should be waived is the ‘extent to which it is possible to de-identify the sample’—possibly suggesting that de-identification need not be complete anonymisation: *Ibid* [15.8], [16.13].

50 However, consent may still need to be sought from collectivities where research involves a collectivity. See *Ibid*, Ch 8.

51 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002.

52 Association of Australian Medical Research Institutes, *Submission G007*, 27 April 2001; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; *Confidential Submission G051CON*, 14 January 2002.

53 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 95.

54 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Question 12–2.

15.36 Some submissions argued for significant reform of the waiver of consent provisions in the National Statement, the s 95 and s 95A Guidelines, or both. In relation to the current operation of the waiver of consent principles in the National Statement, the Centre for Law and Genetics submitted that:

These provisions may require rewriting to emphasise that, where commercial considerations are involved, or where the sample is identified or potentially identifiable (which is usually the case), these principles should not be used to avoid the primary requirement for participant consent. The current high levels of public support for medical research may be damaged irrevocably by misuse of these provisions.⁵⁵

15.37 The Centre for Law and Genetics also stated that the original intent of these provisions was that such applications should be ‘exceptional’.⁵⁶ Privacy NSW expressed similar concerns about the waiver provisions, observing that the National Statement states simply that an HREC *may* take into account the listed matters.

This is not a mandatory requirement and would be difficult to enforce, nor are the listed factors sufficient to ensure that consent is protected in all but strictly defined and exceptional circumstances.⁵⁷

15.38 Privacy NSW emphasised that explicitly defined and enforceable ethical standards should govern the circumstances in which consent may be waived in research involving genetic information and that consent should only be waived in very limited, closely defined, circumstances.⁵⁸

15.39 A number of submissions shared the concerns expressed by Privacy NSW about the non-mandatory nature of the factors to be taken into account in making a waiver decision. The Office of the Federal Privacy Commissioner (OFPC) stated that:

In the interests of maintaining confidence in the HREC system, consideration could be given to making it obligatory for a HREC to consider those factors.⁵⁹

15.40 Other submissions, including from those involved in the conduct or funding of medical research, considered that the existing consent requirements in the National Statement were adequate and appropriate.⁶⁰

15.41 Jeremy Kenner, Ethics Coordinator at the Peter MacCallum Cancer Institute (PMCI), stated that the current guidelines ‘strike the necessary balance between the interests of the research community, research participants and the public exceptionally

55 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

56 Ibid.

57 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

58 Ibid.

59 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

60 Peter MacCallum Cancer Institute, *Submission G028*, 20 December 2001; Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; NSW Health Department, *Submission G303*, 13 January 2003.

well'.⁶¹ The NHMRC Research Committee made a similar point. The Committee noted that HRECs are intended to function as an 'autonomous body which is particularly responsive to the environment and community group with which it is associated'.

Reducing or changing the autonomy of the HRECs by selecting out specific issues over which that body no longer has the final say is acceptable but only if the reason is well justified.⁶²

15.42 The Department of Human Services Victoria Genetics Advisory Committee stated that:

The NH&MRC's National Statement already contains specific and we believe adequate provisions dealing with the use of human tissue samples in research. We believe that the current system of approval and oversight by Human Research Ethics Committees has served the community well, and should not be replaced by a more restrictive system.⁶³

15.43 In relation to the s 95 and s 95A Guidelines, Privacy NSW submitted that the existing framework does not adequately address the specific privacy issues in relation to the use of genetic information for research.

The Guidelines only operate where consent is not sought or obtained. They only apply to the collection and disclosure of identifiable personal information. Given the difficulty in truly deidentifying genetic samples, it would generally be appropriate to treat 'de-identified' genetic information in accordance with the principles contained in the Guidelines and the National Statement.⁶⁴

15.44 Privacy NSW was also critical of the 'public interest' test under which the s 95 and s 95A Guidelines are approved by the Privacy Commissioner and under which HRECs determine whether to approve collection, use or disclosure of health information without consent. As discussed above, the test in each case refers to whether the public interest in research 'substantially outweighs' the public interest in privacy.⁶⁵

It is of concern that this effectively utilitarian test does not address the substantial ethical issues ... in relation to obtaining consent in relation to genetic information. The permissive scope of the public interest test, and its application by HRECs under conditions of limited oversight, also conflicts with the need for consistent and transparent principles that are applied regardless of where information is collected, used and disclosed.⁶⁶

61 Peter MacCallum Cancer Institute, *Submission G028*, 20 December 2001.

62 National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002.

63 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002.

64 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002. The Law Institute of Victoria agreed with these views: Law Institute of Victoria, *Submission G275*, 19 December 2002.

65 *Privacy Act 1988* (Cth) ss 95(2), 95A(3); National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [3.2(b)]; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council [D.4].

66 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

Survey of HRECs

15.45 In order to assess whether there is any need to reform the waiver of consent provisions of the National Statement to take account of any special characteristics of human genetic research, the Inquiry sought to understand more fully how these provisions operate in practice.⁶⁷

15.46 In late 2002, the Inquiry sent questionnaires to the Executive Officers of 216 HRECs, requesting information about ethics review of human genetic research proposals considered between 1 January and 31 December 2002. The questionnaire asked for the number of human genetic research proposals reviewed, approved and denied. The questionnaire then asked for information about HREC consideration of, and decisions about, requests for access to genetic samples or information without consent—that is, about HRECs' exercise of the authority given to them by the National Statement to waive consent and, in particular, about the reasons for waiving consent.

15.47 In response, the Inquiry received 82 completed questionnaires (a response rate of 38%). Twenty-six HRECs had considered 129 proposals involving human genetic research in the reporting period. Ten HRECs had considered 22 proposals that were not designed to obtain the consent of individuals to the use of their genetic samples or information. HRECs decided to waive consent in respect of 17 human genetic research proposals. This amounts to 13% of the human genetic research proposals considered by HRECs in the reporting period, and 77% of proposals where waiver was sought.

15.48 Consultations during the Inquiry identified that the circumstances in which HRECs waive consent typically involve the use of archival tissues originally collected for therapeutic or diagnostic purposes.⁶⁸ The Inquiry noted that waiver of consent may be sought where clinical samples have been collected from a cohort of individuals with serious illnesses, such as cancer. Researchers may be reluctant to seek consent because many of the subjects of the research may have died recently, or may be dying.⁶⁹

15.49 The survey results provided confirmation of this general picture of the circumstances in which waiver of consent is considered by HRECs. At least seven of the research proposals for which waiver was granted involved the use of archival tissue originally taken for therapeutic or diagnostic purposes. The existence of prior prospective consent for research was a factor in at least five research proposals for which waiver was granted.

67 The NHMRC Research Committee referred to the importance, in this context, of knowing how many waivers have actually been approved by HRECs: National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002.

68 HREC Chairs and Officers, *Consultation*, Sydney, 20 June 2002.

69 *Ibid.* In such circumstances, researchers may submit that it is 'impossible or difficult or intrusive' to obtain consent in terms of the National Statement: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [15.8], [16.13]. However, the mere fact that distress may be caused may be insufficient to avoid the consent requirements of the *Privacy Act*, which requires that it be impracticable to collect health information without consent: *Privacy Act 1988* (Cth) NPP 10.3(c). The OFPC emphasised that what may be considered to be impracticable is a 'critical consent issue': Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

15.50 Given the limited nature of the survey, and the low response rate of HRECs, the Inquiry has concluded that there is insufficient information about the operation of the waiver of consent provisions of the National Statement to justify recommending specific reforms. However, as discussed below, the Inquiry does recommend augmented reporting requirements in relation to waiver of consent. Once these reporting requirements have been in operation for a period of time, it may be appropriate for the NHMRC to review this issue.

HREC reporting of waiver of consent

15.51 HRECs report to AHEC on various matters related to their membership and procedures.⁷⁰ These reporting obligations, and proposals for their augmentation, are discussed more generally in Chapter 17. HRECs are specifically required to report annually to AHEC on decisions made under the s 95 and s 95A Guidelines.⁷¹ However, there is no requirement for HRECs to report on waiver of consent under the National Statement itself.

15.52 As discussed above, the s 95 and s 95A Guidelines have limited operation and apply only to Commonwealth agencies and private sector organisations respectively. Much human genetic research in Australia is conducted by universities, teaching hospitals and publicly funded research institutes. Further, the extent of the existing reporting requirements under the s 95 and s 95A Guidelines have been criticised. Privacy NSW stated that

it is not clear that the reporting and auditing processes in any of these instruments are sufficiently transparent and accountable to effectively protect people's genetic information privacy. Accordingly there is a real risk that Sections 95 and 95A will facilitate rather than restrict the conduct of research involving human genetic information other than in accordance with strict ethical principles and practices.⁷²

15.53 In DP 66, the Inquiry proposed that HRECs should be required to report annually to AHEC with respect to human genetic research proposals for which waiver of consent has been granted under the National Statement.⁷³ There was support for this proposal in submissions.⁷⁴ For example, the Department of Health Western Australia submitted that

70 See Australian Health Ethics Committee, *AHEC HREC Annual Report Form*, <www.nhmrc.gov.au/issues/pdfcover/annrpt.htm>, 20 February 2003.

71 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.46]–[2.48]; National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra [3.4]; [4.1]–[4.3]; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council [D.6], [E.1]–[E.3].

72 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

73 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 12–1.

74 Law Institute of Victoria, *Submission G275*, 19 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February

waiver of consent should be allowed by an HREC only after detailed and complex consideration of issues relating to appropriate compliance with the National Statement, privacy legislation and privacy guidelines. As such, compliance reporting and the potential for audit of these decisions of the HRECs are both desirable.

If there are any ongoing concerns as a result of such audit, consideration should be then given to the development in guidelines of more defined circumstances under which such waiver of consent may be applied.⁷⁵

15.54 Jennifer Fleming submitted that:

From a risk management perspective, where protocols involving waiver of consent have been identified as approved by HRECs it would be valuable for the HREC (in consultation with the researcher(s)) to provide a brief statement of justification for that waiver. An AHEC secure database could be held to include (i) responses to the Annual Compliance Reports and (ii) a brief listing of approved protocols where rationales for waiving consent have been endorsed. This would be valuable in terms of promoting transparency of practice, monitoring and education.⁷⁶

15.55 The Inquiry also received a range of comments on the possible content of new reporting obligations. These submissions generally anticipated that reporting would focus on the reasons HRECs grant waiver and, in particular, would address the factors set out in the National Statement and s 95 and s 95A Guidelines.⁷⁷

15.56 The Inquiry agrees with comments made in submissions that new reporting requirements will be of limited benefit, and merely add a new burden to HRECs, unless processes are put in place to ensure that AHEC analyses the information received and provides feedback to HRECs to assist with future ethics review.⁷⁸ The Queensland Government noted that

2003; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; J Fleming, *Submission G241*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003. The AIS Support Group suggested that HRECs should be required to provide written advice to AHEC and to the HGCA within 28 days of making a decision to grant approval to conduct research with a waiver of consent: Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003. The Department of Human Services South Australia suggested that HRECs should also report the number of genetic research proposals they receive. The proposal was opposed by Medicines Australia: Medicines Australia, *Submission G302*, 12 February 2003.

75 Department of Health Western Australia, *Submission G271*, 23 December 2002.

76 J Fleming, *Submission G241*, 20 December 2002.

77 Centre for Law and Genetics, *Submission G255*, 21 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Queensland Government, *Submission G274*, 18 December 2002.

78 Queensland Government, *Submission G274*, 18 December 2002; NSW Health Department, *Submission G303*, 13 January 2003.

if [reporting of waiver] is implemented because of concerns regarding the practices or accountability of the HRECs in waiving consent, further consideration should be given to the processes of establishing and administering the HRECs to ensure compliance with national principles rather than merely adding an extra reporting burden.⁷⁹

15.57 The general requirement to obtain consent in research is fundamental to the protection of interests in privacy and ethical treatment of an individual's genetic samples and information. The Inquiry has concluded that the transparency and accountability of the present system would be better served by requiring HRECs to report specifically on *all* research proposals for which waiver of consent has been granted, and not just those for which waiver has been granted under the s 95 and s 95A Guidelines. This would enable AHEC to record, assess and monitor the operation of these provisions of the National Statement, and provide feedback to HRECs.

15.58 As an interim measure, the existing provisions of the National Statement could be used as the mechanism for extended reporting of waiver of consent. HRECs are currently required to provide information from their records to the NHMRC on request.⁸⁰

15.59 The Inquiry also notes that the relevant provisions of the National Statement pre-date the enactment of the *Privacy Amendment (Private Sector) Act 2000* (Cth) and the approval of the s 95A Guidelines. For this reason, the NHMRC should review the provisions of the National Statement relating to waiver of consent and reporting of decisions, to ensure they are consistent with privacy law.

15.60 The Inquiry further recommends that the NHMRC, in strengthening the level of training and other support provided to HRECs, in accordance with the recommendations in Chapter 17, should ensure that attention is given to the interpretation of the waiver of consent provisions and HREC decision making in relation to waiver.

Recommendation 15–1. The National Health and Medical Research Council (NHMRC), as part of its review of the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) in the 2003–2005 triennium, should amend the National Statement to provide that Human Research Ethics Committees (HRECs) must report annually to the Australian Health Ethics Committee (AHEC) with respect to human genetic research proposals for which waiver of consent has been granted under the National Statement. Until such time as the National Statement has been so amended, the NHMRC should exercise its existing authority to request information from HRECs to require them to report annually to AHEC with respect to such human genetic research proposals.

79 Queensland Government, *Submission G274*, 18 December 2002.

80 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.47].

Recommendation 15–2. The NHMRC, as part of its review of the National Statement in the 2003–2005 triennium, should ensure that the provisions of the National Statement relating to waiver of consent and reporting of decisions are consistent with privacy laws and, in particular, with guidelines issued under s 95 and s 95A of the *Privacy Act 1988* (Cth).

Recommendation 15–3. The NHMRC, in strengthening the level of training and other support provided to HRECs in accordance with Chapter 17 of this Report, should ensure that adequate attention is given to: (a) the interpretation of the waiver of consent provisions of the National Statement; and (b) HREC decision making in relation to such waiver.

Specific consent

15.61 One question that frequently arose in submissions⁸¹ and consultations⁸² related to how specific consent needs to be. In particular, concerns were raised about whether researchers should be able to obtain consent to the future unspecified use of genetic samples or information and, if so, subject to what processes or safeguards.

The National Statement, the Privacy Act and specific consent

15.62 Under the National Statement, obtaining consent should involve:

(a) the provision to participants, at their level of comprehension, of information about the purpose, methods, demands, risks, inconveniences, discomforts, and possible outcomes of the research (including the likelihood and form of publication of research results); and

(b) the exercise of a voluntary choice to participate.⁸³

15.63 This guidance appears to be consistent with the meaning of consent as it is used in the *Privacy Act*. The *Guidelines on Privacy in the Private Health Sector* state that there are three key elements involved in seeking consent to use health information in particular ways: consent must be provided voluntarily, the individual must be adequately informed, and the individual must have capacity to understand, provide and communicate his or her consent.⁸⁴

81 For example, Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; Queensland Institute of Medical Research, *Submission G036*, 14 January 2002; Peter MacCallum Cancer Institute, *Submission G070*, 20 December 2001; Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002; Genetic Support Council WA, *Submission G112*, 13 March 2002; M Dunne, *Submission G041*, 17 December 2001; N Zeps, *Submission G047*, 14 January 2002.

82 For example, Queensland Institute of Medical Research, *Consultation*, Brisbane, 13 December 2001; Children's Cancer Research Institute, *Consultation*, Perth, 4 December 2001; Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 15 March 2002; L Skene, *Consultation*, Melbourne, 15 March 2002.

83 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [1.7].

84 Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, xii. The definition of 'consent' under the *Privacy Act* includes 'express or implied' consent: *Privacy Act 1988* (Cth) s 6.

15.64 The National Statement provides that consent to the collection of human tissue for research purposes should be specific to the purpose for which the tissue is to be used.⁸⁵ Where it is proposed that human tissue samples previously collected and stored with consent for research be used for a different research purpose, separate consent for the different research should be obtained.⁸⁶

15.65 The provisions relating to the use of genetic material and information more clearly anticipate that researchers may seek consent to ‘as yet unspecified future research’. Paragraph 16.10(j) of the National Statement provides that when consent is being sought for prospective collection of genetic material and information, individuals should be informed

about any intention to store their genetic material and information for as yet unspecified future research in accordance with paragraphs 16.12 and 16.16 below. If consent is given, the duration of storage should be specified. If consent for future research use is refused, the genetic material and information should be disposed of at the end of the research, once the sample storage and record keeping requirements of good research practice have been met.⁸⁷

15.66 The NHMRC *Human Research Ethics Handbook* notes, in its commentary on paragraph 16.10(j) of the National Statement, that while consent should normally be specific,

the nature of genetic research may make it difficult to be specific at the time of collection. It is appropriate to ask potential research participants to consent to storage of genetic material for unspecified future use, on the basis that consent will be sought at the time a specific research project is identified.⁸⁸

15.67 According to this interpretation of the National Statement, while researchers may ask participants to consent to the storage of genetic material, further consent to the use of that material in future research will normally need to be sought.

De-identification and specific consent

15.68 The following discussion about obtaining consent to unspecified future use of genetic samples or information is premised on the information or samples remaining identified or potentially identifiable. If information or samples are stored in de-identified form so that that no link can be made between the information or samples and the individuals to whom they relate, the ethical and privacy interests of those individuals are reduced, though not necessarily eliminated.

85 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [15.5(b)].

86 Ibid [15.6].

87 Ibid [16.10(j)].

88 National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), NHMRC, Canberra [C105].

Interpretation of consent requirements

15.69 Many submissions, notably from medical researchers and research organisations, expressed concerns that too strict an interpretation of specific consent requirements would handicap the conduct of genetic research. The Human Genetics Society of Australasia (HGSA) expressed concerns that privacy legislation

gives primacy to privacy (and the consent requirements) making it increasingly difficult to use stored samples, even in de-identified form. It is the experience of the membership of the HGSA that the community is supportive of research, in general and when asked personally to participate. Current privacy legislation, and the application of that legislation, constrains that community support by setting in place requirements that are likely to lead to the situation in which specific current consent is the only acceptable mechanism for using a stored tissue sample for research – it would be unfortunate if that proves to be the case.⁸⁹

15.70 Dr Joe Sambrook from the PMCI submitted that the requirements of genetic research cannot be served by ‘restricting the use of patient material or information to a single specified project or a specific time-span’.

Instead consent should be obtained in a general fashion to enable tissue to be used for both present and future research purposes, even though those purposes may not be precisely defined.⁹⁰

15.71 The Victorian Department of Human Services Genetics Advisory Committee considered that specific consent should not be necessary for de-identified research. It should be sufficient for ‘generic consent’ for de-identified research to be obtained to enable tissue to be used for both present and future research purposes, even though those purposes may not be precisely defined.

We believe that the standard of ‘general’ consent for use in research, combined with the waiver system (described in 15.8 of the National Statement) and review of the use of tissue in specific research projects by a HREC, constitute a model that best protects both the interests of patients and the research community.⁹¹

15.72 Other groups considered that current guidelines relating to consent were inadequate to protect the interests of those whose genetic material or information may be used for research. The Genetic Support Council WA stated that where future research purposes are contemplated but not known at the time consent is given, further written consent from the individual should be required.⁹² The AIS Support Group asked:

Can a person who has no real understanding of the potential ultimate uses of genetic information, truly give informed consent for the taking of a tissue sample for research purposes? Should express consent be obtained each and every time a researcher seeks

89 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

90 Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002.

91 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002.

92 Genetic Support Council WA, *Submission G112*, 13 March 2002.

to use a tissue sample for research? If consent is obtained in an environment where it is felt that adequate understanding of the nature of research exists, is such consent perpetual?⁹³

15.73 The AIS Support Group observed that the complexities of the issues involved require ‘at least a reasonable base level of specific medical or scientific knowledge’ and that to ask a person to give consent to the use of their tissue samples for research purposes without this knowledge ‘challenges the very concept of informed consent’.

15.74 Some submissions emphasised the importance of distinguishing between concepts of consent in the clinical and research settings.⁹⁴ The best known legal test for determining what information doctors should give patients before a medical procedure was established in *Rogers v Whitaker*.⁹⁵ Doctors have an obligation to inform patients about the material risks of a proposed procedure. A risk is considered material if the patient would be likely to attach significance to it.⁹⁶

15.75 Such a test may be appropriate where research involves clinical intervention, but is not necessarily an appropriate guide to how much information should be provided by researchers to research participants about the proposed use of genetic samples or information in medical research.⁹⁷

Defining the research purpose

15.76 Concerns about the practical implications of specific consent requirements were emphasised by individuals and organisations involved in large scale medical research projects using major collections of genetic samples for multiple research projects. For example, the Queensland Institute of Medical Research (QIMR) noted that its studies depend upon

our ability to correlate different measures on the same people over long periods of time. To require that we go back to subjects every time we want to do a different assay on their blood, or type a different gene, would impose an unsustainable burden. Were this to be mandated, it would close us down. We trust that decisions on these issues will continue to be left to the discretion of local [HRECs]. We believe that this system is currently working well for both subjects and researchers.⁹⁸

15.77 The PMCI supported the existing principles relating to consent contained in the National Statement but warned that these principles should not be interpreted

to mean that either retrospective or prospective consent is required for use of genetic information or human tissue in a specific *study* (as opposed to *purpose*) that was not contemplated at the time that the consent was originally obtained. This interpretation,

93 Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002.

94 Peter MacCallum Cancer Institute, *Submission G028*, 20 December 2001; L Skene, *Consultation*, Melbourne, 15 March 2002.

95 *Rogers v Whitaker* (1992) 175 CLR 479.

96 *Ibid*, 634.

97 L Skene, *Consultation*, Melbourne, 15 March 2002.

98 Queensland Institute of Medical Research, *Submission G036*, 14 January 2002.

analogous to that operative in the clinical setting, is not appropriate in the research setting and would seriously impede the research endeavour. Therefore, the only modification that need be made to the current principles, as delineated, is a clear distinction between the application of the informed consent requirement in the clinical setting and the research setting.⁹⁹

15.78 Similar points were made in other submissions and in consultations with researchers.¹⁰⁰ The view was put that it should be sufficient, for example, to obtain consent to a broad research purpose, such as ‘biochemical and genetic studies in cancer’¹⁰¹—without the need to specify exactly what sorts of analysis might be performed on genetic material. The emergence of new genetic research technologies, such as micro-array technology, may mean that researchers wish to re-test samples and this should not necessitate fresh consent, if it is for the same broad research purpose.¹⁰²

15.79 The National Statement refers to a requirement that consent to the use of human tissue samples be specific to the ‘purpose’ for which the tissue is to be used¹⁰³ and that where previously collected samples are to be used for a different ‘research purpose’ further consent should generally be obtained.¹⁰⁴

15.80 While it has been suggested that the parameters of this provision might be clarified,¹⁰⁵ the Inquiry does not believe that any change is necessary. It seems sufficiently clear that a distinction may be made between a broad research purpose and the particular study or research techniques that may be used in the pursuit of that purpose.

Consent to unspecified future research

15.81 It is common practice for researchers to seek consent from donors for their tissue to be used in a particular experiment and then stored for possible later inclusion in other experiments, the details and potential implications of which are unspecified.¹⁰⁶ This practice has been criticised as inappropriate because, as a matter of principle, valid consent cannot be provided in the absence of full and complete disclosure of the uses to which the tissue is to be put.¹⁰⁷

15.82 However, there was also broad consensus within the medical research community about the need for procedures under which prospective research participants may provide consent for unspecified future research—provided that such

99 Peter MacCallum Cancer Institute, *Submission G028*, 20 December 2001.

100 Queensland Institute of Medical Research, *Submission G036*, 14 January 2002; Queensland Institute of Medical Research, *Consultation*, Brisbane, 13 December 2001; Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 15 March 2002.

101 Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 15 March 2002.

102 Ibid.

103 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [15.5(b)].

104 Ibid [15.6].

105 Peter MacCallum Cancer Institute, *Submission G028*, 20 December 2001; Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 15 March 2002.

106 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

107 Ibid.

research is approved by an HREC (which might, in some circumstances require researchers to obtain renewed consent from the donors).

15.83 Suggestions for appropriate procedures often focussed on the idea of ‘tick a box’ consent options. That is, prospective research participants could elect from a graduated set of consent options so that, for example, in addition to the participant being asked to consent to participation in a specified research study, the following options might be presented:

My sample:

May be stored for future research related to this study (including DNA studies) but only if my consent is obtained at the time for this future research and the research is approved by a Human Research Ethics Committee (HREC) Yes/No

May be stored for future research related to this study (including DNA studies) without my further consent being required, provided the research is approved by an HREC Yes/No

May be stored for future research unrelated to this study (including DNA studies) but only if my consent is obtained at the time for this future research and the research is approved by an HREC Yes/No

May be stored for future research unrelated to this study (including DNA studies) without my further consent being required and, provided that the research is approved by an HREC, and may be used

- in de-identified form (anonymous) Yes/No
- in potentially identifiable form (coded) Yes/No
- in identifiable form. Yes/No.¹⁰⁸

15.84 Saunders and Komesaroff noted that an approach presently followed by a few HRECs is for individuals to be given the opportunity to choose whether their samples are to be destroyed after the specified experiment has been completed, to be kept and preserved for possible uses approved by an ethics committee, or to be kept and preserved for possible use subject to subsequent renewed individual consent.¹⁰⁹

15.85 This sort of consent process was seen as having significant advantages for research and, at the same time, respecting the wishes of many donors of human tissue. The QIMR stated that in future studies it envisaged using consent forms with tick boxes to indicate consent to restricted or more general use of samples.¹¹⁰

The suggestion that subjects cannot give informed consent to future uses that have not even been invented yet seems to us to ignore the obvious point that the subjects who tick this box are overwhelmingly altruistic in the matter of medical research, and that

108 B Uren, undated pro forma consent form: *Consent for Collection, Storage and Testing of Human Tissue for Research*.

109 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

110 Queensland Institute of Medical Research, *Submission G036*, 14 January 2002.

an [HREC] is perfectly able to make a judgement on whether a future use is reasonable or not without going back to the subject.¹¹¹

15.86 The advantages of obtaining consent for unspecified future research are significant for the organisations conducting genetic research.¹¹² However, some research organisations expressed concern that presenting a wide range of consent options to prospective research participants may present practical difficulties, both at the point of obtaining consent and in the subsequent handling of samples and information.

15.87 In consultations, medical researchers emphasised the need to balance the potential information needs of prospective research participants with consent forms that are readable and understandable to them. For example, the PMCI Tissue Bank Manager stated that:

In my experience of consenting patients, the length and complexity of the consent document can be quite daunting for the prospective donor. There is a fine balance that must be struck between presenting the individual with all the relevant details, thus obtaining truly informed consent, and effectively ‘confounding’ the donor with detail such that they consent on the basis that they trust the Tissue Bank official or they ‘like the look of you’. This is not informed consent.¹¹³

15.88 In relation to research practice, the QIMR, in commenting on the model consent form discussed above, stated that

it would be possible to have a huge number of variations on the consent form and administration or keeping track of who agreed to what would be very difficult.¹¹⁴

15.89 Similarly, the PMCI Tissue Bank stated that the consent options proposed by the model consent form

do not accurately reflect the nature of consent sought for a large prospective tissue collection such as ours, where all donors are asked to consent for broad research purposes. Within a collection of tissues, having mixed levels of consent places many restrictions on the use of that group of tissues. Each sample must be carefully tracked to ensure the use is in compliance with the wishes of the donor. Administration of the collection becomes increasingly complex and the number of samples available within any disease cohort is potentially reduced.¹¹⁵

111 Ibid.

112 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [12.105]–[12.106].

113 Peter MacCallum Cancer Institute Tissue Bank, *Submission G263*, 20 December 2002.

114 Written comments provided at Queensland Institute of Medical Research, *Consultation*, Brisbane, 13 December 2001.

115 Peter MacCallum Cancer Institute Tissue Bank, *Submission G263*, 20 December 2002. This problem was also highlighted in a recent report for the Nuffield Trust, who stated that ‘Informatics experts say that in large computerised systems it is very difficult to lock-in various patient reservations as to purposes as data are split, merged, transformed, transferred, and otherwise manipulated’: W Lowrance, *Learning from Experience: Privacy and the Secondary Use of Data in Health Research* (2002), The Nuffield Trust, London, 20.

15.90 In the course of the Inquiry, a number of useful models for obtaining consent to unspecified future research were highlighted—in particular those used in relation to the operation of human genetic research databases. These models are discussed in Chapter 18.

Reform of specific consent requirements

15.91 The National Statement anticipates that researchers may seek consent to storage of genetic material for unspecified future research on the basis that consent to its use will be sought at the time a specific research project is identified.¹¹⁶

15.92 The Inquiry understands that the National Statement is currently being interpreted, by at least some HRECs, to permit researchers and research organisations to obtain consent for unspecified future research, as approved by an HREC, and without the need for later, specific, consent.

15.93 This interpretation has met with concern in some quarters, as an unjustified erosion of the protection provided by requirements for specific consent to participation in research. Others have expressed the view that procedures to obtain ‘broad and durable’ consent should be facilitated and that the National Statement may not sufficiently mandate such processes.

15.94 At the centre of this debate are differing views about the application of the concept of consent where people cannot be informed in any detail about the nature of the research. It has been suggested that the language of consent should, in some contexts, be augmented by the concept of ‘donation’ or ‘gifting’: it should be recognised that individuals may choose to ‘gift’ samples for research purposes—for example by leaving their body to science or donating blood to the Red Cross.

15.95 In respect of the last example, Sister Regis Mary Dunne observed that donors to the Red Cross are informed that their blood may be used for transfusion, for the extraction of blood products, or for research.

They do not know, nor have the need to know where their blood has gone. However, the documentation is such that individual identity is protected, yet donors could be contacted again if necessary. Such an Australian system already in place could provide a model for the use in research of altruistically donated blood, and tissue surplus to diagnostic purposes. The initial donation could be accepted with the guarantee that all research will be done only after Ethics Committee approval.¹¹⁷

116 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.10(j)]; National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), NHMRC, Canberra, C105. In its general provisions dealing with privacy the National Statement also states that the consent of participants should be obtained for the use of the personal information where the information is to be held on registers for use by researchers in future research projects: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [18.4].

117 M Dunne, *Submission G041*, 17 December 2001.

15.96 As discussed above, some Australian human genetic research databases currently obtain consent to unspecified future research and their ability to continue doing so is important to the effective operation of the databases. The PMCI observed:

The HREC and the tissue bank act together as the gatekeeper of the samples. Patients consent to the use of the material as controlled by the HREC, which, in turn, can respond to changing expectations and increased information over time. Patients may always elect to have their samples removed from the bank at any time.¹¹⁸

15.97 DP 66 noted that there was much support from medical researchers and others for procedures allowing researchers to obtain consent for unspecified future research, subject to future approval by an HREC. In particular, those involved in the development and ongoing operation of Australian human genetic research databases submitted that the use of tissue banks—with a structure of broad patient consent for research on prospectively collected samples and controlled access to samples through HRECs—is the best way to protect the privacy of patients as well as facilitating research.¹¹⁹

15.98 In DP 66, the Inquiry proposed that the National Statement should provide guidelines dealing specifically with obtaining consent to unspecified future research.¹²⁰ This proposal attracted considerable support in submissions.¹²¹ The Department of Health Western Australia stated that:

The proposal that guidelines should be developed is supported, as the issue of unspecified future research raises many complex issues. ... Further, it is recommended that in these guidelines references to 'consent' be replaced with the phrase 'permitting access' which may or may not refer to consent.¹²²

15.99 The HGSA expressed strong support for guidelines allowing researchers to obtain consent to unspecified future research.

The near future will bring an opportunity to undertake 'parallel processing' of vast amounts of genetic information. This will be of little use if completely anonymous but may help establish genotype-phenotype correlations if coded or identified. Consent

118 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

119 Ibid; N Zeps, *Submission G047*, 14 January 2002.

120 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 12–2.

121 Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Medicines Australia, *Submission G302*, 12 February 2003.

122 Department of Health Western Australia, *Submission G271*, 23 December 2002.

forms should, where appropriate, recognise this and, respecting an individual's right to limit or de-limit use of their genetic material, invite a range of consent options...¹²³

15.100 Similar views were expressed by the Australian Biospecimen Network, who emphasised the importance of medical research carried out on archival materials and associated health information.

Given that human tissue taken during routine surgery will almost certainly be used for research and teaching, prospective consent should be considered a routine part of consent to elective surgery. This consent should be broad and open ... delegating responsibility to HRECs to regulate appropriate usage. To consider a system of requiring re-consent would not only be impractical but could also be considered a re-invasion of privacy of an individual since the person would have to be recontacted.¹²⁴

15.101 The OFPC also supported the Inquiry's proposal and observed that individuals' consent for the storage of their tissue for future research should be obtained and that guidelines relating to such consent

could promote good privacy practices by encouraging researchers to devise practical measures to inform, and in appropriate cases to *keep* informed, the altruistic donors of their samples and their information.¹²⁵

15.102 Other submissions expressed reservations about facilitating the obtaining of consent to unspecified future research, as compared to specific consent.¹²⁶ The Law Institute of Victoria stated that

it is 'fundamental to the concept of informed consent' that researchers, researchers, research organisations and HRECs be required to take **reasonable steps** to obtain specific consent at the time a future research project is identified. The fact that research techniques are changing at a rapid rate simply reinforces the argument that specific consent should, where possible, be obtained.¹²⁷

15.103 The Queensland Government cautioned that the development of guidelines dealing specifically with obtaining consent to unspecified future research may raise strong views from the public, particularly in circumstances where it is proposed that specific, informed consent not be required. It suggested that:

An analysis of community views in respect of consent to unspecified future research is needed to ensure confidence in research in Australia is maintained and individuals feel comfortable allowing their samples to be used. Such analysis could be undertaken by the proposed HGCA.¹²⁸

123 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

124 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

125 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003. One model is the Tumour Bank Newsletter published by the Children's Hospital at Westmead Tumour Bank: Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

126 Law Institute of Victoria, *Submission G275*, 19 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002.

127 Law Institute of Victoria, *Submission G275*, 19 December 2002.

128 Queensland Government, *Submission G274*, 18 December 2002.

15.104 The Inquiry has concluded that the National Statement needs to deal more clearly with the issue of consent to future research. The Inquiry recommends that the NHMRC, as part of its review of the National Statement in the 2003–2005 triennium, should amend the National Statement to provide clear guidance about obtaining consent to unspecified future human genetic research.

15.105 In Chapter 18, the Inquiry recommends that the NHMRC should include specific guidance in the National Statement on obtaining consent to unspecified future research in relation to the operation of human genetic research databases. However, the Inquiry does not believe that the use of consent to unspecified future research should be limited to registered human genetic research databases. Other researchers may also wish to use this mechanism, with the approval of their HRECs,¹²⁹ although it may be expected that consent to unspecified future research will most commonly be sought in the context of the operation of standing research databases.

Recommendation 15–4. The NHMRC, as part of its review of the National Statement in the 2003–2005 triennium, should amend the National Statement to provide clear guidance to researchers about obtaining consent to unspecified future human genetic research.

129 The HGSA agreed, emphasising that ‘information from quite small studies may contribute to much larger enterprises’: Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

16. Encouraging Best Practice in Human Genetic Research

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Introduction

16.1 The National Health and Medical Research Council's (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement)¹ offers guidance on, rather than prescription of, ethically sound research design and practice.

16.2 In relation to human genetic research, comprehensive guidance is provided on a range of issues, including the social significance and consequences of genetic research, privacy and confidentiality, consent, waiver of consent and genetic counselling.² The *Human Research Ethics Handbook*³ provides further advice on the provisions of the National Statement for the assistance of Human Research Ethics Committee (HREC) members, researchers and research participants.

16.3 The Inquiry considers that the provisions of the National Statement provide appropriate general ethical guidance and, in most respects, do not require revision. However, in the course of the Inquiry, suggestions were made about a range of desirable practices in the conduct of human genetic research. These included suggestions about the content of consent forms and participant information sheets, de-identification of genetic samples or information, and disclosure of commercial interests.

16.4 In DP 66, the Inquiry proposed that the Australian Health Ethics Committee should develop model research protocols to provide guidance on various aspects of human genetic research and guidelines for preparing consent forms for human genetic

1 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

2 Ibid Ch 16.

3 National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), NHMRC, Canberra.

research.⁴ Submissions indicated widespread support for these proposals.⁵ For example, the Australian Biospecimen Network submitted that model protocols and consent form guidelines ‘are essential to the effective working of HRECs and would greatly aid the writing of and consideration of ethics applications’.⁶ The Centre for Law and Genetics stated that the Inquiry’s proposals would ‘accelerate and achieve more uniform approaches to best practice in research that directly addresses critical issues of participant privacy in human genetic research’.⁷

16.5 Some cautioned against the risk of these becoming inflexible rules and standards in the face of likely rapid change.⁸ Others emphasised the need for education of researchers and HREC members to ensure sound use of the National Statement.⁹

16.6 The Inquiry has concluded that these submissions support the development and publication by the NHMRC of advice and information on good research practice and consent documentation in research involving genetic information. The function of this advice and information would be to augment the National Statement, as revised, to give further guidance to HREC members, researchers and research participants on what the NHMRC considers to be best practice in the conduct of human genetic research. The range of issues to be included in that advice and information is discussed below.

16.7 The Inquiry notes the suggestions for appropriate involvement of professionals¹⁰ and community members¹¹ in the development of the advice and information. The topics on which advice and information is recommended arise not only in research involving genetic information, but also in research involving any personal information. Indeed, genetic research is now a component of much health research.¹²

4 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 13–1, 13–2.

5 Centre for Genetics Education, *Submission G232*, 18 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; J Fleming, *Submission G241*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Genetic Support Council WA, *Submission G243*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003; Medicines Australia, *Submission G302*, 12 February 2003.

6 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

7 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

8 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Queensland Government, *Submission G274*, 18 December 2002.

9 J Fleming, *Submission G241*, 20 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

10 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

11 Genetic Support Council WA, *Submission G243*, 19 December 2002.

12 R Trent, *Consultation*, Sydney, 1 November 2002.

Advice on research protocols

Coding and de-identification of genetic samples and information

16.8 One area of research practice that may be an appropriate subject for guidance relates to the de-identification of genetic samples and information. The de-identification of samples and information is an important mechanism by which privacy is protected in the conduct of research.

From a privacy-protection perspective, there is a very wide distinction between personally identifiable data and truly anonymized data. But in practice the demarcation between these extremes is not sharp. Attending assiduously to where particular data lie on the spectrum between them, and especially to data that are somewhere in the middle, is a crucial protection strategy.¹³

16.9 The National Statement makes distinctions between identified, potentially identifiable (coded or re-identifiable) and de-identified (not re-identifiable, anonymous) personal information or material.¹⁴

16.10 Genetic samples and information used in human genetic research are commonly coded—that is, identifiers such as the individual’s name, date of birth or address are removed and replaced by a code. This information is potentially identifiable.¹⁵

16.11 For the purposes of the National Statement, information or material is ‘de-identified’ only if the process of de-identification is irreversible—for example where the identifiers have been removed permanently or where the data have never been identified. Coded data is not ‘de-identified’ because coding, by its very nature, is intended to be reversible.¹⁶

16.12 Human genetic research often requires information obtained from genetic testing to be linked to health information contained in clinical records, making true de-identification impractical. Eli Lilly Australia Pty Ltd emphasised the importance of a clear definition of de-identification because of its relation to the scope of consent.¹⁷

13 W Lowrance, *Privacy and Health Research: A Report to the US Secretary of Health and Human Services*, Department of Health and Human Services (US), <www.aspe.os.dhhs.gov/dataacncl/phr.htm>, 8 July 2002, 32.

14 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 9. The National Statement states that “[i]t should be recognised that the term “de-identified” is used frequently, in documents other than this Statement, to refer to sets of data from which only names have been removed. Such data may remain “potentially identifiable””: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 9.

15 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 9.

16 Although one of the factors that may be taken into consideration by an HREC in determining whether consent should be waived is the ‘extent to which it is possible to de-identify the sample’—possibly suggesting that de-identification need not be complete anonymisation: *Ibid* [15.8], [16.13].

17 Eli Lilly Australia Pty Ltd, *Submission G247*, 19 December 2002.

16.13 Submissions and consultations emphasised that complete de-identification is often not a practicable solution to privacy concerns about the collection and use of genetic samples or information in research. For example, in relation to tissue banks, the Peter MacCallum Cancer Institute (PMCI) stated that:

It is important that the samples remain 'identified' to the managers of the tissue bank. Retaining identifiers allows the collection of follow-up information, such as survival data. A recommendation that broadly consented material should be de-identified would prevent the ongoing collection of clinical information. For certain diseases, such as breast or prostate cancer, survival outcome may not be known for years. High quality, centrally collected samples with long term outcome information are extremely valuable for testing potential markers of therapeutic response and outcome

...

16.14 There are other reasons why de-identification may not be desirable in the context of human genetic research. Human genetic research may reveal information of potential importance to the future health of participants or their children. The Human Genetics Society of Australasia (HGSA) noted that:

With regard to research uses, de-identification protects privacy but does not address other interests of research subjects such as the possibility of receiving health information that comes out of the research. Obtaining consent for research that uses identified samples/information allows information to go back to participants and may place an obligation on researchers to do so.¹⁹

16.15 The National Statement notes that researchers should consider carefully the consequences of storing genetic material or information in de-identified form for the proposed research, for future research, and for communication of research results to participants.²⁰

16.16 The HGSA stated that HRECs may consider it inappropriate to approve research using de-identified material or information if the research is likely to produce useful health information for the participants.²¹ Some of the dilemmas were illustrated in the HGSA's submission to the Inquiry.

A problem may arise if the research could identify an important family condition, which may have great significance for [the family's] health. In these cases, both the researchers and the HREC should undertake careful consideration as to which is the best way to approach the condition. For example, if a sample from a baby who had died from SIDS were found to have a genetic problem which could occur in either a parent or future sibling, would the duty of the researchers be to inform that family if further harm could be prevented? The family would then be aware that genetic research had been carried out without their consent. If the samples were anonymised initially and a subsequent research study undertaken where consent was sought, harm may have come to the family because of the delay. However, if the hypothesis about

18 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

19 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

20 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.6].

21 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

the genetic problem were wrong, is it appropriate to contact many parents about the test when there may be no benefit and potentially distress them unnecessarily?²²

16.17 A need to raise awareness about the issues involved in de-identification of genetic samples and information was also evidenced amongst groups most affected by genetic research. Representatives of genetic support groups in Western Australia, in a round table discussion facilitated by the Genetic Support Council Western Australia, stated that they

strongly believed that researchers should only work with de-identified samples, and that a code of practice for de-identifying donor samples should be drafted at the federal level to ensure uniformity in practice. However they noted that more information on the de-identification process and what it means would be useful for the groups.²³

16.18 The need for guidance on this issue was emphasised by the Department of Health Western Australia²⁴ and by researchers at Westmead Hospital who pointed out that even where de-identification is preferred it can be difficult to achieve.²⁵

16.19 The Inquiry considers that advice and information on human genetic research protocols should include guidance on the different mechanisms for coding or de-identifying genetic samples and information used in research, and the relative advantages and disadvantages of each approach in different research contexts.

Managing coded samples and information

16.20 Where genetic samples or information are coded, the privacy of the samples and information is protected because they are not immediately identifiable. The level of protection afforded by coding is dependent on who has access to the identifying code and for what reasons.

16.21 There is an emerging international standard for genetic privacy protection, which provides for the use of independent intermediaries to hold the code linking genetic samples or information with the identifiers.²⁶

16.22 The National Statement recognises the desirability, in some circumstances, of intermediaries in the conduct of human genetic research by stating that '[w]here an HREC approves the use of potentially identifiable data that has been coded, the HREC should decide whether an independent person should hold the code'.²⁷

22 Ibid.

23 Genetic Support Council WA, *Submission G112*, 13 March 2002.

24 Department of Health Western Australia, *Submission G271*, 23 December 2002.

25 Children's Hospital at Westmead, *Consultation*, Sydney, 19 November 2002.

26 W Lowrance, *Privacy and Health Research: A Report to the US Secretary of Health and Human Services*, Department of Health and Human Services (US), <www.aspe.os.dhhs.gov/dataacncl/phr.htm>, 8 July 2002, 35–36.

27 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [14.5].

16.23 The Institute of Community Genetics has developed the concept of the ‘gene trustee’—an independent third party acting on behalf of persons submitting genetic samples for testing. This mechanism is described in some detail in Chapter 18.

16.24 The level of protection provided by a gene trustee mechanism may not be practical for all human genetic research projects, particularly those that are small-scale. However, at the very least it may be argued that the persons who have access to the identifying code should be specified in research protocols.

16.25 There was broad agreement in submissions and amongst those consulted by the Inquiry that the use of coded information and independent intermediaries should be encouraged in genetic research, wherever possible.²⁸ However, some submissions noted that the advantages of such an intermediary would need to be balanced against the additional costs of such arrangements.²⁹

16.26 The Inquiry considers that the advice and information on human genetic research protocols should include guidance on the appropriate use of independent intermediaries to hold codes linking genetic samples or information with the identifiers.

Feedback to research participants

16.27 The possibility of providing feedback of health information to research participants has been identified as raising important ethical issues relevant to human genetic research. This issue is closely related to considerations about the coding or de-identification of genetic samples and information. The National Statement provides that:

When research may reveal information of potential importance to the future health of identified or potentially identifiable participant’s future health or the participant’s offspring, the research protocol must provide for consent procedures, counselling, support, test quality and test result confidentiality as would apply if the participant sought such information in a clinical setting. Otherwise such research may only be performed if the genetic material has been de-identified.³⁰

16.28 When consent is being sought from individuals for the collection of genetic material, the National Statement provides that they should be informed

that the researchers will endeavour to provide information about the outcome of the research. Participants should be advised when it is not intended to provide feedback. If relevant, participants should be asked whether they wish to be notified of research results which relate to them as individuals.³¹

28 For example, Institute of Community Genetics, *Submission G156*, 19 April 2002; Australian Privacy Charter Council, *Submission G120*, 18 March 2002.

29 Department of Health Western Australia, *Submission G271*, 23 December 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

30 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.15].

31 *Ibid* [16.10(d)].

16.29 Nick Saunders and Paul Komesaroff referred to the complexities involved in providing individual results to research participants:

Complexities arise as a result of uncertainty about the reproducibility of results, their implications for the individual concerned or for blood relations, their effect on the ability to take out life insurance, and other matters. In recent years it has come to be regarded as a right of participants in research to receive their individual results; it is likely that this assumption will need to change.³²

16.30 Concerns were also expressed by researchers in submissions and consultations about the discharge of their legal or ethical duties to contact research participants about specific test results,³³ and about the differences of opinion among researchers in population studies.³⁴

16.31 It is possible that, in some circumstances, a legal duty to contact a research participant about a genetic test result might be derived from common law principles relating to the tort of negligence and the concept of a duty of care. There is no legal authority regarding the possible imposition of such a duty.³⁵

16.32 Whatever the legal position, it is clear that some researchers are keenly aware of ethical obligations to inform research participants about the health implications of genetic testing conducted on genetic material provided by them. However, there are recognised practical and resource implications involved in providing individualised feedback to participants.

16.33 The Inquiry considers that advice and information on human genetic research protocols should include further guidance on the discharge of these legal or ethical obligations to research participants. As one submission recommended, that advice should identify 'thresholds of impact', which trigger the need to communicate results or outcomes to participants.³⁶

16.34 Such guidance will be useful, especially for research using large databases.³⁷ One possibility would be to provide that, in appropriate circumstances, researchers' ethical duties to contact research participants about test results may be discharged by providing general information to all research participants, for example, by way of a regular newsletter.³⁸ Information provided in this way could alert research participants to the implication of test results found in the course of the research. It could also advise that, if participants are concerned about these implications, they should obtain individual medical advice about whether clinical genetic testing is indicated.

32 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

33 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

34 WA Genetics Council, *Consultation*, Perth, 28 October 2002.

35 The duty to warn is discussed in more detail in relation to health professionals (see Ch 21).

36 Department of Health Western Australia, *Submission G271*, 23 December 2002.

37 *Ibid.*

38 One model is the Tumour Bank Newsletter published by the Children's Hospital at Westmead Tumour Bank: Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

16.35 The Department of Health Western Australia noted two broader considerations that need to be recognised in the advice and information to be developed by the NHMRC. First,

the level of protection provided by such technical matters may still be compromised by failure in the integrity of the researchers and this is an important matter to be addressed by HRECs, institutions and the researchers themselves.³⁹

Second, the broader function or consequence of such regular advice needs to be recognised because

there needs to be the encouragement of a research culture and ethos which has respect for the outcomes and potential impact of the research on human subjects.⁴⁰

Disclosure of commercial arrangements

16.36 At least in terms of overall funding, it is clear that private sector involvement in health and medical research is significant (see Chapter 11). As a new and burgeoning area of medical research, private sector involvement in human genetic research can be expected to grow. It is express government policy to increase private funding of research and to improve Australia's record in the commercialisation of the outcomes of research.⁴¹

16.37 In consultations the Inquiry was told that it is increasingly common for universities and publicly funded research institutes to 'spin off' private biotechnology firms, including those engaged in aspects of human genetic research.

16.38 Concerns were often expressed in consultations, and in public meetings in particular, about the involvement of commercial enterprises in human genetic research. While people who volunteer as participants in medical research generally have altruistic motives, it was seen as important that the outcomes of research enhance the public good.

16.39 While the Inquiry recognises that the benefits of genetic medicine are unlikely to be achieved without substantial private sector research investment, the commercial background to research projects is an important factor for many prospective research participants. In the absence of Australian data, empirical evidence about public attitudes to research in the United Kingdom may have some relevance in this context. A survey conducted by the Human Genetics Commission in the United Kingdom has found that levels of public trust in the responsible use of human genetic information vary markedly, depending on the nature of the individuals or bodies

39 Department of Health Western Australia, *Submission G271*, 23 December 2002.

40 *Ibid.*

41 Health and Medical Research Strategic Review, *The Virtuous Cycle, Working Together for Health and Medical Research* (1999), Commonwealth of Australia, Canberra, Ch 8, App 1 (Government Response to the Recommendations and Actions in the Report of the National Health and Medical Research Strategic Review—NHMRC to Action).

holding it. In particular, respondents trusted academic scientists more than health and pharmaceutical companies.⁴²

16.40 Extensive public consultation has been conducted in relation to the UK BioBank initiative.⁴³ One of the reported outcomes of this research, conducted through interactive workshop sessions,⁴⁴ was that:

The first reactions were that commercial companies should not have access [to genetic data], although some participants were quick to realise that the results [of research] would be published and therefore widely accessible to anyone. Further debate brought the realisation that if medicines are going to be developed, pharmaceutical companies must have access.⁴⁵

16.41 Another report on qualitative research connected with the development of the BioBank UK initiative concluded that the fact that sample collection would be a 'publicly funded initiative and not set up as a profit-making exercise was reassuring and important in communicating its credibility'.⁴⁶ The report indicated that

[t]here are likely to be questions from the general public and in the media about commercial access to, and use of, the samples and information. Assuming samples are donated freely by donors, there needs to be careful explanation of the financial implications of this.⁴⁷

16.42 Australian academics have expressed the view that, from an ethical perspective, 'the potential for commercial exploitation' of genetic samples and other biological materials is a very relevant consideration when individuals decide whether to consent to participate in research, given that participation is typically altruistic in nature.⁴⁸ Similar views were expressed in some submissions.⁴⁹

16.43 It may be argued that there is a clear need for open and transparent disclosure, to prospective research participants, of the potential commercialisation of research outcomes and the commercial interests of the researchers involved. It has been

42 MORI Social Research, *Public Attitudes to Human Genetic Information: People's Panel Quantitative Study Conducted for the Human Genetics Commission*, <www.hgc.gov.uk/business_publications_morigeneticattitudes.pdf>, 19 February 2003, 41. Respondents were asked the following question (n=1,038): Q68 Please tell me which, if any, you trust to use the human genetic information held on medical databases responsibly? The responses included: An expert government scientific advisory committee (39%); Academic scientists (38%); Health and pharmaceutical companies (20%); Government (13%).

43 UK BioBank is discussed in more detail in Ch 18.

44 People Science and Policy Ltd, *BioBank UK: A Question of Trust: A Consultation Exploring and Addressing Questions of Public Trust Report prepared for the Medical Research Council and the Wellcome Trust March 2002*, People Science and Policy Ltd (2002), People Science and Policy Ltd, London, 1.

45 Ibid, 19.

46 CR Dawson, *Public Perceptions of the Collection of Human Biological Samples*, MRC and Wellcome Trust, <www.mrc.ac.uk/pdf-biobank_public_perceptions.pdf>, 20 February 2003.

47 Ibid, 17.

48 D Nicol, M Otlowski and D Chalmers, 'Consent, Commercialisation and Benefit-Sharing' (2001) 9(1) *Journal of Law and Medicine* 80, 93.

49 E Mussawir, *Submission G235*, 17 December 2002; H Saleh and others, *Submission G218*, 3 December 2002.

suggested that such disclosure may protect the interests of both prospective research participants and researchers themselves:

In order to avoid feelings of exploitation, and possibly even deception, it is of crucial importance that they be given the opportunity to consent to participation in the knowledge that there is a possibility of commercial gain being made from their donated biological material. To do otherwise risks damaging the perception of research and may thereby reduce the willingness of people in the community to participate.⁵⁰

16.44 The National Statement contains a number of provisions relating to the disclosure of funding and financial interests. However, there is no general requirement to disclose this information, or other information about the actual or anticipated commercial arrangements connected with the research, to research participants.

16.45 The National Statement provides that a researcher must disclose to the HREC reviewing the research proposal the amount and sources or potential sources of funding for the research and must declare any affiliation or financial interest. The HREC must consider the extent to which the researcher should disclose information about funding sources to research participants.⁵¹ The HREC may decide that no such disclosure is justified.

16.46 The disclosure requirements in relation to clinical trials are somewhat more rigorous. A researcher must inform an HREC 'of any business or other similar association which may exist between a researcher and the supplier of a drug or surgical or other device to be used in the trial'.⁵² Further, an HREC must examine those aspects of the budgets of clinical trials that raise ethical issues.⁵³

16.47 The Australian Academy of Sciences submitted that 'all applications to HRECs should be required to include details of any commercial support obtained or envisaged'.⁵⁴ It is not clear to what extent this suggestion goes further than the existing provisions of the National Statement requiring disclosure to HRECs. What seems more important is the question of disclosure to research participants.

16.48 The Centre for Law and Genetics suggested that the provisions of the National Statement dealing with disclosure of commercial arrangements to research participants should be tightened.

As a general principle research participants have an ethical right to be informed of all aspects of the research project including any commercial arrangements.⁵⁵

50 D Nicol, M Otlowski and D Chalmers, 'Consent, Commercialisation and Benefit-Sharing' (2001) 9(1) *Journal of Law and Medicine* 80, 93.

51 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.21].

52 *Ibid* [12.5].

53 *Ibid* [12.6].

54 Australian Academy of Science, *Submission G097*, 21 January 2002.

55 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

16.49 Other submissions stressed the importance of disclosure to research participants, as well as to HRECs. The Australian Red Cross Ethics Committee stated that

there should be transparent disclosure to research participants of the potential commercialisation of research outcomes, as well as any conflicts of interest. ... our Committee requires disclosure of commercial arrangements for funding or product development. No researcher has ever raised any objection. In fact most researchers provide a full explanation of the commercial aspects of the research. We should also point out that our standard-model information form includes specific questions on commercialisation. There has been a general acceptance of the disclosure principle.⁵⁶

16.50 There is presently no ethical guidance on the desirable extent of disclosure of commercial arrangements. The Inquiry considers that the NHMRC should develop information and advice on the disclosure by researchers, to research participants, of information about actual or anticipated commercial arrangements connected with human genetic research proposals.

Recommendation 16–1. The National Health and Medical Research Council (NHMRC) should develop information and advice for the preparation of human genetic research protocols, including examples and practical guidance on:

- (a) the mechanisms for coding or de-identifying genetic samples and information used in research, and the relative advantages and disadvantages of each approach in different research contexts;
- (b) the use of independent intermediaries, in appropriate cases, to hold codes linking genetic samples or information with the identifiers;
- (c) the discharge of legal and ethical obligations to inform research participants about the health implications of testing of genetic samples; and
- (d) disclosure by researchers to research participants of information about actual or anticipated commercial arrangements connected with human genetic research proposals.

Advice on consent forms

16.51 The Inquiry considers that information and advice for human genetic research should include guidance on the drafting of consent forms. Guidance is required on the following matters:

56 Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003.

- The use of a graduated set of consent options, from consent to specific research, to related research, or to unspecified future research (as approved by an HREC). Issues concerning consent to the use of genetic samples and information in human genetic research—including issues relating to obtaining consent to as yet unspecified future research are discussed in Chapter 15 above.⁵⁷
- Disclosure by researchers, to research participants, of information about actual or anticipated commercial arrangements connected with human genetic research proposals, as discussed above.
- Clarification of ownership or property interests in any genetic samples, or the information derived from such samples. Issues relating to property in genetic samples are discussed in Chapter 20.

16.52 There was wide support in the submissions for the development of guidelines for preparing consent forms for human genetic research.⁵⁸ Some reservations were expressed about the uncertainty of the ownership issue⁵⁹ and others identified the need for constant communication between governing bodies and researchers⁶⁰ and the need for flexibility.⁶¹

16.53 There is a range of other matters that might usefully be included in guidance on the drafting of consent forms. These include:

- Model statements about how the privacy of research participants is to be protected—that is, through coding or de-identification of genetic samples or information. Saunders and Komesaroff submitted that the implications of decisions about coding or de-identification should be more fully presented to prospective research participants, when their consent is being sought.⁶²
- Model statements about whether, and if so how, consent to participation may be withdrawn. The National Statement provides that a participant must be free at any time to withdraw consent to further involvement in research and the consequences of withdrawal must be explained when obtaining consent.⁶³ Saunders and Komesaroff stated that, in human genetic research, ‘withdrawal may affect not only the research participant but also her or his blood relations’. They also noted that it is often the case that ‘to avoid critical biases in statistical

57 In particular, the documentation developed by the WA Research Tissue Network and the Children’s Hospital at Westmead, Tumour Bank, provide useful models for obtaining consent to the use of stored tissue.

58 Centre for Law and Genetics, *Submission G255*, 21 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; J Guamieri, *Submission G210*, 28 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

59 Queensland Government, *Submission G274*, 18 December 2002.

60 Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

61 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

62 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

63 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [1.12].

analysis withdrawal of a subject enrolled in a research project does not lead to retrospective withdrawal of the data relating to him or her'.⁶⁴ The Department of Health Western Australia recommended that guidance is needed for the rare situations in which the design of the research project precludes the withdrawal of consent.⁶⁵ These sorts of matters may need to be made clearer to prospective research participants on consent forms.

- Who should consent to the access to tumour blocks where the donor of the tissue has died and where access is sought for use in research of relevance to other family members.

Recommendation 16–2. The NHMRC should develop information and advice for preparing consent forms for human genetic research, including examples and practical guidance on such matters as:

- (a) graduated consent options;
- (b) disclosure by researchers about actual or anticipated commercial arrangements;
- (c) ownership or property interests in genetic samples or information;
- (d) methods of protecting the privacy interests of participants; and
- (e) withdrawal of consent by participants.

64 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

65 Department of Health Western Australia, *Submission G271*, 23 December 2002.

17. Strengthening Review by HRECs

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Introduction

17.1 In DP 66 the Inquiry discussed possible reforms to the operation of Human Research Ethics Committees (HRECs).¹ Some of these reforms would require changes to the National Health and Medical Research Council (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement).² Others would be matters for the Australian Health Ethics Committee (AHEC), or research institutions themselves, to examine further. This chapter discusses the options for strengthening ethical review by HRECs and the responses that the Inquiry received to the issues raised in DP 66.

17.2 The Inquiry received a wide range of comment on the adequacy of the existing system of research ethics review—from those who felt that despite resource constraints the current system works well, to those who were critical of it. A number of

1 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney Ch 14.

2 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

submissions suggested possible reforms aimed at strengthening the current system of ethical review of human genetic research proposals and the role of HRECs within that system. While the Inquiry has focused on human genetic research, changes to the ethics review process may have implications for all human research reviewed by HRECs.

17.3 The Inquiry has concluded that there is a need to improve consistency, efficiency, transparency and accountability in HREC review of human genetic research. The Inquiry recommends that the NHMRC initiate a systematic quality improvement program that addresses the range of concerns discussed in this chapter.

The National Statement, AHEC and HRECs

17.4 The National Statement is the foundation of the Australian system of ethical review of research involving humans. As discussed in Chapter 13, the primary purpose of that Statement is the protection of the welfare and rights of participants in research,³ but an important secondary purpose is to ‘facilitate research that is or will be of benefit to the researcher’s community or to humankind’.⁴

17.5 AHEC advises the NHMRC on ethical issues relating to health and develops guidelines for the conduct of medical research involving humans. AHEC therefore has an ongoing role in developing proposals for improving the system of ethical review. During the 2000–2002 triennium, a major element of the strategic plans of AHEC and the NHMRC was to strengthen the level of support provided to HRECs, by way of training workshops, access to advice, and the production of the *Human Research Ethics Handbook*.⁵ The National Statement is scheduled for formal review by AHEC during the 2003–2005 triennium, consistently with NHMRC policy of revising guidelines that have been in place for five years.

17.6 This chapter focuses on the role, function, responsibility and accountability of HRECs, but there are other components of the system of ethical review. These should not be discounted. The National Statement makes it clear that the design and conduct of human research, the integrity of researchers, and their conformity to ethical values and principles are also central to achieving the purposes of the National Statement.

Human Research Ethics Committees

17.7 The National Statement states that the functions of HRECs are to review and, where appropriate, approve all proposals for research involving humans, and to monitor the conduct of that approved research.

3 Ibid, Preamble.

4 Ibid, Preamble.

5 National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), NHMRC, Canberra.

Institutional status

17.8 The status, functions, responsibilities and accountability of HRECs are set out in the National Statement. The National Statement provides that research projects involving human participants must be reviewed and approved by an HREC. HRECs are established by and advise an institution or organisation regarding ethical approval for research projects.⁶

17.9 Institutions or organisations in which such research is undertaken should establish, adequately resource and maintain an HREC, or have research projects reviewed and monitored by an HREC established by another institution.⁷ When establishing an HREC, an institution must set out its terms of reference, scope of responsibilities, accountability and reporting mechanisms.⁸ Institutions must accept legal responsibility for decisions and advice received from the HREC and agree to indemnify its members.⁹

17.10 Institutions with HRECs are required to establish mechanisms for receiving and promptly handling complaints or concerns about the conduct of approved research projects. HRECs must nominate a person to whom complaints may be made and make their identity known to the research participants. Institutions are also required to establish procedures for receiving and promptly handling complaints from researchers about review of research proposals by an HREC.¹⁰

17.11 At present there are 216 HRECs in Australia registered with AHEC, of which 102 are in hospitals, 47 in universities, 35 in government and 32 in other institutions.¹¹ The workload of HRECs varies significantly. Some review in excess of 300 proposals each year, while others deal with far fewer.

Functions and responsibilities

17.12 The primary function of an HREC is to protect the welfare and rights of participants in research.¹² All research projects involving humans must be reviewed by an HREC and must not be undertaken or funded unless and until approval has been granted. HRECs are required to maintain a record of all proposed research projects including specified information. HRECs must also keep a copy of research protocols in the form in which they were approved.

6 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 15.

7 Ibid [2.1].

8 Ibid [2.2].

9 Ibid [2.3].

10 Ibid [2.39]–[2.43].

11 As at 14 February 2003: Data provided by Health Ethics Section, NHMRC. However, it is acknowledged that there are likely to be ethics committees in existence within the community not-for-profit or private sectors that are not registered with the NHMRC.

12 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.5].

17.13 Institutions and HRECs each have responsibilities to ensure that there is appropriate monitoring of the conduct of projects until their completion, at least by an annual report.¹³ The frequency and type of monitoring should reflect the degree of risk to participants, and may utilise existing institutional mechanisms for this purpose.¹⁴

Membership and procedures

17.14 The National Statement specifies a minimum membership for HRECs and requires that the membership has a balance of suitable expertise. The current system of ethics review aims to combine peer review and community involvement. The National Statement also addresses HREC meetings,¹⁵ managing conflicts of interest,¹⁶ documentation and recording of decisions,¹⁷ and expedited review of minimal risk projects.¹⁸

Accountability

17.15 HRECs are accountable to their institutions through reporting and other processes. These may vary among institutions. In addition, the National Statement establishes further accountability processes that relate to the national system of ethical review of research involving humans.

Structure of ethics review

17.16 The National Statement requires that research proposals be reviewed by an HREC. However, organisations may apply additional levels of review or delegate functions to subcommittees.¹⁹

17.17 A separate committee may review the scientific aspects of the project. For example, the Peter MacCallum Cancer Institute utilises a two-stage review process. Research proposals are first reviewed by a biomedical committee to assess the scientific and research value of the proposals, and then by an HREC. One advantage of this approach is that the composition of the HREC can more easily accommodate non-expert and non-institutional members.²⁰

13 Ibid [2.20].

14 Ibid [2.33]–[2.34].

15 Ibid [2.13]–[2.18].

16 Ibid [2.19]–[2.21], [2.23].

17 Ibid [2.24], [2.30]–[2.32].

18 Ibid [2.27]–[2.29].

19 The *Human Research Ethics Handbook* notes that appropriate task-based sub-committees might include scientific review sub-committees, committees for conducting expedited review of low-risk research, discipline-based subcommittees for review of research related to degree qualifications, other special-purpose subcommittees and policy subcommittees: National Health and Medical Research Council, *Human Research Ethics Handbook* (2002), NHMRC, Canberra, [E77].

20 Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 15 March 2002.

17.18 Two-stage review may also be used to review issues arising out of human genetic research involving collectivities, such as research involving Aboriginal and Torres Strait Islander peoples. A joint HREC of the Menzies School of Health Research and the Royal Darwin Hospital convenes an Aboriginal Sub-committee that reviews such research proposals. The sub-committee (assisted by a scientific adviser) reviews proposals; consults with leaders of the relevant Aboriginal communities; and may veto proposals where ethical concerns remain.²¹

17.19 Some submissions suggested that human genetic research should be subject to a more centralised system of ethics review.²² For example, Privacy NSW suggested the establishment of ‘an independent, national ethics review committee with substantial lay and community representation’ and ‘a single, independent and transparent complaints mechanism to deal with decisions made by any HREC’.²³

17.20 A system of centralised review would represent a significant change to the present institutionally based system. In Australia, as in the United States, Canada, New Zealand and the United Kingdom, ethics review committees are based in an institution—usually a research institution within which the research is conducted—or are established within a regional authority, such as an area health service.²⁴

17.21 Complete centralisation of the ethics review process would probably be impractical. The volume of research proposals requiring review is likely to exceed the capacity of a single national body, or series of regional review bodies. The resource implications of establishing a national committee would be significant.

17.22 A centralised system of ethics review would need to include mechanisms to respect the legitimate interests and knowledge of institutions concerning the suitability of their facilities and staff to conduct the proposed research. A centralised system of review also risks introducing a narrow ethical range of perspectives, which are less responsive to community and institutional commitments and change. An institutionally based ethical review system accommodates HREC decisions that may differ in how they apply the principles and values set out in the National Statement.

17.23 One precedent for a national centralised level of review is the Gene and Related Therapies Research Advisory Panel (GTRAP). GTRAP is a subcommittee of the Research Committee of the NHMRC, and was established to provide the NHMRC and HRECs with advice on human gene therapy trials, and to assist researchers in the establishment of best practice standards.²⁵ Although GTRAP is primarily an advisory body, it plays a role in the HREC approval process for human gene therapy trials.

21 Menzies School of Health Research, *Consultation*, Darwin, 18 March 2002.

22 L Martinovic, *Submission G006*, 30 May 2001; Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

23 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

24 P McNeill, ‘International Trends in Research Regulation: Science as Negotiation’ in *Research Ethics* (1999) Elsevier Science, Oxford, 243.

25 National Health and Medical Research Council, *Gene and Related Therapies Research Advisory Panel (GTRAP)*, <www.health.gov.au/nhmrc/research/gtrap/about.htm>, 23 July 2002.

AHEC has stated that HRECs should not approve research that involves gene therapy without the prior approval of both the Institutional Biosafety Committee and GTRAP.²⁶

17.24 This review process was thought necessary because gene therapy involved ‘novel approaches to treatment, the short or long term potential for harm was still undetermined, and it would be difficult for HRECs to gain expertise as the number of research studies would be small’.²⁷ It needs to be emphasised that this approach was specifically based on the possibility that human gene therapy might accidentally affect the germ cell line and that it was therefore unreasonable for a single HREC to be expected to make ethical judgments on such studies without access to the best available scientific advice.

17.25 The Inquiry does not believe that there are cogent reasons to establish a national ethics committee to review human genetic research and not other research involving humans. While human genetic research involves a particular constellation of ethical issues, the ethical hazards are no greater than some other categories of research—such as clinical trials or research involving vulnerable persons. Similarly, while some genetic research may involve the need for access to specialised scientific advice, much will not.

17.26 The Inquiry considers that there are a number of ways to strengthen review of human genetic research by HRECs without establishing a centralised system. As discussed below, these include revised procedures, improved resources, and better education and training. Nevertheless, the NHMRC, as part of the quality improvement program recommended below, should consider whether there is a need for greater consolidation of ethical review, by region or subject-matter.

Quality of review

Membership of HRECs

17.27 The minimum membership of an HREC must comprise:

- a chairperson;
- at least two members who are lay people, one man and one woman who have no affiliation with the institution or organisation, are not currently involved in medical, scientific or legal work, and who are preferably from the community in which the institution is located;
- at least one member with knowledge of and current experience in the areas of research that are regularly considered by the HREC;

26 Australian Health Ethics Committee, *Information Sheet for HREC's #2* (2001) Australian Health Ethics Committee, Canberra.

27 Ibid.

- at least one member with knowledge of and current experience in the professional care, counselling or treatment of people;
- at least one member who is a minister of religion or a person who performs a similar role in a community such as an Aboriginal elder; and
- at least one member who is a lawyer.²⁸

17.28 Institutions must ensure that the membership of their HRECs will equip them to address all relevant considerations arising from the categories of research likely to be submitted to them.²⁹ Each HREC must ensure that it is sufficiently informed on all aspects of each research protocol, including its scientific and statistical validity, to decide whether the protocol is acceptable on ethical grounds and conforms to the National Statement. Institutional appointment of additional members with relevant expertise may be necessary, but in making additional appointments institutions should maintain the diversity of categories of members and the relative proportion between institutional and non-institutional members.³⁰

17.29 Institutions may recruit members in such manner and shall appoint them for such terms and on such conditions as they determine—although members are appointed for their expertise and not as representatives of any professional, political or social group. Members should receive a formal notice of appointment together with an assurance that the institution will provide legal protection in the event of claims arising from bona fide conduct of committee duties.³¹

17.30 There need be only two members of the HREC who have no affiliation with the institution. An informal survey by AHEC of HRECs from the hospital and university sectors conducted in 2002 showed that on average three or four of the six core membership categories were filled by persons who had no affiliation with the institution, although this varied quite widely.

17.31 In submissions and consultations, concerns were expressed about the mix of institutional and non-institutional members on HRECs and the effects of this composition on the independence of HRECs. A particular concern was that some community members may be disempowered in their contributions to the review process by the presence of members with specialist expertise, and by a preponderance of institutional members, who may be employees of the research organisation whose research proposals are being reviewed.

17.32 In expressing concern about the perceived inadequacies in the audit of HRECs' use of guidelines issued under the *Privacy Act 1988 (Cth)* (*Privacy Act*), Privacy NSW stated that:

28 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.6].

29 Ibid [2.7].

30 Ibid [2.8]–[2.9].

31 Ibid [2.10]–[2.12].

This lack of transparency is a significant concern in light of the composition of HRECs, which may include interests that are not independent of the interests attached to the research itself. Inevitably, implementation of the Guidelines varies depending upon the personal, institutional and commercial context in which an HREC operates.³²

17.33 The New South Wales Health Department made similar points, relating the balance of membership to the transparency of HREC processes.

The structure and transparency of HRECs could be enhanced by including more community representation than is currently required by the National Statement. The current preponderance of 'institutional' members may lead to lay and community members feeling disempowered in their contributions to the review process.³³

17.34 An alternative view is that institutions are entitled to have their own interests and values maintained by the appointment of persons who hold the values of the institution strongly, including staff members, and that such persons also understand that the primary purpose of the HREC is to protect the interests of human participants. It is not in the interests of institutions to be seen to support unethical research.

17.35 There is an international trend towards stronger community representation on ethics review committees. This consists of a trend towards the appointment of a greater number of community members; the election of chairpersons from among the community members; the selection of people 'of standing' as community members because they may be better able to withstand pressure from other committee members; and the appointment of community members as representatives of research participants or particular groups of participants.³⁴

Advice for HRECs on review of human genetic research proposals

17.36 The Inquiry received submissions on the need for HREC members to have sufficient expertise in genetics. The Familial Cancer Centre of the Royal Melbourne Hospital recognised the ubiquity of genetic issues in health research and recommended that:

Since so many research projects now relate to human genetic research, HRECs should include at least one member who has genetic expertise and understands the ethical and social issues relating to such research.³⁵

17.37 This suggestion was supported by other submissions,³⁶ and is consistent with the requirement of the National Statement that the membership of HRECs be equipped

32 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

33 NSW Health Department, *Submission G303*, 13 January 2003.

34 P McNeill, 'International Trends in Research Regulation: Science as Negotiation' in *Research Ethics* (1999) Elsevier Science, Oxford, 243, 254.

35 Familial Cancer Centre — Royal Melbourne Hospital, *Submission G249*, 20 December 2002.

36 New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002.

to address issues arising from the categories of research likely to be submitted to them.³⁷

17.38 Some submissions suggested that HRECs need further sources of advice on ethical considerations related to human genetic research. The Australian Academy of Science stated that:

there are many HRECs that are facing new challenges in areas such as genetics. Each HREC has the responsibility to consider each proposal on its merits, but we recommend that there should be a system in place where HRECs can ask for advice from a central body. If a Standing Advisory Group on Human Genetics is established, either it, or the Australian Health Ethics Committee, or the two together, could fill this role.³⁸

17.39 As discussed in Chapter 15, the Inquiry conducted a survey of all HRECs to determine the number of research proposals that involved human genetic research and were considered by HRECs during 2002. The Inquiry received 82 completed questionnaires (a response rate of 38%). Twenty-six HRECs had considered 129 proposals involving human genetic research in the reporting period.

17.40 Although the number human genetic research proposals submitted for review is likely to increase, the Inquiry does not recommend the establishment of a specialist advice service for HRECs. However, as part of the quality improvement program for HRECs recommended below, the needs of HRECs for expertise, and the obligations of institutions and committees to be satisfied that they have adequate expertise, should be emphasised.

Monitoring of human genetic research by HRECs

17.41 Research organisations and HRECs have responsibilities to ensure that there is appropriate monitoring of approved research projects. The National Statement requires that regular reporting, at least annually, is a minimum monitoring requirement. Reports should address progress of the research, maintenance and security of records, compliance with the approved protocol and with any conditions of approval.³⁹ Additional monitoring mechanisms may be employed by the HREC.⁴⁰ HRECs are directed to require that researchers report immediately anything that might warrant review of the ethical approval of the project, including serious or unexpected adverse effects on participants, proposed changes to the protocol and unforeseen events that might affect the continued ethical acceptability of the project.⁴¹ HRECs are to impose, as a condition of approval, the requirement that researchers inform the HREC, with reasons, if the research is discontinued before the expected date of completion.⁴²

37 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.7].

38 Australian Academy of Science, *Submission G097*, 21 January 2002.

39 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.35].

40 *Ibid* [2.36].

41 *Ibid* [2.37].

42 *Ibid* [2.38].

17.42 Where an HREC is satisfied that a research project is not, or cannot be, conducted in accordance with the approved protocol and that, as a result, the welfare and rights of participants are not, or will not, be protected, it may withdraw ethical approval, inform the researcher and institution of such withdrawal, and recommend to the institution that the research be discontinued or suspended. A researcher is required to discontinue research if ethical approval is withdrawn.⁴³

17.43 The Inquiry has received differing views on the adequacy of the current monitoring practices, which vary widely in different types of research, from clinical trials to social science research. Some individuals and organisations were critical of the existing arrangements for monitoring of research by HRECs. However, the resource implications of implementing more thorough monitoring were also widely recognised.

17.44 The New South Wales Genetics Service Advisory Committee stated that there should be uniform policies and guidelines on the reporting and monitoring requirements of HRECs.⁴⁴ Privacy NSW submitted that monitoring should be conducted by HRECs throughout the research period and that the current minimum requirement of annual inspection was inadequate.⁴⁵ Professor Nick Saunders and Associate Professor Paul Komesaroff stated that, in general, HRECs do not effectively monitor approved research.

It is likely that to achieve adequate monitoring a fundamental change in approach will be needed, including possibly routine questioning of research participants. This is an important question that will in due course require detailed attention.⁴⁶

17.45 The Queensland University of Technology stated that it currently exceeds the requirements in the National Statement by conducting 'limited random audit of active ethical clearances'. The University noted that this approach should be considered as an additional minimum monitoring requirement. The University is also in the early stages of considering compliance audits to capture research not submitted for review.⁴⁷ Other suggested additions to the monitoring process included mechanisms to check on consent processes⁴⁸ and the development of criteria for identifying genetic research that involves high risks and requires more intensive monitoring.⁴⁹

17.46 The NHMRC Research Committee emphasised the resource constraints on monitoring.

Monitoring of research by HRECs presently requires annual reports and the HREC can undertake further monitoring if this were felt to be essential. Again, the administrative load currently required of a HREC would make the requirement for

43 Ibid [2.44]–[2.45].

44 New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002.

45 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

46 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

47 Queensland University of Technology, *Submission G109*, 14 March 2002.

48 H Saleh and others, *Submission G218*, 3 December 2002.

49 Department of Health Western Australia, *Submission G271*, 23 December 2002.

more active or frequent monitoring difficult to implement unless some other part of the regulatory process could be relaxed or simplified.⁵⁰

17.47 Other submissions highlighted the importance of adequate resources.

The current minimum monitoring requirements by HRECs are adequate. The existing provisions allow HRECs to use discretion when monitoring human research proposals, including human genetic research. The limiting factor for adequate monitoring by HRECs is their lack of adequate resources.⁵¹

17.48 The existing provisions of the National Statement relating to monitoring of research are flexible and allow institutions and HRECs to monitor the conduct of human genetic research to the extent that they consider appropriate, or practicable. However, given the importance of effective monitoring procedures and the concerns expressed to the Inquiry, monitoring procedures should be reviewed by the NHMRC as part of the quality improvement program recommended below.

Needs of HRECs, HREC members and researchers

Resources

17.49 The resourcing of HRECs was examined by the 1996 *Report of the Review of the Role and Functioning of Institutional Ethics Committees* (the 1996 IEC Report).⁵² Concerns about the overall resources available to HRECs, not only for monitoring of research, were expressed in submissions and consultations. For example, the Office of the Federal Privacy Commissioner (OFPC) argued that while the current structures for ethics review are ‘fundamentally sound’ there needs to be sufficient resources made available to support members of HRECs.⁵³

The current HRECs structure currently depends heavily on the good will of respected members of Australian society, who generously give their already limited time to this very important process. HRECs are finding it increasingly difficult to meet additional obligations and processes as they are introduced.⁵⁴

17.50 With particular reference to privacy compliance, the OFPC stated that:

From a privacy perspective, compliance with HRECs requirements by researchers should be at a premium. If the growing research challenges are to be met in the interests of the community and participants, HRECs will need every assistance by way of resourcing and administrative support and in the provision of guidance on such matters as applying the Guidelines under [the *Privacy Act*].⁵⁵

50 National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002.

51 NSW Health Department, *Submission G303*, 13 January 2003. See also J Fleming, *Submission G241*, 20 December 2002.

52 D Chalmers, *Report of the Review of the Role and Functioning of Institutional Ethics Committees: Report to the Minister for Health and Family Services* (1996), Commonwealth of Australia, Canberra.

53 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

54 Ibid.

55 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

17.51 The need for resources to enable HRECs to carry out their functions was repeated in many submissions.⁵⁶ Submissions linked the inadequacy of resources to all functions of HRECs. The Human Genetics Society of Australasia (HGSA) stated that the role of HRECs is ‘constrained by resources and lack of expertise’.

Additional resources are needed to allow HRECs to carry out their assessment of research submissions and to meet their reporting and monitoring obligations. Members of HRECs, most of whom are unpaid volunteers, should be provided with general training and any specific training that relates to the type of research carried out in their institution. However, it is essential that the additional resources do not come from the limited funding available in Australia for research.⁵⁷

17.52 The Queensland University of Technology submitted that there is a need for greater resourcing for the exchange of information between HRECs, for the promotion of excellence in the ethics review, and for the training of HREC members and researchers.⁵⁸ The Australian Biospecimen Network commented on the need for better resources to streamline administrative processes.

Resources should be made for improved secretarial support and for online facilities for expedited completion of ethics applications. Such online facilities would provide links to all relevant sections of the National Statement, recommendations on how to ensure compliance including model consent forms. Such a facility would solve many of the issues related to the time needed to fill in an ethics application by an investigator, the quality of such submissions and therefore the time taken to have it reviewed and its reciprocal acceptance by other HRECs.⁵⁹

17.53 Although there was widespread agreement that HRECs needed more resources, there was no unified opinion as to the source of those resources. For example, while the HGSA opposed the use of research grants for these purposes, the OFPC stated that:

specific funding allocations should be made in research grants to enable researchers to properly inform HRECs of all pertinent issues and to comply with the relevant protective guidelines, such as the Section 95 and 95A Guidelines under the [*Privacy*] Act. Those research grants should also include specific funding allocations for the involvement of HRECs in the research project.⁶⁰

17.54 The cost to researchers of complying with ethics review and other requirements is a related issue. Professor Nick Martin, Head of the Queensland Institute of Medical Research stated:

56 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; J Fleming, *Submission G241*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Queensland Institute of Medical Research, *Submission G190*, 26 November 2002; NSW Health Department, *Submission G303*, 13 January 2003.

57 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

58 Queensland University of Technology, *Submission G109*, 14 March 2002.

59 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

60 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

We receive no extra budget to cope with compliance costs, and inevitably this means less research gets done. For example, simply to comply with ethical requirements for my own projects requires 1.5 FTE positions. In the US, the National Institutes of Health have now recognised this and now add 8% for compliance costs to all successful grants. I hope that your report will recommend a similar measure here. The government and community have to recognise that they simply cannot have this degree of regulation without paying for it. Furthermore, the amount of time now required to look after compliance issues greatly decreases job satisfaction for scientists and I know many who are now actively avoiding tackling important clinical problems for this reason. I don't believe this is in the community's best interest.⁶¹

17.55 The Inquiry recognises that, under the National Statement, it is the responsibility of institutions to resource HRECs. However, submissions and consultations indicated that this responsibility is not always fulfilled. The Inquiry recommends that, as part of the process of strengthening HREC review of human genetic research, each relevant institution and organisation should provide adequate resources to enable its HREC to fulfil its institutional responsibilities and achieve the standards required by the quality improvement program recommended below.

Education and training

17.56 The need for education and training of HREC members was recognised by the 1996 IEC Report.⁶² The report recommended that AHEC should develop a statement of core competencies for HREC (then called an Institutional Ethics Committee) members to assist in the development of courses for their in-service training and that institutions should make available sufficient ongoing funding to enable their committee members to take up opportunities for training and development.⁶³

17.57 The Inquiry received similar suggestions in submissions.⁶⁴ The Inquiry considers that the education and training requirements of HREC members and support staff should be addressed by the NHMRC, as part of the quality improvement program recommended below.

Payment of HREC members

17.58 HREC members are not paid a sitting fee but may be reimbursed for out-of-pocket expenses. Many HREC members are employed by the institution within which the HREC is located. These 'institutional members' may be remunerated for work connected with service on HRECs (as it forms part of their duties of employment).

61 Queensland Institute of Medical Research, *Submission G036*, 14 January 2002.

62 D Chalmers, *Report of the Review of the Role and Functioning of Institutional Ethics Committees: Report to the Minister for Health and Family Services* (1996), Commonwealth of Australia, Canberra Ch 7.

63 Ibid, Rec 14–15.

64 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Queensland Institute of Medical Research, *Submission G190*, 26 November 2002; NSW Health Department, *Submission G303*, 13 January 2003; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

Non-institutional members are usually unpaid.⁶⁵ However, Commonwealth agencies with HRECs pay sitting fees to non-institutional members on the Remuneration Tribunal scale. This applies, for example, to non-institutional members of HRECs in the Australian Institute of Health and Welfare, the Department of Health and Ageing, and the Department of Defence.

17.59 Payment of HREC members was raised in submissions to the Inquiry and had also been identified in the 1996 IEC Report. It has been suggested that lack of payment for non-institutional members may present a barrier to the effective operation of HRECs.

Because non-institutional members are volunteers, one feels constrained not to ask too much of them and so meetings are not scheduled frequently enough. I believe the solution is to offer payment for attendance at [HREC] meetings at consultancy rates, so the institution does not feel embarrassed at calling on people's voluntary time. I see no conflict of interest for members in accepting such payments, any more than should be felt by any other consultant such as an auditor. Of course, [HREC] members are free to waive their fee if they prefer that their service be seen as for the public good, but this should not be expected. I believe that only in this way can [HRECs] attain the efficiency now required for their important role in the regulatory process for medical research.⁶⁶

17.60 The Inquiry notes that there is no necessary correlation between making payment to non-institutional members and an improvement in the system of ethics review. Most people who agree to assist institutions by becoming members of HRECs do so for altruistic reasons, according to the findings of the 1996 IEC Report. Formalising the HREC system, including introducing member payments, may not necessarily attract the right sort of HREC members. Time, rather than money, may be the more important limiting factor on members' involvement in ethics review. Funding ways to optimise the use of members' time, for example by streamlining expedited ethics approval of low risk research, augmenting HREC secretariat resources and using information technology may be better means of achieving improved ethics review than paying members.

17.61 The New South Wales Health Department suggested that further consideration be given to the issues involved.⁶⁷ The Inquiry makes no specific recommendation in this regard but notes that the issue of payment for HREC members would be appropriate for the NHMRC to consider, in the course of implementing the recommendations in this chapter.

65 Most institutional members add HREC membership to their duties, while a small number, especially committee chairpersons, are relieved of other duties in order to serve as HREC members. Some institutions pay non-institutional members. For example, the Alfred Hospital in Melbourne pays HREC members \$150 per meeting.

66 N Martin, *Correspondence*, 22 April 2002.

67 NSW Health Department, *Submission G303*, 13 January 2003.

Accountability and reporting

17.62 The National Statement requires the NHMRC, through AHEC, to audit the activities of HRECs to ensure compliance with the National Statement.⁶⁸ Institutions and HRECs are required to report annually on several matters including:

- membership and membership changes;
- number of meetings;
- confirmation of participation by required categories of members;
- the number of protocols presented, the number approved and rejected;
- monitoring procedures in place and problems encountered; and
- complaints procedures and the number of complaints handled.⁶⁹

17.63 Institutions and HRECs are also required to provide information from their records to the NHMRC on request.⁷⁰ In the event that annual or other reporting reveals that an institution and its HREC have not complied with the National Statement, NHMRC policy requires that the institution be given an opportunity to explain the non-compliance and to show cause. A proven breach may result in withdrawal of funds, the institution ceasing to be eligible to receive further research funding and the possible naming of the institution in the NHMRC report to Parliament (see Chapter 14).

17.64 The debate about the accountability of the HREC system is on-going. The House of Representatives Standing Committee on Legal and Constitutional Affairs, in its inquiry into human cloning and stem cell research, heard evidence criticising the structure and operation of HRECs including ‘the lack of public accountability in the process and the in-house nature of committees’ and the non-representative nature of their membership.⁷¹ The Committee recommended in September 2001, that the federal government establish an independent review of the institutional ethics committee system in Australia.

Reporting on human genetic research by HRECs

17.65 Submissions emphasised the importance of reporting mechanisms in ensuring that ethical standards in the National Statement are upheld.⁷² There are additional reporting requirements where HRECs are involved in decision making

68 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [2.46].

69 Ibid [2.48].

70 Ibid [1.16], [2.30]–[2.31], [2.47]–[2.48].

71 House of Representatives Standing Committee on Legal and Constitutional Affairs, *Human Cloning: Scientific, Ethical and Regulatory Aspects of Human Cloning and Stem Cell Research* (2001), Parliament of the Commonwealth of Australia, Canberra, 162–165.

72 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002; Australian Academy of Science, *Submission G097*, 21 January 2002.

processes that implement statutory standards, such as the guidelines under s 95 and s 95A of the *Privacy Act*. All ethics review processes need to be transparent and accountable to both research participants and the public in order to ensure that strict ethical standards are upheld. Reporting requirements are critically important in achieving this goal.

17.66 Privacy NSW criticised existing reporting obligations as inadequate, particularly to protect privacy, and suggested a number of reforms, including the establishment of an independent national ethics review committee, and that records of HREC decisions be provided to research participants, the national committee and the federal Privacy Commissioner.⁷³ Other submissions also referred to the need for improved reporting.⁷⁴ The Australian Academy of Science stated that there should be a more comprehensive reporting system for applications that come before HRECs and their outcomes, and that this ‘would allay some concerns over pressures on the system due to commercial arrangements’.⁷⁵ The Department of Human Services South Australia submitted that:

the reporting relationship between HRECs and AHEC/NHMRC should be improved. Requiring a committee to report annually is probably not sufficient. HRECs should become more accountable and transparent, especially given their role in Australia.⁷⁶

17.67 However, other submissions doubted there was any need for further reporting or opposed its introduction.⁷⁷ The Centre for Law and Genetics expressed the view that, provided that changes were made in relation to reporting of waiver of consent decisions, there was no evidence to support further changes in reporting requirements.⁷⁸

17.68 Some were concerned that additional reporting obligations may place further stress on the system for ethics review without any significant benefit.⁷⁹ The NHMRC Research Committee considered that it would be ‘counter productive’ to add more reporting requirements.

Pressure on researchers and HRECs has continued to mount over the years, and it would be counterproductive to add more reporting requirements, or other demands at either the researcher or HREC levels. This is not to say that changes might be

73 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

74 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Australian Academy of Science, *Submission G097*, 21 January 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

75 Australian Academy of Science, *Submission G097*, 21 January 2002.

76 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

77 Centre for Law and Genetics, *Submission G255*, 21 December 2002; D Cavaye, *Submission G110*, 14 March 2002; National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Queensland Government, *Submission G274*, 18 December 2002; NSW Health Department, *Submission G303*, 13 January 2003; Queensland Government, *Submission G274*, 18 December 2002; Medicines Australia, *Submission G302*, 12 February 2003.

78 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

79 D Cavaye, *Submission G110*, 14 March 2002; National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Queensland Government, *Submission G274*, 18 December 2002.

beneficial, but if so, they should be seen within the global context so that changes at one level lead to a relaxing or facilitation of the regulatory process rather than just adding another layer of bureaucracy.⁸⁰

17.69 The New South Wales Health Department firmly indicated that additional reporting would not be beneficial—because of its impact on HRECs and AHEC. In responding to a question about whether HRECs should report commercial arrangements in research to AHEC, the Department stated:

Such an option would inundate AHEC with information about which they may not have the resources to action. HRECs currently review the commercial arrangements for a range of research proposals and the National Statement provides some guidance in this matter. However, HRECs would benefit from more thorough guidelines, other than the advice offered in the National Statement, on matters associated with commercial arrangements and potential conflicts of interest.⁸¹

17.70 The balance to be struck and the interests at stake were clearly put by the Queensland Government.

Commercial arrangements relating to human genetic research proposals have the potential to substantially increase the risk to the privacy of the genetic information if not managed appropriately. There is also a risk that public perception of research could be tainted if sufficient mechanisms are not put in place to protect privacy where there are commercial interests involved.

Commercialisation of human genetic research is a community concern repeatedly expressed in correspondence received by the Queensland Government. Adequate oversight and reporting mechanisms and safeguards are required. However, as discussed in the Inquiry's paper, it is necessary to ensure HRECs are not overburdened with reporting requirements that inhibit their core functions.⁸²

17.71 In Chapter 15 the Inquiry recommended that HRECs should be required to report annually to AHEC with respect to human genetic research proposals for which waiver of consent has been granted under the National Statement (see Recommendation 15–1). The Inquiry makes no further specific recommendations concerning reporting to AHEC, but notes that the issue of reporting, including in relation to commercial arrangements, should be addressed by the NHMRC as part of the quality improvement program recommended below.

Accreditation of HRECs

17.72 The 1996 IEC Report received submissions suggesting a need for greater transparency, monitoring and supervision of HRECs.⁸³ However, the report stated that there was

80 National Health and Medical Research Council Research Committee, *Submission G128*, 18 March 2002.

81 NSW Health Department, *Submission G303*, 13 January 2003.

82 Queensland Government, *Submission G274*, 18 December 2002.

83 D Chalmers, *Report of the Review of the Role and Functioning of Institutional Ethics Committees: Report to the Minister for Health and Family Services* (1996), Commonwealth of Australia, Canberra.

no persuasive evidence of unsatisfactory or poor conduct in the current operation of IECs [as HRECs were then known] to justify the introduction of more stringent inspection (for example, external independent audits) of IECs. Independent audits and the like should not be routinely introduced and should be a 'last-choice' option used when there is evidence of misconduct. There was little support in the submissions for the conduct of random audits.⁸⁴

17.73 The 1996 IEC Report concluded that the most effective method to ensure the accountability of the institutional ethics committee system was more detailed reporting by HRECs to AHEC and by AHEC to the NHMRC.⁸⁵

17.74 The present Inquiry did not identify evidence of poor performance or conduct on the part of HRECs. However, submissions expressed much support for the accreditation of HRECs, reflecting a significant shift in opinion in the years since the 1996 IEC Report.⁸⁶ Submissions argued that accreditation would help ensure high and consistent standards of review, suitable expertise, adequate and continuous training and sufficient resources to fulfil the institutional responsibilities of HRECs.

17.75 The HGSA expressed its support for accreditation and related the need for accreditation to standards and resourcing.

HGSA supports the accreditation of HRECs to ensure that ethical oversight is of the highest standard and also to stimulate organisations to provide an appropriate level of resourcing.⁸⁷

17.76 The Department of Human Services South Australia agreed that accreditation would be a 'positive move'.⁸⁸ The Australian Biospecimen Network saw accreditation as part of a solution to the problem of multi-site approvals.

Moreover, it would be helpful if members of HRECs were provided free training in all aspects relevant to their membership of an HREC, preferably with some form of formal recognition of this training. Such a formal training would then provide a means for 'accreditation' of an ethics committee, which would permit more free reciprocal agreements between HRECs.

At the moment, the requirement to make multiple submissions to potentially dozens of HRECs for multicentre trials implies that not all HRECs are equal, and that by extension individual HRECs are not 'recognised' by other HRECs as having authority to give approval. This absurd situation makes a mockery of the ideal that each HREC abides stringently to the National Statement.

84 Ibid, 54.

85 Ibid, 54.

86 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; J Fleming, *Submission G241*, 20 December 2002; Queensland Institute of Medical Research, *Submission G190*, 26 November 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003; Medicines Australia, *Submission G302*, 12 February 2003.

87 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

88 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

We propose that training be provided to volunteer members of HRECs to provide a certificate of competence to abide by the National Statement, and that completion of such by each member of an HREC provide de facto accreditation to that HREC such that its decisions are recognised as sufficient for reciprocal approval by other HRECs. Random scrutiny of an approved protocol by AHEC would ensure standards are adhered to according to the National Statement and would ensure the individual HRECs provide adequate reporting on how their approval was provided, as well as clear reasons as to why projects were not recommended for approval.⁸⁹

17.77 The Centre for Law and Genetics stated that there 'is no pressing case for the introduction of a formal system of accreditation of HRECs'⁹⁰ but, along with several other submissions, argued that although the present system is satisfactory it needs work to ensure that HRECs and institutions are fully aware of their responsibilities.⁹¹ The Australian Red Cross Ethics Committee stated that:

In view of the type of applications considered by our Committee, we believe that it is not inappropriate for HRECs dealing with human tissue research applications to be more regulated than other HRECs. Such research raises unique issues ... Our Committee would not object to accreditation procedures being developed.⁹²

17.78 In North America, accreditation of human research ethics review processes has been either recommended or implemented. In Canada, a task force set up by the National Council on Ethics in Human Research (NCEHR) recommended in March 2002 that the NCEHR affirm the need for nationwide oversight of ethics review processes, in the form of an accreditation program to be conducted by 'an arm's-length, non-governmental organization'.⁹³

17.79 In the United States, the Institute of Medicine recommended accreditation as one mechanism for enhancing the effectiveness and reliability of programs for the protection of human research participants.⁹⁴ The Institute reaffirmed these recommendations in a later report.⁹⁵ In a comprehensive and detailed report, the United States National Bioethics Advisory Commission (NBAC) also recommended accreditation of those programs.⁹⁶

17.80 There are several current accreditation programs in the United States. One is conducted by the American Association for Accreditation of Human Research Protection Programs Inc, a non-profit corporation that offers accreditation of programs

89 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

90 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

91 Department of Health Western Australia, *Submission G271*, 23 December 2002; NSW Health Department, *Submission G303*, 13 January 2003.

92 Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003.

93 The National Council on Ethics in Human Research, *Final Report of the Taskforce to Study Models of Accreditation for Human Research Protection Programs in Canada* (2002), The National Council on Ethics in Human Research, Ottawa.

94 Institute of Medicine, *Preserving Public Trust: Accreditation and Human Research Participation Protection Programs* (2001), National Academies Press, Washington.

95 Institute of Medicine, *Responsible Research: A Systems Approach to Protecting Research Participants* (2002), National Academies Press, Washington.

96 National Bioethics Advisory Commission, *Ethical and Policy Issues in Research Involving Human Participants Vol I: Report and Recommendations of the National Bioethics Advisory Commission* (2001), National Bioethics Advisory Commission, Bethesda.

on a user pays basis, using site visits. The National Committee for Quality Assurance administers an accreditation program that is mandatory for all health centres operated by the Department of Veterans Affairs. The Office for Human Research Protections, within the Department of Health and Human Services, has established a program of quality improvement based on an initial self-assessment, followed by site visits and assistance.

17.81 The term accreditation is capable of referring to a wide range of processes, from simple registration based on annual returns, to detailed scrutiny of committee procedures and decision making processes. The essential characteristics were described by the NBAC as follows:

These programs generally involve experts and peers developing a set of standards that represents a consensus of the best practices in the profession. Therefore, the choice of standards and the criteria for evaluating whether an institution has met them are critically important. The emphasis of these programs should be on education, on assuring that appropriate protections are in place, and on avoiding excessively bureaucratic procedures. Accrediting programs may lose their value if they are seen as merely another administrative burden.

Despite concerns that accreditation might add another layer of bureaucracy to an already regulated environment, several advantages of accreditation programs have been noted. They are generally voluntary and represent a profession's desire to self-regulate. Many accrediting programs strive to meet higher standards than are required by law, so that having the credential implies a higher level of competence than what may be minimally required. Properly governed and organized, accrediting bodies can improve competence (and thus performance) within a profession, which helps to achieve the goals of the profession and build public trust (Hamm 1997). In addition, accreditation serves an educative role.

Institutions seeking accreditation, for example, usually go through a periodic review process involving a self-study and a site visit from a team of experts in the profession. The self-evaluation of the institution in preparation for a site visit helps it identify strengths and weaknesses in its programs and make improvements. The site visit can be an especially useful learning experience that encourages the institution to sustain best practices.⁹⁷

17.82 Consistently with submissions to the Inquiry, an accreditation system would need to be based on the articulation of detailed standards of performance, an initial quality improvement phase to inform, train and equip HRECs, and commitment of institutions to those standards. There may need to be a process of site visits and certification for identifying and remedying defects. These initiatives would require a significant increase of resources for AHEC.

17.83 The Inquiry has concluded that a system of formal accreditation offers the prospect of substantially strengthening HREC review of human genetic research proposals. As such, the Inquiry believes that accreditation is worthy of further detailed consideration by the NHMRC, but that such a scheme is feasible only when clear

97 Ibid, 49.

standards for consistency, efficiency, transparency and accountability of HREC review of human genetic research have been established.

Inquiry's views

17.84 The Inquiry notes the developments in the regulatory framework for review of research proposals by HRECs, which followed the 1996 IEC Report and the extensive consultations prior to the publication of the National Statement in 1999.

17.85 The Inquiry also notes that many of the HREC reforms proposed in submissions and consultations do not relate solely to human genetic research but to the conduct of research involving humans generally. In Chapter 14, the Inquiry has recommended that the NHMRC review mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.

17.86 The Inquiry considers that, on the basis of submissions and consultations, the present ethics review system could be significantly improved. The weaknesses identified have attracted criticism and may constitute threats to the effectiveness and public standing of the system.

17.87 The evidence available to the Inquiry does not show that the conduct of review and monitoring of human genetic research has exposed research participants to unacceptable risks. However, the evidence does show that many HRECs lack resources and relevant expertise. Submissions also suggest that improvements in reporting by HRECs are needed to ensure that the relevant standards are consistently and expertly applied.

17.88 The Inquiry has concluded that the first priority for the NHMRC should be to develop and implement a quality improvement program for HRECs to promote consistency, efficiency, transparency and accountability in the review of human genetic research by HRECs. A second priority should be for the NHMRC to undertake a detailed examination of the need for an accreditation system for HRECs. Fundamental to both initiatives is the provision of adequate resources by institutions to enable HRECs to fulfil their responsibilities.

Recommendation 17–1. The National Health and Medical Research Council (NHMRC) should develop and implement procedures to promote consistency, efficiency, transparency and accountability in the review of human genetic research by Human Research Ethics Committees (HRECs). In developing such procedures, the NHMRC should initiate a systematic quality improvement program that addresses:

- (a) consolidation of ethical review by region or subject-matter;
- (b) the membership of HRECs and, in particular, the balance between institutional and non-institutional members;

- (c) the need for expertise of HRECs in considering proposals for human genetic research;
- (d) on-going monitoring of approved human genetic research projects;
- (e) the education and training of HREC members;
- (f) payment of HREC members for their work in reviewing research proposals;
- (g) independent audit of HREC processes; and
- (h) standardised record keeping and reporting to the NHMRC, including in relation to commercial arrangements.

Recommendation 17–2. In the course of developing a quality improvement program for HRECs in accordance with Recommendation 17–1, the NHMRC should review the need for an accreditation system for HRECs in their ethical review of human genetic research.

Recommendation 17–3. As part of the process of strengthening HREC review of human genetic research, each relevant institution and organisation should provide adequate resources to enable its HREC to fulfil its institutional responsibilities and achieve the standards set in accordance with Recommendations 17–1 and 17–2.

Part E. Human Genetic Databases

18. Human Genetic Research Databases

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Introduction

18.1 Increasingly, researchers are compiling collections of genetic samples and information to aid studies into the causes of disease, drug reactions, and the interaction between genetic status and the environment. The importance of human genetic databases has come in part as a result of developments in the field known as 'bioinformatics'. Bioinformatics combines computer science, biology and mathematics to produce tools that enable the storage and analysis of large quantities of biological information.¹ The advent of this science has lent new value to large collections of genetic material and information, allowing researchers to perform studies on a scale impossible previously.

1 K Dearne, 'Biotech Industry Booming', *The Australian*, 14 May 2002, 3.

18.2 This chapter is concerned with the collection, use, storage, disclosure and transfer of genetic samples and information that have been collected primarily for use in research and are held in collections by hospitals, public and private research organisations, and in the archives of pathology laboratories.

18.3 Other collections of genetic samples and information exist, which can be used in research, although they were not created primarily for that purpose. These include pathology collections, banked tissue, newborn screening cards, and other collections, such as those maintained as part of a population screening program. Collections of this kind may have great value for research because they include samples taken over a long period of time, or from a unique class of patients. These databases are examined in Chapter 19.²

18.4 This chapter begins by explaining the terms ‘human genetic database’ and ‘human genetic research database’ and describes (with reference to Australian and overseas examples) how these databases are formed and how they are used in research.

18.5 The chapter then examines aspects of the existing regulatory framework that applies to the operation of human genetic research databases. As with other aspects of research involving humans, the relevant regulatory framework centres on the National Health and Medical Research Council’s (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement)³ and on statutory restrictions on dealing with personal information under the *Privacy Act 1988* (Cth) (*Privacy Act*)⁴ and similar state and territory legislation.

18.6 The Inquiry has concluded that new regulation of human genetic research databases is necessary. The Inquiry has taken a ‘light-touch’ approach, recommending the inclusion of new provisions in the National Statement to provide ethical guidance on the operation and use of human genetic research databases. The Inquiry also recommends the introduction of a registration system, administered and overseen by the NHMRC, and the use of a gene trustee to protect the privacy of samples and information held in databases, where appropriate. In addition, the Inquiry is of the view that policies should be developed to control the use and disclosure of genetic samples and information held in human genetic research databases for other purposes, including law enforcement.

What are human genetic research databases?

18.7 The term ‘human genetic database’ may refer to many kinds of collections of genetic samples and genetic and other health information. Genetic samples contained in research collections can include a wide range of human biological materials such as

2 Human genetic databases created for law enforcement and forensic purposes are discussed in Part J.

3 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

4 And related guidelines: National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (2000), NHMRC, Canberra; National Health and Medical Research Council, *Guidelines Approved Under Section 95A of the Privacy Act 1988* (2001) National Health and Medical Research Council.

extracted DNA, body fluids, cells and sections of tissue. The information in a human genetic database may include molecular genetic data, standardised clinical data, genealogical data, and information on the health, lifestyle and environment of an individual.⁵

18.8 This chapter uses the term ‘human genetic research databases’ to refer broadly to collections of genetic samples and genetic and other health information, in any combination, which have been established for the purpose of human research. Some databases may be maintained for the study of a specific genetic condition; others will be used in many types of genetic research. However, the unifying element is that human genetic research databases have been created primarily for the purposes of medical or other human research. In contrast to samples and information contained in archival pathology collections and other human tissue collections (see Chapter 19), the samples and information in human genetic research databases are stored with consent for their possible use in research projects, which are submitted to a Human Research Ethics Committee (HREC) for approval, and for which further specific consent is sometimes required.

18.9 Other terms such as ‘biobank’, ‘gene bank’ and ‘tissue bank’ have also been used to denote what this chapter refers to as human genetic research databases. In Australia, the term ‘tissue bank’ is often used to denote collections of genetic material that may be used in human genetic research—for example, the Peter MacCallum Cancer Institute (PMCI) Tissue Bank.

18.10 These additional terms demonstrate the lack of consensus over what is meant by a genetic database, which itself stems from the diversity of existing collections. For example, the term ‘biobank’ has been used in Sweden and the United Kingdom. In the United Kingdom, the UK Biobank project encompasses the collection of blood samples and health and lifestyle information.⁶ In Sweden the term has been used more restrictively to mean collections of identifiable tissue specimens.⁷ Legislation establishing the Estonian Genome Project defines a ‘gene bank’ as

a database established and maintained by the chief processor consisting of tissue samples, descriptions of DNA, descriptions of state of health, genealogies, genetic data and data enabling the identification of gene donors.⁸

5 W Lowrance, ‘The Promise of Human Genetic Databases’ (2001) 322 *British Medical Journal* 1009. See also the definition of ‘human genetic database’ in House of Lords Select Committee on Science and Technology, *Human Genetic Databases: Challenges and Opportunities* (2001), The Stationery Office Limited, London [3.3], and the WHO definition of genetic database in F Dekkers and others, *Genetic Databases: Assessing the Benefits and the Impact on Human & Patient Rights* (2001), World Health Organization, Geneva, 6.

6 People Science and Policy Ltd, *BioBank UK: A Question of Trust: A Consultation Exploring and Addressing Questions of Public Trust* (2002), Medical Research Council and the Wellcome Trust, London, 8.

7 Swedish National Board of Health and Welfare, *Biobanks in Medical Care*, <www.sos.se/sos/publ/refereng/0077011e.htm>, 20 February 2003.

8 *Human Genes Research Act 2000* (Estonia) s 2(10).

Research value

18.11 Much of the research value of human genetic research databases is derived from linkages created between clinical, personal and genetic information. Examining these linkages is an important tool in identifying the genetic causes of disease and in other forms of human genetic research.

18.12 Different forms of genetic research can be conducted using human genetic databases. These include:

- linkage studies to identify the gene sequences associated with inherited diseases;
- association studies to find correlations between a disease and a genetic change where there is no obvious pattern of inheritance;
- genetic epidemiology studies of the interaction between genes and environment; and
- pharmacogenetic studies to determine if there is a genetic basis for certain adverse reactions to drugs.⁹

18.13 Each of these studies requires access to a different type of human genetic database, or uses databases in a different way, and may raise different issues.¹⁰ Some linkage studies map genetic sequences to identify genetic changes linked to the existence of an inherited disorder, where a distinct familial pattern of disease inheritance can be seen. They require collections of tissue taken from family members and information about which members suffer from the disorder.¹¹

18.14 By contrast, association studies require large collections of samples from people with a given condition, combined with detailed medical histories. These studies examine the genetic causes of diseases that do not follow a clear inheritance pattern, and attempt to make statistical correlations between gene sequences and a disease. Large populations are studied because such diseases may show a marked genetic cause in only some individuals, or a weak genetic cause in many individuals. Large populations also increase the accuracy of statistical correlations.¹²

18.15 Genetic epidemiology studies examine the interaction between genetic and environmental factors in causing diseases and susceptibilities. Studies of this type require access to very large population collections as well as detailed medical histories.¹³

9 J Kaye and P Martin, *The Use of Biological Sample Collections and Personal Medical Information in Human Genetics Research* (1999), Wellcome Trust, London, 9–13.

10 *Ibid.*, 11 and following.

11 *Ibid.*, 10–11.

12 *Ibid.*, 11–12.

13 *Ibid.*, 12–13.

18.16 Databases are particularly valuable for pharmacogenetics—the study of genetic causes of variable drug responses. By linking clinical records with genetic information, databases correlate the effects of medication and variations in genetic makeup on a scale large enough to produce statistically significant results. As a result, many pharmaceutical and biotechnology companies seek access to human genetic databases, or build their own collections.

Types of human genetic research databases

18.17 Human genetic research databases vary in scale from limited collections, created for a specific study or series of studies, to major population databases that store samples and information from very large sections of a population and are used for different studies over many years. Research databases may be established by research organisations for use in their own studies or they may be established on a commercial basis for sale to interested researchers or research organisations.

18.18 In Australia, most human genetic research databases have been established by research organisations for use in their own studies.¹⁴ For example, the Menzies Centre for Population Research maintains a research database comprising extensive genealogical data, genetic samples, and health information supplied by donors, to search for genetic causes of disease. All material is donated by volunteers specifically for the Centre's research projects.¹⁵ Sometimes, research databases are established by research organisations connected with hospitals and other health care organisations. For example, the PMCI Tissue Bank was established in 1998 to conduct molecular genetic studies in cancer.¹⁶ Cancerous tissue is obtained from patients via surgical and pathology staff attached to the PMCI.

18.19 Although not yet a feature of Australian research culture, donor tissue banks are being constructed on a commercial basis in the United States. Such banks collect tissue from hospitals and process them ready for research. The banks themselves do not conduct research, but sell processed tissue to researchers at other institutions. An example is Gene Logic, a US tissue repository containing more than 10,000 tissue samples that are made available to researchers and pharmaceutical companies. Samples are supplied by hospitals once consent has been given.¹⁷

18.20 Human genetic research databases may also be created on a grand scale to encompass the genetic samples and information of large sections of the population. These databases may be used for a wide variety of research projects by different groups of researchers. The Estonian Genome Project, the Iceland deCODE project and UK Biobank are examples of such databases.

14 Although other researchers may also be granted access.

15 Menzies Research Institute, *Home Page*, <www.menzies.utas.edu.au/>, 20 February 2003.

16 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

17 GenomeWeb Staff Reporter, *New Tissue Repository Firm to Close Microarray, IP Collaborations in 6 Months*, GenomeWeb, <www.genomeweb.com/articles/view.asp?Article=2002627173511>, 27 June 2002.

18.21 In Estonia, a non-profit government project to form a database of the genetic information from three-quarters of the country's population is underway. The centralised electronic database will contain blood samples and health data information, each collected and stored separately, for use in large-scale association studies. Control of the database is split between the Estonian Genome Project Foundation (the government body that will own the database) and EGeen (the exclusive commercial licensee that also finances the project). EGeen will have access to the database for research into the causes of disease and to develop drugs and treatments.¹⁸

18.22 A similar project has already been undertaken in Iceland by deCODE—a company that holds an exclusive licence to create and operate a database of genetic and other health information, known as the Iceland Health Sector Database. The project brings together three types of information—coded health information taken from Iceland's national health care system records, genealogical data, and genetic information from samples obtained and analysed with the consent of Icelandic donors.¹⁹

18.23 Both these projects involve an exclusive licence for access to medical and genetic information, and to genetic samples, demonstrating the growth of major commercial interest in databases. In each case, genetic information is not publicly available, but is controlled in part by commercial entities.

18.24 A different approach to the development of a major genetic research database has been taken in the United Kingdom. UK Biobank is a publicly funded project to collect samples and health information from 500,000 volunteers. The project aims to provide sufficiently comprehensive health information and a large enough sample size to enable more effective studies of the interactions between genotype and environment. Participants will supply updated medical and environmental exposure information every five years.²⁰ Researchers will be given access to the central database following application and approval from an oversight body.²¹

Regulation of human genetic research databases

18.25 The collection, storage, use and disclosure of genetic samples and information held in human genetic research databases are regulated by a mixture of legislation, guidelines and standards. These include:

- the legislative framework for the protection of information and health privacy based on the *Privacy Act* and similar state and territory legislation (see Chapters 7 and 8);

18 Estonian Genome Foundation, *Genome Project*, <www.genomics.ee/index.php?lang=eng&show=20>, 20 February 2003.

19 DeCODE Genetics, *Home Page*, <www.decode.com>, 20 February 2003. See also H Rose, *The Commodification of Bioinformation: The Icelandic Health Sector Database* (2001) Wellcome Trust.

20 The UK Biobank, <www.ukbiobank.ac.uk>, 19 February 2003.

21 Medical Research Council and the Wellcome Trust, *Draft Protocol for BioBank UK: A Study of Genes, Environment and Health*, 1 February 2002 [2.3.1].

- state and territory Human Tissue Acts, which require consent for donation of human tissues for research;
- ethical guidelines contained in the National Statement²² (see Chapters 13–15); and
- the Human Genetics Society of Australasia *Guidelines for Human DNA Banking* (the HGSA Guidelines).²³

Information and health privacy legislation

18.26 Chapter 7 summarised the legislative framework for the protection of information and health privacy based on the *Privacy Act* and similar state and territory legislation.²⁴ Chapter 8 discussed the application of this legislation to genetic samples.

18.27 The *Privacy Act*, and similar state and territory legislation, is intended to protect the personal information of individuals and to give them control over how that information is collected, used and disclosed. This legislation sets out safeguards that organisations must observe in collecting, storing, using and disclosing personal information. It also gives individuals rights to access and correct their personal information.

18.28 The protection provided by information and health privacy legislation extends to genetic information and is an important element in regulating the operation of human genetic research databases. An organisation covered by information or health privacy legislation must handle genetic information (including that in a human genetic research database) in accordance with legislative privacy principles. A range of issues relevant to the privacy of genetic samples and information held in human genetic research databases is discussed below.

Harmonisation of health privacy law

18.29 Chapter 7 highlighted problems that arise from the absence of a comprehensive framework for the regulation of health information across the public and private sectors, at the federal, state and territory level. There, the Inquiry recommended that the Commonwealth, States and Territories should pursue the harmonisation of information and health privacy legislation as it relates to human genetic information (Recommendation 7–1). The absence of uniform laws has particular implications for the regulation of human genetic research databases, which constitute an additional reason for Commonwealth, state and territory governments to pursue harmonisation.

22 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

23 Human Genetics Society of Australasia, *Guidelines for Human DNA Banking*, Guidelines, 1 July 1990.

24 *Privacy and Personal Information Protection Act 1998* (NSW); *Health Records and Information Privacy Act 2002* (NSW); *Health Records Act 2001* (Vic); *Information Privacy Act 2000* (Vic); *Health Records (Privacy and Access) Act 1997* (ACT).

18.30 The *Privacy Act* applies to genetic information in databases maintained by federal and Australian Capital Territory government agencies and private sector organisations,²⁵ including private hospitals. In general, commercial organisations are covered by the *Privacy Act* unless they are established for a public purpose by state or territory law (other than as incorporated companies).²⁶

18.31 Major human genetic research databases are generally maintained by public hospitals, universities or research institutes. In most cases, as state or territory authorities,²⁷ they are covered by state or territory information and health privacy legislation, where it exists.²⁸

18.32 Some human genetic research databases are not covered by any information or health privacy legislation. For example, the database held by the Menzies Centre for Population Research, as an organisation within the University of Tasmania, is not subject to privacy legislation, because Tasmania has not enacted such legislation.

18.33 It may not be clear which privacy legislation, if any, will apply where researchers affiliated with hospitals or universities are funded by an external source or work in collaboration with private sector organisations. In such instances, the application of legislation may need to be considered on a case-by-case basis taking into account factors like the nature of the contract, if any, between the researcher and funding body, and the impact of the funding on the relationship between the university or hospital and the researcher.²⁹

18.34 For example, the Murdoch Children's Research Institute carries out genetic research using samples and information obtained from the Royal Children's Hospital and is affiliated with the University of Melbourne. Researchers are employed by the Institute, which is an incorporated body covered by the *Privacy Act*. However, the hospital and university are covered by the Victorian legislation.

18.35 Concerns about the lack of uniform principles for storage, use and disclosure of genetic information were expressed in submissions.³⁰ In relation to databases, the Centre for Law and Genetics commented that:

25 *Privacy Act 1988* (Cth) s 6C(1).

26 *Ibid* s 6C(3)(c).

27 *Ibid* s 6C(1)–(3). Most universities in Australia are state or territory authorities, for example *University of Sydney Act 1989* (NSW); *University of Tasmania Act 1992* (Tas). However, three private universities, Bond University, the University of Notre Dame, and Melbourne University Private may be covered by the federal *Privacy Act*.

28 *Privacy and Personal Information Protection Act 1998* (NSW); *Health Records and Information Privacy Act 2002* (NSW); *Health Records Act 2001* (Vic); *Privacy Act 1988* (Cth); *Health Records (Privacy and Access) Act 1997* (ACT).

29 M Otlowski, 'Protecting Genetic Privacy in the Research Context: Where to From Here?' (2002) 2 *Macquarie Law Journal* 87.

30 Genetic Support Council WA, *Submission G112*, 13 March 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002.

The lack of uniform privacy legislation throughout the country remains a major issue in this area. Were the states and territories to introduce complimentary privacy legislation to the Commonwealth legislation there would be an improved framework for the protection of human genetic databases.³¹

Coverage of genetic samples

18.36 In Chapter 8, the Inquiry recommended that the *Privacy Act* be amended to cover genetic samples, as well as the genetic information derived from them (Recommendation 8–2). This recommendation addresses the gap in the existing framework for protecting the privacy of genetic samples. Genetic samples are closely analogous to genetic information and the *Privacy Act* appears capable of extending appropriate privacy protection to them.

18.37 Human genetic research databases often include genetic samples, information derived from the analysis of the samples, as well as other health information. The samples and information are used in close association to facilitate research. Similar privacy principles should be applied to the handling of genetic samples and the information derived from them. The exclusion of samples from the coverage of privacy legislation poses a problem because, as discussed in Chapter 8, individual privacy rights in respect of samples may not be asserted by the individuals from whom the samples were taken.

Prospective application

18.38 The National Privacy Principles concerning the collection, use and disclosure of information apply only to information collected after 21 December 2001.³² The *Privacy Act* does not constrain the use or disclosure of existing information stored in human genetic research databases by private sector organisations, even where fully informed consent has not been obtained. Privacy NSW stated:

This is a very serious concern with respect to information contained in human genetic databases, including human tissue samples, collected before the commencement of the *Privacy Act*'s private sector amendments. The use of such information for purposes other than which it was collected should be strictly regulated and consent should generally be required for such use.³³

18.39 Research organisations have emphasised the problems involved in complying with new regulation in relation to the samples collected in the past.³⁴ For example, the Queensland Institute of Medical Research (QIMR) stated:

31 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

32 *Privacy Act 1988* (Cth) s 16C.

33 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

34 Peter MacCallum Cancer Institute, *Submission G028*, 20 December 2001; Queensland Institute of Medical Research, *Submission G036*, 14 January 2002.

we feel we can comply with pretty well any new regulations as they are introduced, and these have grown more demanding over the last 15 years. What we are quite unable to cope with is the idea that use of samples or data collected in the past should have to comply with new regulations.³⁵

18.40 The cost, time and ethical tensions involved with obtaining subsequent consent from donors to unspecified future research was discussed in Chapter 15. There is clearly a tension between the interest of the research participant in privacy and the interest of researchers in gaining access to samples and information. In the long term, the problem is best addressed by allowing human genetic research databases to obtain broad consent to the use of the samples in future research.³⁶ The use of mechanisms such as a gene trustee can reduce significantly the tension between interests in privacy and research.

Human Tissue Acts

18.41 The Human Tissue Acts³⁷ lay down consent provisions for the donation of three classes of tissue—blood, regenerative tissue and non-regenerative tissue.³⁸ Regenerative tissue is any tissue that can be replaced by the body after removal, while non-regenerative tissue is any tissue other than regenerative tissue.³⁹ Distinctions are also made between donations from living and deceased persons.

18.42 Living persons may donate blood for transfusion and use for therapeutic, medical or scientific purposes with verbal consent.⁴⁰ It is not clear whether consent to one use constitutes consent to the other uses, but the Acts appear to require specific consent to each use. For example, consent to transfusion would not necessarily entail consent to use for research purposes (which the Acts call ‘scientific’ purposes). Similar provisions apply to living donations of regenerative tissue; however, consent must be in writing and may require either certification from a doctor or witnessing by a family member.⁴¹

18.43 Any tissue from a deceased person may be donated for transplantation or therapeutic uses, or for other medical or scientific purposes where consent to do so was expressed during life. Tissue also may be donated if the donor expressed no objections during life and the senior available next of kin gives consent. Only the Western Australian Act explicitly requires that tissues be used only for the purpose for which

35 Queensland Institute of Medical Research, *Submission G036*, 14 January 2002.

36 See Ch 15.

37 *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1979* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1985* (Tas); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplant Act 1982* (WA); *Transplantation and Anatomy Act 1978* (ACT); *Human Tissue Transplant Act 1979* (NT).

38 Sperm, ova and foetal tissue are expressly excluded from the ambit of the legislation: *Human Tissue Act 1983* (NSW) s 6 and cognate state and territory legislation. The exclusion only applies to donations from living persons, but the legislation has generally been taken to exclude donations from the dead also.

39 *Ibid* s 4(1) and cognate state and territory legislation.

40 *Ibid* ss 19, 21 and cognate state and territory legislation.

41 *Ibid* ss 7, 9, 12 and cognate state and territory legislation.

they were removed,⁴² but this appears to be the meaning of the provisions in the remaining Acts.

18.44 The Human Tissue Acts do not comprehensively regulate the collection, storage and use of tissue samples as part of human genetic research databases. For example, they contain no provisions regulating the storage, use or transfer of samples. In relation to consent, the Acts require only that donors be informed of the nature and effect of the removal of tissue. They do not require donors to be informed that their tissue may be stored, disclosed to other researchers or linked with health information.

18.45 The Human Tissue Acts could be amended to include new provisions to cover collection, storage, use or transfer of samples (including those held in human genetic research databases). This option is discussed in Chapter 20 but is not supported by the Inquiry. The Inquiry has concluded that comprehensive reform of the law relating to the handling of genetic samples should build on existing information and health privacy legislation rather than the Human Tissue Acts (see Chapter 8).

National Statement

18.46 The National Statement⁴³ contains no provisions dealing specifically with human genetic research databases.⁴⁴ However, many of its general provisions are relevant to the operation of databases.

18.47 The National Statement requires researchers to ensure the confidentiality and privacy of stored genetic information, where identified or potentially identifiable.⁴⁵ Research protocols must make clear whether information is to be stored and in what form—identified, potentially identifiable (coded), or de-identified (not identifiable, anonymous).⁴⁶ Individuals should be informed of any intention to store genetic samples or information.⁴⁷

18.48 Where research involves linkage of data sets (as is common with research involving databases), the National Statement provides that an HREC may approve the use of identifiers to ensure that the linkage is accurate, but once the linkage has been completed the HREC should require that the resulting data be coded or de-identified.⁴⁸

42 *Human Tissue and Transplant Act 1982* (WA) s 22(3).

43 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

44 With the possible exception of [15.7], which relates to consent to the use of tissue samples held in 'banks'. In addition, the NHRMC has released guidelines for genetic registers: National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra. While genetic registers are used primarily in the provision of health care, they may also be used to facilitate research (see Ch 22).

45 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [16.3].

46 *Ibid* [16.5].

47 *Ibid* [15.5(c)], [16.10(j)], [18.4].

48 *Ibid* [14.8]. In certain circumstances, an HREC may permit use of personal information for record linkage without consent: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra [18.5].

18.49 In addition, the National Statement generally requires consent to the use of human tissue samples, genetic material and genetic information in medical research. As discussed in Chapter 15, these consent requirements can be waived by an HREC.

Guidelines for Human DNA Banking

18.50 The HGSA Guidelines use the term ‘DNA bank’ to refer to collections of genetic material maintained by both clinical service groups and researchers.⁴⁹ The HGSA Guidelines set standards for banking of DNA and are directed, in part, at ensuring that possible future health service needs of families will be met where DNA is stored.

18.51 While the HGSA Guidelines mainly address storage of DNA in the context of clinical health services, they also consider situations where research into genetic disorders may later have implications for health service delivery. The HGSA Guidelines state that researchers have obligations to the families they are studying. Databases should be properly documented, and the records made available to whoever will be providing for the health service needs of the families. There should be clear policy about the use of DNA samples from an elderly subject with a reduced life expectancy, to allow some to be set aside for future needs. Proper consideration should be given to the later health service needs of families once research has been completed.⁵⁰

18.52 The HGSA Guidelines recommend the establishment of a central directory of DNA banks storing material for both clinical and research purposes, with the register to be maintained by one centre in each region. Any persons storing DNA would be under an obligation to notify the appropriate centre in their region, and any extraction and storage of DNA should be reported to the centre. It is recommended that the centre be a regional clinical genetics service.⁵¹ The Guidelines offer no recommendations specifically directed to the conduct of research, but state that research using DNA samples should be regulated by the NHMRC.⁵²

The need for reform

Privacy

18.53 Genetic databases raise privacy concerns because they store large quantities of genetic samples and information that may be accessed by many different researchers, over many years, and for many different research purposes. These concerns include questions about consent to storage, use and re-use of genetic samples and information, linking of information to genetic samples, and the extent of disclosure of samples and information. The volume of information that might come to be held in human genetic research databases magnifies these concerns.

49 Human Genetics Society of Australasia, *Guidelines for Human DNA Banking*, Guidelines, 1 July 1990.

50 *Ibid* [2.5].

51 *Ibid* [4.7].

52 *Ibid* [2.7].

18.54 Linkages within databases create privacy concerns, as the combination of samples and information can yield new information in itself. Collections may build up a more comprehensive picture of an individual's health than is held in any other form, such as by linking detailed genealogical information with the health records of a number of family members and data derived from testing genetic samples.

18.55 Storage of genetic samples or information in human genetic research databases may also increase the chances that such samples or information can be re-identified. While genetic samples, medical records, and lifestyle information may all have been coded or de-identified,⁵³ bioinformatic technologies may link samples and information in a way that reveals the identity of the individual to whom they relate.⁵⁴

18.56 Protecting privacy must be balanced against the promotion of research. Maintaining public confidence in genetic research is crucial because much of the tissue used in research in Australia is obtained through altruistic donation. Such altruism might be adversely affected if community trust in the operation of human genetic research databases is eroded. The Office of the Federal Privacy Commissioner (OFPC) observed:

It is vital to acknowledge the importance of maintaining public confidence in the operation of human genetic databases and the altruistic donation of samples and information to those databases.⁵⁵

Consent for unspecified future research

18.57 An issue of particular relevance to the operation of human genetic research databases is the extent to which researchers are effectively able to obtain consent for unspecified future research. This issue was also examined in Chapter 15, where the Inquiry recommended that the National Statement should establish new guidelines dealing specifically with obtaining consent to unspecified future research (Recommendation 15–4).

18.58 Some human genetic research databases in Australia already operate by obtaining consent to unspecified future research and there was support for facilitating this process. Major human genetic research databases may not be able to operate effectively without the ability to seek broad and durable consent to the use of genetic samples and information in research, given the cost and time involved in obtaining specific consent from large numbers of donors.

53 Under the National Statement, information or material is 'de-identified' only if the process of de-identification is 'irreversible'—for example because the identifiers have been removed permanently or because the data have never been identified: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra, 9.

54 On linkage of medical information, see R Magnusson, 'Data Linkage, Health Research and Privacy: Regulating Data Flows in Australia's Health Information System' (2002) 24(1) *Sydney Law Review* 5.

55 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003. The RCPA also referred to the importance of maintaining the confidence and support of the public in the operation of human genetic databases: Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002.

18.59 Where samples and information are already stored in a human genetic research database, and neither consent to unspecified future research nor specific consent to proposed research has been obtained, the issue of retrospective consent will also arise. This issue will be especially pertinent for databases that have been developed from existing collections, from samples and information left over from clinical use, and from the conversion of clinical databases to research use.

18.60 Obtaining subsequent consent from donors for future research is a costly and time-consuming process for research organisations. The primary reason for favouring broad initial consent is that further consent is not necessary when researchers wish to apply new research techniques⁵⁶ or conduct new research that could not have been foreseen at the time of the donation.

Research techniques are developing at such a rapid rate that it was not possible 5 years ago to describe with any detail projects that are being pursued today. The same is true of the relationship between techniques available today and research five years from now.⁵⁷

18.61 In the context of cancer research there are other important considerations. For example, some cancers, such as ovarian and gastric cancer, have high rates of mortality.⁵⁸ To obtain consent retrospectively for individual research projects would be difficult in cases of aggressive and fatal disease. To 're-consent' patients for new studies may be impossible, due to death, or disturbing to surviving patients.⁵⁹ The PMCI submitted that the timely conduct of research into the causes of cancer would be significantly prejudiced if, instead of being able to use existing collections of human tissue for new cancer research, fresh collection of tissue were to be required.⁶⁰

18.62 Many Australian human genetic research databases utilise procedures that involve obtaining broad consent for the use of tissue in research, generally from donors whose tissue is being surgically excised for diagnostic and therapeutic purposes. In effect, this consent purports to cover unspecified future research.

18.63 For example, the PMCI uses broad consent, rather than specific protocol-based consent, to facilitate the future research use of tissue, as approved by its ethics committee.⁶¹ A similar approach is used by the Western Australia Research Tissue Network.⁶² The consent form for donors to the PMCI tissue bank states that the sample will be used for 'biochemical and genetic studies on the cause of cancer'.⁶³ Donors are

56 For example, new DNA microarray techniques: Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

57 Peter MacCallum Cancer Institute, *Submission G070*, 20 December 2001.

58 For example, in the case of a national study of ovarian cancer the majority of patients die within three to five years of diagnosis: Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

59 Peter MacCallum Cancer Institute, *Submission G070*, 20 December 2001.

60 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

61 Peter MacCallum Cancer Institute, *Submission G070*, 20 December 2001; Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002; Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 15 March 2002.

62 N Zeps, *Submission G047*, 14 January 2002; Children's Cancer Research Institute, *Consultation*, Perth, 4 December 2001.

63 Peter MacCallum Cancer Institute, *Information and Consent Form Protocol No 98/36* (2002).

informed that researchers associated with the PMCI will have access to the samples for approved research purposes only and that access to donated tissue will be regulated by the PMCI ethics committee.⁶⁴ The consent form for the Western Australia Research Tissue Network simply refers to donation of tissue or blood for medical research and states that storage of and access to tissue will be managed by an Advisory Committee and only released where approved by an HREC.⁶⁵

18.64 Specimens collected from oncology patients at the Children's Hospital at Westmead are stored within the Children's Hospital at Westmead Tumour Bank and research on the specimens is regulated by the Tumour Bank Committee under the auspices of the hospital HREC.⁶⁶ The hospital seeks consent for unspecified future research by adding to its admission form the following clause:

I understand and agree that blood, urine, and other samples may be taken for the diagnosis and treatment of myself/my child. Any unused part of the samples may be retained for future research.⁶⁷

18.65 A two-stage consent process is used. Initial consent is obtained, as described above. At a later stage, consent to store samples in the Tumour Bank for use in future research is obtained, at a time when prospective research participants are in a better position to give informed consent. The rationale for this process was explained as follows:

It is our belief that to approach parents of newly diagnosed childhood cancer patients immediately upon admission is insensitive to the emotional state of the family and hence, is unethical by nature. ... However, so as to ensure that the consent given by patients for the diagnostic samples to be stored in the Tumour Bank and used for subsequent research is thoroughly informed we obtain *retrospective consent* and approach the parents at a later stage with specific information about the Tumour Bank as well as a further consent form for the collection of control tissue. The option to have the specimens withdrawn from the Bank is offered at this point.⁶⁸

18.66 A number of research institutions keep research participants informed about ongoing research activities. The value of this process was highlighted in consultations with research organisations.⁶⁹ Clearly, it is desirable for research participants who have consented to unspecified future research to be kept informed about the nature of the research that is eventually conducted, should they wish to know more about it. The Children's Hospital at Westmead Tumour Bank referred in its submission to a system of 'continual information flow' to the parents, guardians and patients within the hospital.

64 Ibid.

65 WA Research Tissue Network, *WA Research Tissue Network Consent Form MR 730.8* (2002).

66 Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

67 Ibid.

68 Ibid.

69 Children's Cancer Research Institute, *Consultation*, Perth, 4 December 2001; Queensland Institute of Medical Research, *Consultation*, Brisbane, 13 December 2001; Peter MacCallum Cancer Institute, *Consultation*, Melbourne, 23 October 2002; University of Tasmania, *Consultation*, Hobart, 27 November 2001; Sir Charles Gairdner Hospital, *Consultation*, Perth, 3 December 2001; Institute of Molecular Bio-Science, *Consultation*, Brisbane, 12 December 2001.

We believe that people who donate tissue for research expect that research be done on the specimens, even if it is unspecified at the time of consenting. Hence, the Newsletter is the means by which we continually inform the oncology parents what research is taking place as well as reminding them of the agreement conditions under which we use the tumour tissue.⁷⁰

Is existing regulation adequate?

18.67 The existing regulation of human genetic research databases is limited. Although the operation of human genetic research databases is covered to some extent by information and health privacy legislation, the Human Tissue Acts and ethical guidelines, there are significant gaps.⁷¹ Overwhelmingly, submissions agreed that the current regulation of human genetic research databases is inadequate and requires reform.⁷²

18.68 A major gap is the absence of rules governing the collection, storage, use, transfer or disclosure of genetic samples and information. For example, the *Privacy Act* and most other state and territory information and health privacy legislation do not cover genetic samples. Information that has been de-identified in such a way as to fall outside the definition of ‘personal information’ is also excluded.⁷³

18.69 In any case, the distinct ethical and privacy issues raised by the large-scale and long-term storage and use of linked genetic samples and information may not be addressed adequately simply through the application of information and health privacy legislation. The operation of research databases may require specific regulation.

18.70 The existing ethical principles and guidance set out in the National Statement do not deal with some of the distinct privacy and ethical issues raised by human genetic research databases. There are, for example, no specific provisions to ensure that major collections of genetic samples and information are subject to appropriate governance and accountability. The National Statement provides insufficient guidance on mechanisms for obtaining consent to unspecified future research, particularly in the context of standing research databases.

Options for reform

18.71 The Inquiry has considered a number of options for regulating human genetic research databases, including:

⁷⁰ Children’s Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

⁷¹ Ibid.

⁷² For example, Familial Cancer Centre — Royal Melbourne Hospital, *Submission G249*, 20 December 2002; R Muller, *Submission G197*, 27 November 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002. Compare Queensland Institute of Medical Research, *Submission G190*, 26 November 2002.

⁷³ For example, *Privacy Act 1988* (Cth) s 6(1).

- the inclusion of new provisions in the National Statement;
- a licensing or registration scheme; and
- the use of a gene trustee system to protect the privacy of samples and information stored in databases.⁷⁴

18.72 These options are not mutually exclusive, and could be adopted as a regulatory ‘package’ if this would most effectively address the privacy and ethical concerns about the operation of human genetic research databases.

New provisions in the National Statement

18.73 The National Statement lays down ethical guidelines that help to regulate the operation of human genetic research databases. However, some specific issues that arise from the establishment and operation of large-scale research databases should be addressed by new provisions in the National Statement.

18.74 In DP 66 the Inquiry proposed the inclusion of a new chapter in the National Statement to provide further ethical guidance on the operation of human genetic research databases, notably in relation to consent to unspecified future research.⁷⁵ However, there is a range of other matters relating to the operation and governance of human genetic research databases that might usefully be dealt with in the National Statement, such as ethics review where external researchers are seeking access to samples and information stored on the database; guidance on the use of independent intermediaries to hold codes linking genetic samples or information with identifiers; and governance structures.

18.75 Submissions expressed support for the proposed expansion of the National Statement to include a new chapter covering human genetic research databases.⁷⁶ The Androgen Insensitivity Syndrome Support Group Australia stated that:

Amending the NHMRC national statement to provide ethical standards for the operation of human genetic research databases would provide a valuable ethical baseline against which the operation of human research genetic databases could be measured. Already used by HRECs in such a way, the national statement would then encompass all steps of the research process.⁷⁷

74 The Inquiry has also considered whether the Human Genetics Commission of Australia should have a role in regulating research databases.

75 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 15–1.

76 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

77 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

18.76 The Department of Human Services South Australia noted that because of the sensitivity of samples and information held in research databases, ‘it is important that ethical guidance on their operation is formalised’. The Department suggested that ‘[t]his will ensure that each database follows protocol and abides by the guidelines set down by the National Statement with regard to privacy and data access’.⁷⁸

18.77 The Centre for Law and Genetics emphasised that a new chapter of the National Statement was ‘particularly desirable to provide guidance on the issue of consent to future research using samples and information stored in human genetic research databases’.⁷⁹

18.78 Another submission made the point that new provisions in the National Statement would avoid the need to develop legislative measures to regulate human genetic research databases.⁸⁰ Others explicitly rejected the imposition of legislative controls on the operation of databases.⁸¹ Genetic Health Services Victoria cautioned that ‘the potential benefit to the community from such research must be encouraged and not hampered by unrealistic and even unethical requirements for consent’.⁸²

Licensing or registration of research databases

18.79 In DP 66, the Inquiry asked whether human genetic research databases should be subject to a licensing or registration scheme.⁸³ A licensing scheme would require all human genetic research databases maintained in Australia to obtain a licence to operate. Organisations maintaining databases would be subject both to the general rules applying to licence-holders, as well as any specific conditions of the licence itself. Licence conditions could be used to impose specific limitations on how databases are maintained, and could be adapted for different types of databases.

18.80 An alternative, ‘light touch’ approach would be to implement a system of registration. A registration scheme could be modelled on the existing processes for the registration of HRECs with the Australian Health Ethics Committee (AHEC). Operators of human genetic research databases (or their HREC) could be required to report to AHEC on their activities, including on their consent procedures and on the nature of research undertaken using the database during the reporting period. AHEC could have authority to audit the activities of research databases to ensure compliance with the National Statement. In addition, the National Statement might be amended to state that an HREC should not approve research using unregistered genetic databases.

78 Department of Human Services South Australia, *Submission G288*, 23 December 2002. See also Department of Health Western Australia, *Submission G271*, 23 December 2002.

79 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

80 Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

81 Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

82 Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

83 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Question 15–1.

18.81 Most submissions agreed upon the need for more scrutiny and oversight of the operation and use of human genetic research databases. Some were satisfied with either scheme—licensing or registration—whichever would most effectively promote the protection of privacy.⁸⁴ Of the remaining submissions in favour of an oversight scheme, registration was preferred to licensing⁸⁵ because it was seen as a less onerous and costly method of regulation.⁸⁶

18.82 Some submissions did not support either licensing or registration and expressed concern about the practical implications of such a reform for medical research. The Children’s Cancer Institute Australia asserted that

the requirement to register any database which contains “gene information” would be costly and in-effective. Moreover, such a regulatory system would be unworkable in the research setting.⁸⁷

18.83 QIMR strongly opposed any idea that research databases should be subject to a licensing or registration scheme.⁸⁸ In relation to its own database, QIMR stated that:

The nature of research is ... a fluid and rapidly permuting process. To require documentation of the [database] resource, and the activities using it would require almost as many staff again and would impose an unsupportable burden on our research. The academics would simply move to theoretical work and simulation studies, away from real diseases, and staff supporting the databases would lose their jobs.

Our belief is that the use of genetic databases in a research setting is perfectly well regulated by the existing HRECs, to which we have to submit requests for any new use, or variation in existing use. Approval from our HREC, at least, is far from a pushover, since we frequently have to submit amendments in response to critiques from them. While we strongly believe that HRECs should have national accreditation, we also believe that monitoring use of genetic databases should remain the responsibility of local HRECs who are familiar with the context of research, and not be devolved to a remote regulator.⁸⁹

84 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; R Muller, *Submission G197*, 27 November 2002; Familial Cancer Centre — Royal Melbourne Hospital, *Submission G249*, 20 December 2002.

85 Australian Biospecimen Network, *Submission G238*, 19 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002.

86 For example, Department of Health Western Australia, *Submission G271*, 23 December 2002.

87 Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

88 Queensland Institute of Medical Research, *Consultation*, Brisbane, 11 November 2002; Queensland Institute of Medical Research, *Submission G190*, 26 November 2002.

89 Queensland Institute of Medical Research, *Submission G190*, 26 November 2002.

18.84 The Inquiry asked what conditions should attach to licensing or registration of human genetic research databases if such a system were introduced.⁹⁰ Annual reporting of database activities and regular audits were suggested in some submissions.⁹¹ Transparency and accountability were regarded as necessary to promote best practice.⁹² Jennifer Fleming made the point that regulatory controls should also respect the confidentiality of researchers and their intellectual property.⁹³ The Centre for Law and Genetics recommended that minimum registration requirements be imposed. These could include

undertakings to be bound by the NPPs or IPPs and to restrict access to the database to projects approved by properly constituted HRECs in compliance with the National Statement.⁹⁴

18.85 The use of sanctions and the revocation of licences or registration were regarded by a number of submissions as an appropriate means of ensuring compliance with prescribed operational requirements.⁹⁵

18.86 A particular issue noted in submissions was the need to define clearly what constitutes a ‘human genetic research database’ for the purposes of a registration or licensing scheme.⁹⁶ For example, the Centre for Genetics Education noted that it is sometimes unclear when a clinical genetic database that is used for research becomes a research database.⁹⁷

Oversight of human genetic research database regulation

18.87 A licensing or registration scheme would require administration and oversight by an appropriate body. Oversight functions could be undertaken by the NHMRC, through AHEC, or be left entirely to HRECs.

18.88 Submissions expressed differing views on the appropriate body to oversee a licensing or registration scheme. Some submissions saw AHEC as the appropriate body to which database operators should report, or proposed that AHEC should have a role

90 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Question 15–1.

91 R Muller, *Submission G197*, 27 November 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

92 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; J Fleming, *Submission G241*, 20 December 2002.

93 J Fleming, *Submission G241*, 20 December 2002.

94 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

95 Ibid; Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

96 Centre for Genetics Education, *Submission G232*, 18 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Familial Cancer Centre — Royal Melbourne Hospital, *Submission G249*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

97 Centre for Genetics Education, *Submission G232*, 18 December 2002.

in auditing the operation of databases.⁹⁸ Other submissions suggested that the primary responsibility for monitoring the operation of research databases should rest with HRECs.⁹⁹ The NHMRC Research Committee reasoned that

it is likely that the most effective regulatory bodies will be those established in close proximity to databases, with the capacity to interact regularly with the database coordinators on matters of importance.¹⁰⁰

18.89 The NHMRC Research Committee expressed the view that

supervision of ethics practised through reporting mechanisms that allow scrutiny, would be more likely to achieve the outcome desired than those involving onerous barriers to achieving registration, or barriers that interfered with sample data collection.¹⁰¹

18.90 Some submissions considered that it would be appropriate for the Human Genetics Commission of Australia (HGCA) to have some regulatory role with regard to databases.¹⁰² For example, the Cancer Council of Victoria stated that:

All research collections of human DNA samples should be registered with a national authority with oversight by the proposed HGCA. National regulation is preferable to a State based approach because of issues relating to privacy legislation variations in some States. Also genetic research databases have the potential to be forced to deal with new technology leading to issues not considered at the establishment of these databases. Further, a national approach is preferable because families often extend across multiple states and consistency is essential for proper handling of all databases, and for making it easier for families to understand conditions under which data is held. In theory, one national body should handle such issues more efficiently.¹⁰³

18.91 Others made the point that the HGCA, as described in Chapter 5, is intended to be an advisory body, not a regulator. Consequently, a number of submissions doubted that it would be appropriate for the HGCA to have a role in directly regulating

98 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; J Fleming, *Submission G241*, 20 December 2002; R Muller, *Submission G197*, 27 November 2002.

99 Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Queensland Institute of Medical Research, *Submission G190*, 26 November 2002.

100 National Health and Medical Research Council Research Committee, *Submission G262*, 20 December 2002.

101 Ibid.

102 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Familial Cancer Centre — Royal Melbourne Hospital, *Submission G249*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

103 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

databases,¹⁰⁴ while accepting that the HGCA might, for example, ‘be responsible for monitoring issues common to all databases and to report accordingly’.¹⁰⁵

Inquiry’s views

18.92 In the Inquiry’s view, further regulation is required to protect the privacy of genetic samples and information held in human genetic research databases. The Inquiry has concluded that reform in keeping with the present framework for the ethical conduct of research, centred on the National Statement and on review of research proposals by HRECs, is preferable to the introduction of new legislative constraints on the operation of research databases.

18.93 However, as noted above, the ethical principles and existing guidance set out in the National Statement do not adequately deal with some of the distinct privacy and ethical issues raised by human genetic research databases. The inclusion of new provisions in the National Statement to deal specifically with the operation of human genetic research databases could address these issues. The National Statement already includes specific chapters on the use of human tissue samples and human genetic research.¹⁰⁶ A new chapter, or new provisions in existing chapters, could also be developed to deal with human genetic research databases.

18.94 While it will be for the NHMRC to develop new provisions in the National Statement, the Inquiry recommends that these provisions include specific guidance on obtaining consent to unspecified future research. In drafting such provisions, the NHMRC should balance the needs of researchers with the need to protect the privacy of individuals’ genetic information and promote public trust in research.¹⁰⁷ The forms and processes for seeking consent to unspecified future research used by the PMCI Tissue Bank, the Western Australia Research Tissue Network and the Children’s Hospital at Westmead Tumour Bank, discussed above, may provide useful models.

18.95 Under the new provisions of the National Statement recommended by the Inquiry, the NHMRC would have oversight of the ethical operation of human genetic research databases and would take advantage of existing regulatory structures and reporting mechanisms.

18.96 However, it is also the case that regulation of human genetic research databases under the National Statement would be subject to the existing limitations in relation to enforcing compliance. Given the existing and likely future extent of private sector research using human genetic research databases, new mechanisms may be

104 Centre for Law and Genetics, *Submission G255*, 21 December 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; J Fleming, *Submission G241*, 20 December 2002; Queensland Government, *Submission G274*, 18 December 2002.

105 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

106 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra Ch 15–16.

107 The NHMRC and AHEC have statutory obligations to consult the community before issuing ethical guidelines, including new provisions of the National Statement: See *National Health and Medical Research Council Act 1992* (Cth) ss 8, 13–15.

required to achieve compliance with the National Statement. These issues are addressed in Chapter 14.

18.97 The Inquiry has concluded that a system for the registration of human genetic research databases is capable of providing greater transparency and accountability in the operation and use of such databases, without subjecting institutions to onerous compliance costs. Registration will oblige institutions to identify and regularise the research collections that they currently maintain and to ensure that the operation of these collections is subject to appropriate governance structures and proper institutional and HREC scrutiny.

18.98 Registration would mean that, for the first time, there will be comprehensive information available to AHEC, and possibly to the public, about the number and type of research databases, and the kinds of research being conducted using the samples and information they contain.¹⁰⁸ Such transparency may become increasingly important in allaying public concerns about the privacy, ethical and other implications of the continuing development of research databases. Registration will also provide AHEC with information necessary to enable it to properly advise the NHMRC, and provide guidance to HRECs, on ethical issues relating to the operation of research databases.

18.99 For these reasons, the Inquiry recommends that the new provisions of the National Statement should incorporate a requirement for institutions to register their human genetic research databases with the NHMRC. In addition to the general registration requirement, the Inquiry considers that there should be requirements in relation to:

- the nomination of a database keeper/custodian¹⁰⁹ who will have clear responsibility for the day-to-day operation of the database;
- compliance with standards for the collection, use, storage, disclosure and transfer of genetic samples and genetic information held by the database;
- annual reporting to the institutional HREC and AHEC on database operations; and
- provision for audit of the database and its operations, on request by the institutional HREC and/or AHEC.

18.100 In addition, guidance could be provided on a range of other matters relating to the operation of human genetic research databases including:

- governance structures, including guidance on the appropriate relationships between the institution, database custodian and the institutional HREC;

108 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

109 Compare National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra [2.1(e)].

- ethical approval processes where external researchers seek access to samples and information stored on the database; and
- guidance on the appropriate use of independent intermediaries to hold codes linking genetic samples or information with identifiers (see also below).

18.101 The constraints of the Terms of Reference mean that the recommendations made in this Report are generally directed to the protection of human genetic information, rather than to other forms of personal information. However, the Inquiry recognises that there may be good reason to consider extending the reforms recommended below to all research databases that contain identifiable bodily samples or personal information.

Recommendation 18–1. The National Health and Medical Research Council (NHMRC), as part of its review of the *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement) in the 2003–2005 triennium, should amend the National Statement to provide ethical guidance on the establishment, governance and operation of human genetic research databases. The amendments (whether by means of a new chapter or otherwise) should include specific guidance on obtaining consent to unspecified future research. (See also Recommendation 15–4.)

Recommendation 18–2. The NHMRC should establish and administer a public register of human genetic research databases. The National Statement, as revised in accordance with Recommendation 18–1, should establish conditions of registration and provide that no genetic research under the National Statement can be conducted using information from a database unless it is duly registered.

The gene trustee

18.102 The Inquiry suggests that the new provisions of the National Statement should provide guidance, among other things, on the appropriate use of independent intermediaries to hold codes linking genetic samples or information with identifiers. The concept of a ‘gene trustee’, which was referred to in Chapter 16, is relevant in this context.

18.103 The use of a gene trustee could be required, in appropriate situations, as a condition of the registration of a human genetic research database, or as a stand alone reform. A gene trustee system might be able to operate with a single body administering registration and acting as trustee. Persons or bodies taking on the role of gene trustee could also be subject to some system of scrutiny to ensure their integrity and independence from research organisations.

18.104 The model discussed here is based on the gene trustee approach developed for the Sydney Tay-Sachs disease screening program within the Jewish community as outlined in a submission made by the Institute of Community Genetics.¹¹⁰ The scheme is based around an independent third party, which controls information used to identify data and samples held within a database. The gene trustee acts as an intermediary between the persons maintaining the database and the individuals who supply their tissue and information.

18.105 Consent to donation of samples and information is given using a three-part consent form. The donor fills out Part A with his or her personal details and provides consent to the storage and use of the samples and information. Part A is marked with a unique personal identification number (PIN) and a unique database control identification number. Part B of the form contains only the database control identification number while Part C contains only the PIN.

18.106 Part A is then separated from the consent form and held by the gene trustee. Samples and information, along with Part B, are supplied to the persons maintaining the database and are stored using the database identification number as the only identifier. The donor of the samples and information is given Part C of the consent form, containing the PIN and contact details of the gene trustee.

18.107 Only the gene trustee can link the donor's personal information with any samples or information held by the database. If further consent to research is required, the holders of the database must pass the request to the gene trustee who will then contact the individual. Re-consent to future use could be obtained through the trustee, which would hold contact information for donors and details of the consent they have given. Destruction of the linking information held by the gene trustee (that is, Part A of the form) will permanently de-identify all samples and information held on a person in the database.

18.108 The gene trustee could act as a central body that holds linking information from materials held in numerous databases. Alternately, large databases could be required to establish their own gene trustee system. In either case, gene trustees would have to handle linking information in accordance with regulatory requirements.

18.109 The value of this approach lies in the separation of any identifying information from all sensitive data and material held in a database. No matter who obtains access to this material, they will be unable to identify it without contacting the gene trustee, who will be bound not to release any identifying information without the consent of the individual.

18.110 There are, however, a number of problems with this approach. It would present significant administrative costs, particularly where re-consent was sought for numerous samples. It also does not take account of the means by which many research

110 Institute of Community Genetics, *Submission G156*, 19 April 2002. The details of the scheme are elaborated in L Burnett and others, 'The 'Gene Trustee': A universal identification system that ensures privacy and confidentiality for human genetic databases' (2003) 10 *Journal of Law and Medicine* (in press).

subjects are contacted and asked to donate tissues and information. Often this occurs in a clinical context, where sufferers of a genetic condition are identified by their treating clinician and asked if they would like to participate in research. In other instances, researchers seek out participants through genetic counselling services and clinics. There may not be the initial degree of separation between the donor and researcher that would be necessary to make the gene trustee system workable in all situations.

18.111 The system may have greater value where large research databases are established for broad purposes, like UK Biobank or the Estonian database. The system may also be appropriate where commercial organisations develop databases from patient samples and information acquired from hospitals—a practice that is becoming common in the United States.¹¹¹

Submissions and consultations

18.112 DP 66 asked whether the use of a gene trustee should be a condition of the licensing or registration of databases. Considerable support for the use of a gene trustee system was expressed in submissions.¹¹² The OFPC maintained its support for ‘the adoption of practical mechanisms for the protection of an individual’s privacy, such as a gene trustee’ and commented that such a trustee

offers...an effective, transparent and accountable form of protecting an individual’s health information against the risks inherent in the establishment and operation of large databases.¹¹³

18.113 Criticism of the gene trustee system focused on costs.¹¹⁴ These concerns were raised both by supporters of the system as well as those who opposed it. The gene trustee system was regarded as unsuitable for smaller and more informal databases. The Human Genetics Society of Australasia (HGSA) submitted that the gene trustee system may be too complex and cumbersome for some smaller databases.¹¹⁵ However, the Children’s Hospital at Westmead Tumour Bank stated:

111 See GenomeWeb Staff Reporter, *New Tissue Repository Firm to Close Microarray, IP Collaborations in 6 Months*, GenomeWeb, <www.genomeweb.com/articles/view.asp?Article=2002627173511>, 27 June 2002.

112 Australian Biospecimen Network, *Submission G238*, 19 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; D Robins, *Submission G181*, 29 October 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Queensland Government, *Submission G274*, 18 December 2002; Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002.

113 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

114 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Familial Cancer Centre — Royal Melbourne Hospital, *Submission G249*, 20 December 2002.

115 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

Many institutional tumour banks and tissue repositories that have been established, including our own, are well placed to take on the custodial 'gene trustee' role with regards to the use of human tissue in genetic research.¹¹⁶

18.114 Not all human genetic research databases hold sensitive linked information, and hence only some may require an intermediary to protect privacy.¹¹⁷ One solution, suggested by the Institute of Actuaries of Australia, is to allow HRECs to decide when the use of a gene trustee is necessary.¹¹⁸

Inquiry's views

18.115 The Inquiry has concluded that use of an independent intermediary (such as a gene trustee) is an effective method of protecting the privacy of samples and information held in human genetic research databases. The system works well by maintaining the privacy of samples and information held in databases, while still allowing donors to be contacted if necessary. It avoids the problematic aspects of anonymisation and ensures that anyone who obtains access to samples and information will be unable to re-identify them without the authorisation of the gene trustee.

18.116 The Inquiry recognises that the use of an independent intermediary will not be necessary or practicable in all circumstances. For this reason, the Inquiry recommends that the new provisions of the National Statement provide guidance on when its use will be appropriate, rather than requiring all research databases to establish such a mechanism.

18.117 Where human genetic research databases do not have an established an independent intermediary, it may nevertheless be desirable for HRECs to make the use of such a system a condition of approving research, and the National Statement should provide guidance to HRECs on this issue (see also Recommendation 16–1). In this way, all genetic research involving the use of databases may be made subject to a gene trustee system if that is necessary to protect privacy in the particular circumstances of the research.

Recommendation 18–3. The NHMRC, in revising the National Statement in accordance with Recommendation 18–1, should provide guidance on the circumstances in which the use of an independent intermediary is to be a condition of: (a) registration of a human genetic research database; or (b) approval by an Human Research Ethics Committee of research involving a human genetic research database.

116 Children's Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

117 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

118 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

Secondary uses of research databases

18.118 Individuals who donate genetic samples and information to human genetic research databases do so with the aim of aiding medical researchers in their endeavours to improve their understanding of disease and to develop new treatments. In some instances, others may seek to obtain access to these samples and information for different purposes. These could include law enforcement and forensic purposes, disaster victim identification, paternity testing or to aid diagnosis of an inherited condition.¹¹⁹

18.119 DP 66 asked whether legislation is needed to govern the disclosure, for law enforcement purposes, of genetic samples and information held in human genetic research databases. Such legislation might also extend to other secondary uses of samples and information.

18.120 In the United Kingdom, the Human Genetics Commission (HGC) has recommended that genetic databases established for health research should not be used for any other purpose and that this should be put beyond doubt, by legislation if necessary.¹²⁰ The HGC referred, in particular, to the ‘pressure for allowing police access for purposes of identification’.¹²¹

18.121 In Australia, the *Privacy Act* allows personal information to be disclosed without consent where the disclosure is reasonably necessary for the enforcement of the criminal law or of a law imposing a pecuniary penalty, or for the protection of the public revenue.¹²² Some state and territory information privacy legislation contains similar provisions.¹²³ These provisions are permissive in that they permit agencies or organisations that hold personal information to disclose it without breaching privacy legislation, but do not require disclosure.

18.122 Disclosure of genetic samples and information for purposes other than those for which they were collected constitutes an interference with privacy. In donating samples and information to a research database, an individual consents to their use for research, not for other purposes. The use of samples and information for unrelated purposes might be a violation of the consent agreement between the database operator and the donor. Over time, people may be less willing to donate samples for research if they fear the samples will be used for other purposes.

119 For example, to diagnose whether someone has a particular inherited mutation, it is necessary to know the exact mutation and the disease symptoms it causes. A sample taken from a family member who has displayed the symptoms associated with the disease is needed to determine the exact mutation that causes the disease in that family. Other family members are then tested to see whether they carry this mutation, which will determine whether they also will develop the disease.

120 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 105.

121 *Ibid.*, 105.

122 *Privacy Act 1988* (Cth) IPP 11.1(e); NPP 2.1(h).

123 For example, *Health Records Act 2001* (Vic) Health Privacy Principle 2.2(i), (j); *Privacy and Personal Information Protection Act 1998* (NSW) ss 18, 23(5).

18.123 The need for tighter controls on the use, disclosure and transfer of genetic samples and information held in research databases was recognised in many submissions. A number supported the introduction of legislation to deal specifically with this issue.¹²⁴ Some suggested that the same principles as those laid down in the *Privacy Act* should be applied.¹²⁵

18.124 The HGSA noted that there are practical problems with the use of genetic samples and information held in research databases for law enforcement or other purposes. For example, establishing that samples and information are accurately identified might not always be possible. Police may also be unlikely to seek access to samples and information in databases, as they would first need to be aware they had been donated. Samples and information that have been de-identified would be unlikely to be of use to them.¹²⁶

18.125 Recognising the need to protect the privacy of samples and information donated to databases, the Australian Biospecimen Network stated:

It is essential that ... legislation be established to ensure that the present situation in which a person can obtain a court order to obtain genetic samples or information be more tightly restricted. This is essential for future compliance of consent as individuals may withhold consent if they feel that others may invade their genetic privacy for a purpose different to that [for] which the 'information' was given. Such protection would be consistent with the *Privacy Act's* spirit of collecting information only for the purpose [for] which consent was given, and for example demanding a sample for paternity testing would be a clear violation of the spirit of the consent to keep the sample/information only for medical research.¹²⁷

18.126 The OFPC also noted that public health should not be prejudiced by individuals' concern that the health information they provide may be disclosed for insurance, employment, or law enforcement purposes.¹²⁸

Inquiry's views

18.127 There is a need to develop a more formalised approach to requests for transfer or disclosure of samples and information held in databases to ensure that these are released only in appropriate circumstances. The Inquiry recommends that the Australian Health Ministers' Advisory Council, in consultation with state and territory Attorney-General's Departments and police services, the HGCA and the NHMRC, should develop rules for transfer or disclosure of samples and information held in human genetic research databases for law enforcement purposes.

124 Department of Human Services South Australia, *Submission G288*, 23 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Australian Biospecimen Network, *Submission G238*, 19 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; J Fleming, *Submission G241*, 20 December 2002.

125 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

126 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

127 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

128 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

18.128 A national policy developed in this way will be able to define how requests for the transfer or disclosure of samples and information to third parties are to be dealt with. The rules will help avoid the current inconsistencies of practice whereby database administrators develop their own policies on disclosure for law enforcement purposes, sometimes through memoranda of understanding with law enforcement agencies.¹²⁹ Police access to stored samples is also considered from the perspective of law enforcement in Chapter 41.

Recommendation 18–4. The Australian Health Ministers’ Advisory Council, in consultation with state and territory Attorney-General’s Departments and police services, the Human Genetics Commission of Australia and the NHMRC, should develop nationally consistent rules governing the disclosure, for law enforcement purposes, of genetic samples and information held in human genetic research databases. These rules should provide for disclosure only: (a) with the consent of the sampled person or a person authorised to consent on his or her behalf; or (b) pursuant to a court order.

129 As is the case with the Memorandum of Understanding between the New South Wales Newborn Screening Programme and the New South Wales Police (see Ch 19).

19. Human Tissue Collections

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Introduction

19.1 This chapter is concerned with the collection, storage, use and disclosure of, and access to, genetic samples and information held in tissue collections maintained mainly by hospitals or pathology laboratories.¹ These collections, which have not been collected primarily for use in research, are referred to in this chapter as 'human tissue collections' to distinguish them from human genetic research databases (see Chapter 18).

19.2 Tissue samples are collected and stored in many medical contexts. Pathology laboratories receive blood and other tissue for testing and analysis. Blood banks take donations of blood for transfusion. Hospitals remove tumours during surgery, which may be stored for later examination. Tissue repositories 'bank' bone marrow, skin and blood for use in treatment. Organs and tissues are also removed and retained following autopsy. In each of these cases, samples containing human genetic material are stored.

1 In this chapter the term 'tissue' includes blood and other bodily samples.

19.3 In some instances, tissue is stored only for a short time and then disposed of. Other tissue is archived and retained for a significant period of time, whether for teaching purposes, to meet quality assurance requirements, or to allow future re-testing. Retention with no single, defined purpose also occurs, as is the case with newborn screening cards created as part of newborn screening programs.

19.4 Developments in genetic technology have made it possible to perform almost all available genetic tests on stored tissue, provided it has been adequately preserved. Large amounts of potentially sensitive information about the person from whom the tissue was taken and his or her family can be obtained from archived tissue.

19.5 As a result, archived collections of preserved human tissue have taken on a new importance as both a scientific and economic resource, particularly where they can be linked with an individual's medical history. In particular, they are invaluable research resources for studies into the genetic causes of disease. Genetic testing of stored tissue samples has potential uses in other contexts, including criminal or police investigations, as evidence in court proceedings and for parentage or other kinship testing. These secondary uses raise issues of privacy and consent.

19.6 This chapter examines aspects of the regulatory framework that applies to human tissue collections and, in particular, to newborn screening cards. The Inquiry has concluded that there is a need for nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to newborn screening cards and other human tissue collections.²

Types of human tissue collections

19.7 Human tissue collections are established and maintained in three main contexts: by pathology laboratories in conducting medical testing, by health authorities in the conduct of newborn screening programs, and by tissue banks that hold stores of tissue for transplantation and therapeutic use. Each type of human tissue collection is described below.

Archived pathology samples

19.8 Pathology laboratories receive samples of blood, body fluids and tissue from doctors and hospitals for testing. Such tissue may have been removed during surgery or via a medical examination. Pathologists examine test results to aid in determining the cause of a condition and how it should be treated.³

19.9 The pathologist's findings and analysis are reported to the doctor who ordered the tests. However, the laboratory will retain any remaining portion of the sample once testing has been completed. These samples are archived and may be kept

2 Ch 24 examines the development and implementation of population genetic screening programs, including newborn screening programs.

3 For example, a general practitioner who suspects a patient has anaemia may take a blood sample and send it to a pathology laboratory for analysis. The sample will be tested for haemoglobin levels and examined under a microscope to look for signs of iron deficiency like abnormally small cell size.

for some time. Samples are retained to fulfil laboratory accreditation requirements⁴ or to meet laboratory standards. There may also be legal reasons for keeping samples, and a laboratory may wish to be able to re-test samples to confirm results at a later stage. Collections of pathology samples are, therefore, comprised of residual tissue taken primarily for therapeutic and diagnostic purposes. Although samples are generally coded when a pathology laboratory receives them, the laboratory retains the means to unite a sample with its identifying information.

19.10 Samples are stored in a variety of forms. Some are fresh-frozen and kept refrigerated. Others, like sections of tissue, are embedded in paraffin to enable slides to be made from cut sections. These blocks of tissue can be stored for a long time without significant deterioration.

19.11 Samples are sometimes disposed of once they have no further use. Usually this involves steam-heating samples to melt containers and destroy the contents. The heat at which they are melted is sufficient to denature any DNA contained in samples, while the process prevents separation of individual containers. Once cooled, destroyed samples are generally deep-buried.

Newborn screening cards

19.12 In hospitals around Australia, blood has been taken from newborns, and the sample identified and stored, for the last 30 years. Between two and five days after birth, virtually all children are screened for a variety of conditions. A small sample of blood is taken and placed on a piece of blotting paper. The child's name, date of birth, hospital of birth and birth weight, and the mother's name and date of birth, are also recorded on the card.⁵

19.13 These cards are referred to in this Report as 'newborn screening cards'. The cards are also commonly known as 'Guthrie cards', a name derived from the scientific test once used to detect phenylketonuria. They are now more correctly referred to as newborn screening cards because the Guthrie test is no longer universally used and newborn screening also includes other tests.

19.14 Samples are obtained with verbal parental consent. Parents are supplied with an information leaflet detailing the screening program, the tests that are performed and (in some cases) the fact that cards are stored and may be used or disclosed for other purposes. In South Australia and New South Wales parents are usually also notified that the cards, once de-identified, might be used in approved research.⁶ Screening program staff are generally required to discuss the contents of this leaflet with parents to ensure they have fully understood the information provided before giving consent. However, as discussed below and in Chapter 24, doubts have been expressed about whether this constitutes proper consent.

4 See Ch 11.

5 A newborn screening card is reproduced at the end of Ch 24.

6 NSW Health Department, *Test to Protect Your Baby* (2000), NSW Health Department, Sydney; South Australian Neonatal Screening Centre, *Screening Tests for Your New Baby* (2002), Adelaide.

19.15 Cards are stored for different periods in each State and Territory, ranging from two years in Western Australia to much longer periods in most other jurisdictions. Cards are retained for quality assurance and laboratory auditing purposes and for retrospective diagnosis. Retrospective diagnosis can directly assist some children.⁷ In addition, where a child has died, stored cards can be used to confirm the mutation for a genetic condition, enabling carrier testing of family members.⁸ Cards are also stored to help testing services to devise and trial new screening tests.

19.16 In 1999, the Senate Legal and Constitutional Legislation Committee referred to newborn screening card collections as 'inadvertent DNA sample banks', noting that identified blood samples containing genetic material from almost all people under the age of 28 are currently stored in most States and Territories.⁹

Tissue banks

19.17 A variety of tissue banks have been established in Australia to hold donor tissues for transplantation and therapeutic uses. Unlike the human genetic research databases discussed in Chapter 18 (which are also sometimes referred to as tissue banks), these banks have not been created for research purposes. They may, however, house collections of human tissue that might be used for research or other secondary purposes.

19.18 For example, the Perth Bone and Tissue Bank receives donations of bone for transplantation and therapeutic use. Bones are received from living individuals (who, for example, donate bone removed during hip replacement surgery) and from deceased individuals. Sections of bone obtained from failed grafts are sometimes used in research projects by the Bone Bank.¹⁰ The Australian Red Cross Blood Service collects blood donations for whole-blood transfusions and the manufacture of therapeutic products. On rare occasions, the Australian Red Cross Ethics Committee considers requests for access to blood samples for research purposes.¹¹

19.19 Human genetic samples are also banked commercially in the form of cord blood. Firms such as Cryosite, based in Sydney, store blood taken from the umbilical cords of newborn infants. Parents pay for storage so that they will be able to access the blood, which can be used in the treatment of some diseases, if their child becomes ill.¹²

7 For example, in relation to testing for congenital cytomegalovirus (CMV) infection: Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

8 For example, in relation to testing for Duchenne muscular dystrophy: *Ibid.*

9 Senate Legal and Constitutional Legislation Committee, *Provisions of the Genetic Privacy and Non-discrimination Bill 1998*, The Parliament of Australia, <www.aph.gov.au/senate/committee/legcon_ctte/genetic/index.htm>, 21 August 2002, 3.

10 A Cowie (Perth Bone and Tissue Bank), *Correspondence*, 2 July 2002. Specific consent for research use is always sought from donors.

11 Australian Red Cross Ethics Committee, *Submission G292*, 6 January 2003. Ethics approval to date has always required individual consent, frequently restricted access to de-identified or coded samples only, and has never involved a waiver of consent.

12 D Teutsch, 'Born to be Insured', *Sun Herald* (Sydney), 30 June 2002, 27.

19.20 Some genetic registers also maintain banks of genetic samples. These may be used for presymptomatic diagnosis and carrier detection. Where a disorder has been identified but a reliable molecular test has not yet been developed, samples may be banked for testing once a reliable test becomes available.

Secondary uses of human tissue collections

19.21 Human tissue collections have a variety of potential secondary uses, including use in medical research, law enforcement and parentage testing.¹³ Submissions expressed concerns about possible secondary uses of human tissue collections,¹⁴ including those that may occur without the knowledge of the person from whom the samples were taken.¹⁵ Submissions also raised concerns about the storage of genetic samples taken from children who cannot consent on their own behalf.¹⁶

19.22 Individuals may not be aware that their genetic samples or information have been retained and could be used again for other purposes. The Neurofibromatosis Association of Australia submitted the results of a survey of its members, which showed that few people were aware that their tissue could have been stored after it was removed for diagnosis or treatment; nor were they aware of the uses to which it might be put.¹⁷ Women's Health Victoria submitted that:

The thought that parts of our human tissue may be stored in a bank or laboratory, for some other research, without our knowledge, is abhorrent to some and feels like a violation.¹⁸

19.23 Secondary uses of genetic samples and information collected for other purposes raise privacy and consent issues. However, some secondary uses are closely related to the primary purpose of collection and could reasonably be expected. For example, the re-testing of pathology samples for quality assurance purposes is a closely related secondary purpose.

Research value

In the post genome sequencing era, the ability to easily and extensively access the staggering amount of medical and biological information locked up within tissue archives is paramount—molecular pathology could play a pivotal role in unlocking the bases of multiple disease types, including infectious diseases, cancer and developmental disorders.¹⁹

13 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

14 *Confidential Submission G023CON*, 29 November 2001; UnitingCare NSW & ACT, *Submission G052*, 14 January 2002; Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

15 National Council of Women Australia, *Submission G095*, 31 January 2002.

16 For example, T Struzina, *Submission G016*, 25 November 2001.

17 Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002.

18 Womens Health Victoria, *Submission G076*, 3 January 2002. See also *Confidential Submission G051CON*, 14 January 2002.

19 *Confidential Submission G034CON*, 14 January 2002.

19.24 As is the case with human genetic research databases (see Chapter 18), human tissue collections have great value as research tools. Samples are generally collected in a clinical setting and so will often have identifying information attached to them, which enables them to be linked to medical records.

19.25 This is one reason that newborn screening cards and archived pathology samples are very useful research tools. Newborn screening cards have been used, for example, to look for genetic mutations that may cause Sudden Infant Death Syndrome (SIDS). Researchers have been able to test genetic samples taken from the cards of children known to have died of SIDS. Such targeted research would have been impossible if the cards were not identified.²⁰ Other uses, such as in epidemiological studies to determine the prevalence of a genetic mutation in the population, can be conducted on de-identified samples.

19.26 Human tissue collections, particularly newborn screening cards, have significant potential value for population studies, including those that may help government and health system administrators to plan for the future health needs of the Australian population.²¹ The collections can be used to study the interaction of genetic and environmental factors in disease over time, to examine the causes of genetic diseases, and to locate genetic mutations. Through such research, new diagnostic tools and treatments can be developed, which will have economic as well as medical value.

19.27 Other human tissue collections can be used to confirm diagnoses to facilitate research on familial disorders:

[T]here are circumstances where it is essential to review, for clinical and research purposes, the pathology on samples collected for diagnosis. For example, in many large molecular-epidemiology studies, confirmation of diagnosis is critical for substantiating apparent familial histories of specific cancers.²²

19.28 Tissue collections created over long periods of time have unique value for researchers. Access to historical collections can enable research that otherwise would have been logistically very difficult and time-consuming, if not impossible, given the problems of establishing such databases from scratch. Many studies require access to large sample sets. In some cases collections created for research are not sufficiently large but they can be supplemented with archived material.

19.29 This may apply, for example, to some research uses of material held in human tissue collections, where the dividing line between clinical care and research may be unclear.²³ In its submission, the Australian Academy of Science observed that research into the causes of disease is an intrinsic part of diagnosis by pathologists and that the clinical and research activities of pathologists cannot be regarded in isolation: in trying to diagnose a condition, the pathologist is also searching for the most

20 B Williamson, *Consultation*, 5 September 2001.

21 Advisory Committee members, *Advisory Committee meeting*, 31 May 2002.

22 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

23 Australian Academy of Science, *Submission G097*, 21 January 2002.

effective cure.²⁴ A crossover between pathological diagnosis and ongoing research is regarded as good clinical practice, particularly as tests on historical samples can help find treatments for family members.²⁵

19.30 The Australian Academy of Science also emphasised the need to recognise the research uses of archived pathology material and submitted that research utilising these materials should be encouraged in the interests of the community. The Academy argued that restricting access to samples would be a major impediment to research.²⁶

19.31 The Peter MacCallum Cancer Institute stated that it is ‘hard to overstate the importance of controlled access to archival material’,²⁷ a perspective emphasised by other submissions.²⁸

Although the movement to prospectively collected consented material will ultimately result in an increase in the number of samples available for research, this will take time. ... the increasing sophistication of (molecular) sub-typing of cancer requires large sample sets to achieve statistical power. Hence, there will continue to be a need to supplement tissue bank sets with archival material for the foreseeable future.²⁹

19.32 The research value of human tissue collections may depend on how the tissue has been stored. It has been said that proper long-term storage of newborn screening cards for research purposes would be complex and expensive.³⁰

Forensic use of human tissue collections

19.33 Genetic samples stored in human tissue collections may be sought for forensic and law enforcement purposes.³¹ Newborn screening cards in particular may provide a useful resource in some criminal investigations. The cards contain sufficient DNA to enable matching with other samples collected in an investigation, and they have reasonably reliable identifying information attached to them. Newborn screening cards may be of use in identifying human remains and in providing samples for DNA profiling to obtain matches to suspects where no other samples (apart from those collected from a crime scene) can be obtained.

19.34 Newborn screening cards are not regularly used for law enforcement purposes. However, police do sometimes seek disclosure of newborn screening cards and hospitals do sometimes comply with requests for disclosure. For example, in New

24 Ibid.

25 Australian Society for Medical Research, *Submission G124*, 18 March 2002; Children’s Hospital at Westmead, *Consultation*, Sydney, 19 November 2002. The Children’s Hospital at Westmead Tumour Bank agreed: Children’s Hospital at Westmead Tumour Bank, *Submission G276*, 17 December 2002.

26 Australian Academy of Science, *Submission G097*, 21 January 2002.

27 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

28 Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002.

29 Peter MacCallum Cancer Institute, *Submission G071*, 7 January 2002.

30 WA Genetics Council, *Consultation*, Perth, 28 October 2002.

31 *Confidential Submission G023CON*, 29 November 2001; New South Wales Legal Aid Commission, *Submission G087*, 21 January 2002.

South Wales more than a dozen requests for cards have been made since 1996, mainly for use in murder inquiries and to identify bodies. Parental consent was given in almost all cases, and cards were released in all but a few cases.³² Where consent is not given, disclosure may be obtained by police under a court order, most often a search warrant issued by a magistrate.

19.35 Public concerns about police access to samples came to prominence when the Western Australian police took possession of some newborn screening cards held by the Princess Margaret Hospital in Perth. Although the police had a warrant to take possession of the cards, some parents were nevertheless disturbed by this action and sought the return of their children's cards. These events are said to have led to a decrease in participation in the screening program.³³

19.36 In the course of the Inquiry, the issue of law enforcement use of samples held in human tissue collections has been raised mainly in relation to newborn screening cards. Identified pathology samples and other stored tissue might also be used for law enforcement purposes. However, the increased use of police powers to compel the provision of DNA samples under forensic procedures legislation may decrease the need for police to access newborn screening cards or other stored samples, at least where samples may be obtained from living persons.³⁴

Parentage and other kinship testing

19.37 In some instances, individuals may seek access to stored genetic samples for parentage and other kinship testing,³⁵ sometimes in the context of court proceedings.³⁶ Some newborn screening programs in Australia have been approached for samples for parentage testing. In Western Australia a newborn screening card was released for parentage testing, with the consent of both parents.³⁷

Regulation of human tissue collections

19.38 The collection, storage, use and disclosure of genetic samples and information held in human tissue collections are regulated by a mixture of legislation, guidelines and standards. These include:

- the legislative framework for the protection of information and health privacy based on the *Privacy Act 1988* (Cth) (*Privacy Act*) and similar state and territory legislation (see Chapters 7 and 8);

32 NSW Privacy Commissioner, *Correspondence*, 4 June 2002.

33 Department of Clinical Biochemistry at Princess Margaret Hospital in Perth, *Consultation*, Perth, 3 December 2001.

34 The content of forensic procedures legislation is covered in Part J.

35 Parentage and other kinship testing is discussed in Ch 35.

36 *Roche v Douglas* [2000] 22 WAR 331.

37 Department of Clinical Biochemistry at Princess Margaret Hospital in Perth, *Consultation*, Perth, 3 December 2001.

- state and territory Human Tissue Acts, which require consent for the donation of human tissues for research;³⁸
- standards and guidelines released by the National Pathology Accreditation Advisory Council (NPAAC), which apply to pathology laboratories accredited in Australia;
- relevant ethical guidelines and policy statements; and
- government health authority policies on the storage, use and disclosure of newborn screening cards.

19.39 The regulation of tissue collections also takes place against the background principles of ownership of samples, which provide that laboratories have a right to possession of preserved samples (see Chapter 20). Newborn screening cards may be regarded as a medical record. It is therefore possible that, according to common law and equitable principles, newborn screening cards are owned by the health authorities that create them.³⁹

Information and health privacy legislation

19.40 Chapter 7 summarised the legislative framework for the protection of information and health privacy based on the *Privacy Act* and similar state and territory legislation.⁴⁰ Chapter 8 discussed the application of this legislation to genetic samples.

19.41 Personal information held by private sector organisations, such as private sector pathology laboratories, is governed by the *Privacy Act*. Where it is held by public hospitals, pathology-related personal information will be subject to state and territory information and health privacy legislation. Newborn screening cards are held by state public hospitals⁴¹ and screening is done in public laboratories. The cards are

38 *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1979* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1985* (Tas); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplant Act 1982* (WA); *Transplantation and Anatomy Act 1978* (ACT); *Human Tissue Transplant Act 1979* (NT).

39 *Breen v Williams* (1996) 186 CLR 71. See also C Lawson and R Smith, 'Protecting Genetic Materials and Genetic Information: A Case Study of Guthrie Cards in Victoria' (2001) 9 *Journal of Law and Medicine* 215, 223; L Skene, 'Access to and Ownership of Blood Samples for Genetic Tests: Guthrie Spots' (1997) 5(2) *Journal of Law and Medicine* 137, 140. However, *Breen v Williams* did not deal with records that included blood or tissue samples.

40 *Privacy and Personal Information Protection Act 1998* (NSW); *Health Records and Information Privacy Act 2002* (NSW); *Health Records Act 2001* (Vic); *Information Privacy Act 2000* (Vic); *Health Records (Privacy and Access) Act 1997* (ACT).

41 Victorian cards are held by the Murdoch Childrens Research Institute, part of the Royal Children's Hospital; Tasmanian, South Australian and some Northern Territory cards are held by the Women's and Children's Hospital in Adelaide; New South Wales and Australian Capital Territory cards are held by the Westmead Children's Hospital; Western Australian cards are held by the Princess Margaret Hospital in Perth; Queensland cards are stored at the Prince Charles Hospital.

therefore subject to state and territory information and health privacy legislation, where it exists.⁴²

19.42 As discussed in Chapter 8, newborn screening cards are clearly covered by the *Privacy Act* or similar state and territory information and health privacy legislation because they are ‘personal information’—the cards contain identifying information, such as date of birth, as well as the blood sample, and the card is a ‘record’ for the purposes of the Act.⁴³ However, the exclusion of samples from the coverage of most privacy legislation poses a problem because individual privacy rights in respect of samples, including those held in human tissue collections, may not be asserted by the individuals from whom the samples were taken. This issue is discussed in Chapter 8.

Human Tissue Acts

19.43 Human genetic samples collected for diagnostic or therapeutic purposes are expressly excluded from the ambit of the Human Tissue Acts.⁴⁴ Samples collected for pathology purposes and newborn screening cards are thus not covered by the Acts. Tissues, body parts and organs donated to tissue banks are covered by the Acts under provisions outlined in Chapter 18. Under these provisions consent to donate tissue for one purpose, for example transplantation, probably does not constitute consent to other uses, such as research.

National Pathology Accreditation Advisory Council guidelines

19.44 NPAAC produces guidelines and standards for the accreditation of pathology laboratories and services. The quality assurance and accreditation standards that regulate genetic testing laboratories are discussed in Chapter 11.

19.45 The minimum standards for pathology laboratory practice in Australia are set out in NPAAC’s *Standards for Pathology Laboratories*.⁴⁵ These standards state that confidentiality of patient information must be a primary consideration in the operation of a pathology service and that pathology laboratories should have policies and procedures to maintain ethical standards of laboratory practice and to ensure human tissue samples are treated with due respect.⁴⁶

42 At present, only New South Wales, Victoria and the Australian Capital Territory have privacy legislation which protects health information held in their public sectors: *Privacy and Personal Information Protection Act 1998* (NSW); *Health Records and Information Privacy Act 2002* (NSW); *Health Records Act 2001* (Vic); *Privacy Act 1988* (Cth).

43 *Privacy Act 1988* (Cth) s 6(1). However, once a section of the blood spot is punched out and physically detached from card on which it was stored, the *Privacy Act* no longer governs it. See Ch 8.

44 *Human Tissue Act 1983* (NSW) s 34(1)(a)–(b) and cognate state and territory legislation.

45 National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (2002), Department of Health and Ageing, Canberra.

46 *Ibid*, Standard 1.

19.46 In addition, the NPAAC *Guidelines for the Retention of Laboratory and Diagnostic Material*⁴⁷ prescribe minimum time periods for retaining different types of diagnostic material. For example, in anatomical pathology, different retention periods are prescribed for frozen section tissue (20 years); frozen tissue blocks for immunofluorescence studies (three months); unblocked tissue removed at surgery (one month) and unblocked tissue retained at autopsy (generally up to three months after autopsy).⁴⁸ Importantly, newborn screening cards are to be retained until the child reaches the age of 25 years.⁴⁹

Other relevant guidelines and policy statements

19.47 A range of ethical and other guidelines has been issued by bodies, including the National Health and Medical Research Council (NHMRC), the Human Genetics Society of Australasia (HGSA) and specialist medical colleges. These are relevant to the collection, storage, use and disclosure of, and access to, genetic samples and information held in human tissue collections, including newborn screening cards.

19.48 The NHMRC *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement)⁵⁰ generally requires consent to the collection and use in research of human tissue, including samples that have been collected and stored after clinical procedures. As discussed in Chapter 15, these consent requirements can be waived by a Human Research Ethics Committee (HREC).

19.49 The HGSA's *Guidelines for Human DNA Banking* cover DNA stored to meet the future service needs of families affected by inherited genetic disorders, including for confirmation of diagnosis at the molecular level, presymptomatic diagnosis, carrier detection and prenatal diagnosis by family linkage analysis.⁵¹ The guidelines list matters to be discussed with individuals prior to obtaining consent to store their DNA, which include the uses to which the DNA will be put, the minimum period for which the DNA will be stored, and the conditions under which the DNA can be used for research.⁵²

19.50 In relation to newborn screening, the HGSA and the Royal Australasian College of Physicians have released the *Policy Statement on the Retention, Storage and Use of Sample Cards from Newborn Screening Programs*.⁵³ The Policy Statement provides that cards should be stored in a manner appropriate to their intended future

47 National Pathology Accreditation Advisory Council, *Guidelines for the Retention of Laboratory Records and Diagnostic Material* (2002), Department of Health and Ageing, Canberra.

48 Ibid [2] (Anatomical Pathology).

49 Ibid [9.9]. The 25 years is calculated on the basis of seven years (the notional statute of limitations) from the time a child becomes an adult at 18 years: B O'Connor (National Pathology Accreditation Advisory Council), *Correspondence*, 20 May 2002.

50 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

51 Human Genetics Society of Australasia, *Guidelines for Human DNA Banking*, Guidelines, 1 July 1990 [2.1].

52 Ibid [2.6].

53 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *Policy Statement on Newborn Screening*, 1 June 1999.

uses and notes that the primary purpose of retaining cards is to enable confirmation of test results.⁵⁴ The policy statement recommends particular consent or approval requirements for the use or disclosure of newborn screening cards for various purposes:

- Screening program development: No consent is required where cards have been anonymised; however, ethics committee approval is necessary if cards are to be used outside the screening laboratory or where the samples remain.
- New health information about the person from whom the sample was obtained is generated: The approval must specify who should be informed of abnormal test results, what they will be told and the nature of any follow-up that may be necessary.⁵⁵
- Individual requests: Test information should not be released to anyone other than the person whose blood is on the card, or parents where that person is a minor. Release should be discouraged if the person is still living and could give another sample. If a card is returned, the family should be encouraged to retain it or, if they will not, then destruction of the card at an agreed time is to be sought.⁵⁶
- Requests from health professionals: A card may be released to determine cause of death or to gain genetic information for family reasons, but only where parental consent has been given.⁵⁷
- Research studies: Cards may be used in research studies where approval has been obtained from both an ethics committee and the screening program advisory committee (if one exists). Research should be conducted in accordance with NHMRC guidelines and follow any other requirements placed on it by the screening program committee.⁵⁸
- Coronial and forensic inquiries: Cards may be released with parental or next-of-kin permission or in accordance with a legal requirement.

19.51 The Royal College of Pathologists of Australasia (RCPA) has released a policy statement on the secondary use of human tissue samples collected for diagnostic purposes.⁵⁹ The RCPA policy statement includes guidance on the re-use of samples for education, research, commercial, medico-legal, insurance, employment and legal determinations.⁶⁰ For educational and quality review purposes, consent is generally considered to be an implied part of patient care while the individual is still a patient at

54 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *Policy Statement on the Retention and Use of Sample Cards From Newborn Screening Programs*, 7 June 2001 [4.1], [4.3.1].

55 *Ibid* [4.3.2].

56 *Ibid* [4.3.3].

57 *Ibid* [4.3.4].

58 *Ibid* [4.3.5].

59 Royal College of Pathologists of Australasia, *Provision of Second Opinions with Particular Reference to Histopathology Cytology and Specimens for Morphological Examination*, 1 March 2001.

60 *Ibid* [3.3]–[3.6].

the institution where the review is taking place.⁶¹ Research use of pathology samples requires consent unless HREC approval for a waiver of consent has been obtained in accordance with the National Statement. Otherwise samples should be used only for the purpose for which consent was obtained.⁶²

19.52 The RCPA policy statement provides that samples should not be released for insurance, employment or medico-legal purposes unless a subpoena or warrant has been obtained or where the individual has consented to the sample's use in this manner. Once the use has been completed, samples should be returned to the laboratory where they were held.⁶³ This would cover police access to samples. Generally an individual's request for the return of his or her own material will be refused.⁶⁴

Health authority policies on newborn screening cards

19.53 Health authorities have developed their own policies in relation to newborn screening cards, which differ from each other in significant respects. For example, the Western Australian Newborn Screening Program has different policies to those adopted in most other Australian programs, most notably the requirement that cards be destroyed after two years.⁶⁵ In other States and Territories cards are generally stored for much longer periods.

19.54 In Western Australia cards may be released for research after the removal of identifiers, although demographic information may be provided if necessary. The program's policy provides that only one blood spot from each card will be released and the research project should aim to make some contribution to public or family health, or to the goals of public health screening.⁶⁶ Requests from individuals and medical practitioners are considered by a Review Panel.⁶⁷ Cards are not to be disclosed for law enforcement purposes unless the consent of next-of-kin or a subpoena has been obtained, and even then release will be restricted unless the requesting party can demonstrate that no alternative sample can be found. The Review Panel also considers requests for disclosure for law enforcement purposes.⁶⁸ South Australia has similar policies; however, the Executive Director of Medicine of the hospital holding the cards deals with requests for the disclosure of cards for law enforcement purposes.⁶⁹

61 Ibid [3.3].

62 Ibid [3.4].

63 Ibid [3.5], [3.6].

64 Ibid [3.7].

65 WA Newborn Screening Program, *Policy for the Retention, Storage and Use of Dried Blood Spot Samples Collected by the Western Australian Newborn Screening Program*, 1 February 2002 [5.2.4].

66 Ibid [5.3.1.5].

67 Ibid [5.3.1.3], [5.3.1.4].

68 Ibid [5.3.1.6].

69 Women's and Children's Hospital, *Protocol for the Retention, Storage and Use of Neonatal Screening Cards (Guthrie Cards) collected by the South Australian Newborn Screening Program* (2002) [B 1]. South Australia and Western Australia retain cards from Tasmanian and the Northern Territory newborn screening respectively.

19.55 In Victoria, Genetic Health Services Victoria deals with the release of cards on a case-by-case basis. Cards are sometimes released for research and, rarely, to police. Genetic Health Services Victoria has signed a Memorandum of Understanding (MOU) with the Victorian Police to provide a framework for police access to newborn screening cards. Under the MOU, the parties agree that access to cards should be sought through a court order⁷⁰ and that the information contained on the card is confidential.⁷¹

19.56 In New South Wales, the Children's Hospital at Westmead releases cards only with written parental consent and authorisation from the Director of the Screening Program.⁷² Cards may be released to the police under the terms of an MOU between the New South Wales Health Department and the New South Wales Police. The New South Wales MOU differs from the Victorian agreement in setting out a protocol for the release of cards where there is no court order. The MOU provides that the Department will disclose cards for the purpose of identifying human remains or where the Police possess a forensic sample suspected to come from a victim of a crime, taken from the scene of the crime, where the victim cannot be located. New South Wales Police must make reasonable efforts to secure the consent in writing of the next-of-kin of the person whose sample is requested, unless it would be impractical to do so or would compromise an ongoing investigation. The protocol is not intended to affect disclosure as required by law, such as by a search warrant.⁷³

19.57 In New South Wales, cards may also be released for certain forms of research, including special studies for families where the child is deceased; other special studies for families; forensic studies as directed by the Director-General of Health or the Department Head of the Newborn Screening laboratory; and (when de-identified) for epidemiological studies approved by the hospital HREC.⁷⁴

Issues and problems

19.58 A number of ethical and privacy concerns are raised by aspects of the handling of genetic samples and information held in human tissue collections. The samples and information held in these collections have often been obtained for one purpose—such as pathology testing, newborn screening or other therapeutic use—and not explicitly for other, secondary purposes. Issues of consent and privacy arise when it is sought to use or disclose the samples or information for purposes for which consent may not have been obtained. Consent to the storage of samples in human tissue collections is discussed below, followed by discussion about the use of such samples in research and law enforcement.

70 Usually a search warrant obtained by application to a magistrate under s 465 of the *Crimes Act 1958* (Vic): Victoria Police, *Consultation*, Sydney, 7 March 2003.

71 Victoria Police and Genetics Health Services Victoria, *Memorandum of Understanding*, 23 January 2003.

72 B Wilcken, *Consultation*, 6 June 2001; The Children's Hospital at Westmead, *NSW Newborn Screening Programme*, <www.chw.edu.au/prof/services/newborn/>, 20 February 2003.

73 New South Wales Commissioner of Police and New South Wales Health Department, *Memorandum of Understanding*, 17 April 2002.

74 Children's Hospital at Westmead, *Sample Manual — NSW Newborn Screening Manual* (2000), Sydney.

Consent to storage

19.59 When samples are taken for pathology testing, consent to *testing* is obtained but consent to *storage* of the sample is not always explicit. Once stored, samples may later be used or disclosed for other purposes, some of which are related to the primary purpose of collection and others of which are not.⁷⁵

19.60 Many people are unaware that tissue samples collected for pathology tests are subsequently stored and might be used for other purposes. Almost all stored tissue samples will contain DNA, and some secondary uses may reveal information about an individual's genetic status, health, parentage or kinship. The familial nature of genetic information means that information may also be revealed about members of his or her family, such as the mother named on a newborn screening card.⁷⁶

19.61 Submissions confirmed the general lack of community awareness about the storage of genetic samples.⁷⁷ For example, the Association of Genetic Support of Australasia stated:

The community have little knowledge or understanding about the existence of stored samples from the Newborn Screening programs. Those who are aware are very concerned. This is a grey area which must be urgently resolved and community awareness increased.⁷⁸

19.62 When members of the public do become aware that samples are stored, they may become concerned about possible use or disclosure of the cards.⁷⁹ These concerns sometimes lead individuals to request their own or their child's newborn screening card from the hospitals where the cards are stored.⁸⁰

19.63 Submissions emphasised the need to obtain informed consent for the storage of tissue samples beyond the period for which it is necessary to retain them to carry out the primary purpose for which they were collected.⁸¹ The Centre for Genetics Education submitted that written consent should be obtained for retention of samples

75 The Office of the Health Services Commissioner Victoria and the HGSA submitted that quality assurance should be considered as a primary purpose for which cards are collected and retained: Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

76 C Lawson and R Smith, 'Protecting Genetic Materials and Genetic Information: A Case Study of Guthrie Cards in Victoria' (2001) 9 *Journal of Law and Medicine* 215, 217.

77 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002.

78 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

79 Centre for Genetics Education, *Submission G232*, 18 December 2002.

80 It has been claimed that one such request was refused by the administrators of the NSW Newborn Screening Program on the basis of the program's obligation under NPAAC guidelines, New South Wales Department of Health directives and the *Privacy and Personal Information Act 1998* (NSW): *Confidential Submission G103CON*, 25 February 2002. Cards have been returned on request in other cases: See C Lawson and R Smith, 'Protecting Genetic Materials and Genetic Information: A Case Study of Guthrie Cards in Victoria' (2001) 9 *Journal of Law and Medicine* 215.

81 Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002.

beyond that necessary for quality assurance.⁸² Other submissions argued that newborn screening cards should not be retained beyond the time necessary to conduct diagnostic tests.⁸³ The New South Wales Legal Aid Commission questioned the legitimacy of retaining newborn screening cards beyond this time, noting that:

The retention of the cards after the testing has been completed conflicts with section 12 of the New South Wales *Privacy and Personal Information Protection Act 1998*, which provides that personal information must be kept for no longer than is necessary for the purposes for which the information may lawfully be used.⁸⁴

19.64 In contrast, Genetic Health Services Victoria noted that the ability to make retrospective diagnostic use of newborn screening cards was a reason to retain cards.⁸⁵

19.65 The storage and subsequent use of newborn screening cards is a particularly sensitive example of the consent problems raised by human tissue collections. The primary purpose of screening—the early diagnosis of treatable conditions—needs to be balanced with the need to obtain fully informed consent to storage and other possible uses. Obtaining explicit consent to the storage of newborn screening cards may have the unwanted effect of decreasing participation in screening programs. Parents might refuse to allow their child to be screened for possibly life-threatening but preventable disorders because they do not wish their child’s sample to be disclosed to the police later in life or used in research.

19.66 Submissions voiced concerns about the implications of requiring consent to the storage of newborn screening cards. The New South Wales Legal Aid Commission stated:

Up until the present time there has been almost universal participation in the new born screening program, as a result of the desire of parents to provide the best possible care for their babies. If the community becomes aware that the Guthrie cards are retained indefinitely, and becomes concerned about other uses to which the blood samples and information may be put, there may be some drop off in participation in this vital preventative health care program.⁸⁶

19.67 Genetic Health Services Victoria remarked that ‘[m]entioning potential forensic use at such a vulnerable time could lead to refusal which would be of potential harm to the baby’.⁸⁷ The Inquiry has heard that some parents have refused newborn screening tests because of concerns about storage of samples.⁸⁸

82 Centre for Genetics Education, *Submission G232*, 18 December 2002.

83 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002. See also National Legal Aid, *Submission G314*, 19 February 2003.

84 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

85 Genetic Health Services Victoria, *Submission G211*, 28 November 2002; National Legal Aid, *Submission G314*, 19 February 2003.

86 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

87 Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

88 Children’s Hospital at Westmead, *Consultation*, Sydney, 19 November 2002.

Research use

19.68 Unlike samples collected as part of a research project, future research use of pathology samples and newborn screening cards is often not envisaged at the time they are collected. In many cases, consent to future research will not have been obtained, although some general consent forms do notify the sample provider that the tissue might be used in research. In some cases the lack of clear guidance on the use of samples, particularly newborn screening cards, for research may discourage researchers from seeking to use the cards.⁸⁹

19.69 Submissions expressed support for the use of human tissue collections for research. For example, the Victorian Breast Cancer Laboratory submitted:

Ethical research using [tissue banking and studies on archival specimens] is to be lauded, as quantum improvements in clinical outcome will result from studies that profile genetic changes in cancer and other disease states. ... Public debate needs to be encouraged so that there is an appreciation of the importance of tissue samples for basic and clinical research. ... The vast majority of patients are only too pleased to allow excess tissue obtained as part of their routine clinical care (that would otherwise be discarded) to be used for research.⁹⁰

19.70 The Association of Australian Medical Research Institutes and the HGSA stated that research should be allowed to continue on historical collections of tissue samples provided the samples are de-identified, the samples were collected in the course of diagnosis or treatment of diseases, and approval from an HREC is obtained.⁹¹ The Institute of Actuaries of Australia supported long-term retention of cards for research:

Guthrie cards collections provide unique and potentially invaluable genetic databases of children born in Australia in the last thirty years. Long term retention is essential.⁹²

19.71 The Department of Human Services South Australia suggested an approach that could be appropriate for research using newborn screening cards. The Department suggested that cards be in two sections, with the blood spot divided from the information on the card by perforations. The information could then be easily detached once there is no longer a need to retain the card for re-testing and quality assurance.⁹³

89 WA Genetics Council, *Consultation*, Perth, 28 October 2002.

90 Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002.

91 Association of Australian Medical Research Institutes, *Submission G007*, 27 April 2001; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

92 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

93 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

19.72 Many submissions emphasised the need to gain consent and ethics committee approval for research using stored samples.⁹⁴ However, requirements to seek new consent were considered by some to constitute an unjustifiable administrative burden. Dr Nikolajs Zeps submitted that obtaining consent for each and every use of archival tissue would be ‘a logistical problem that would effectively stymie the function of routine pathology’.⁹⁵ Some individuals from whom samples have already been taken may have died, or may be distressed at being reminded of a time when they were ill. On this issue, Dr Rosemary Balleine stated that ‘the propriety of contacting these patients in itself poses an ethical dilemma’.⁹⁶

19.73 Other submissions criticised the research use of stored samples without consent, even if de-identified. For example, the Androgen Insensitivity Syndrome Support Group Australia submitted that community attitudes frowned upon subsequent re-use for research purposes without specific consent.⁹⁷ They also submitted that use of samples stored previously without consent ‘must surely fail any reasonable ethical or moral test’.⁹⁸ Dr Trevor Kerr stated:

I would find it very disturbing if the state allowed unfettered access to material that once belonged to me, in order to give enormous profit to private organisations from products that are sold back to humanity.⁹⁹

19.74 Although de-identifying stored samples to allow their use in research does solve some problems, members of the National Public Health Partnership Genetics Working Group suggested that it might be unethical to de-identify cards if significant personal health information would be generated by the research, as de-identification prevents patients from being contacted if the research produces findings relevant to their health.¹⁰⁰ The Children’s Cancer Institute of Australia agreed, stating that samples should be coded where possible, rather than de-identified, to enable linkages to be made between clinical data and research outcomes.¹⁰¹

19.75 The HGSA summarised the options for dealing with consent to the use in research of tissue samples collected primarily for clinical purposes:

94 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Royal College of Pathologists of Australasia, *Submission G287*, 23 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

95 N Zeps, *Submission G047*, 14 January 2002.

96 R Balleine, *Submission G035*, 14 January 2002.

97 Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002.

98 *Ibid.*

99 T Kerr, *Submission G223*, 4 December 2002.

100 National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002.

101 Children’s Cancer Institute Australia, *Submission G221*, 29 November 2002.

- Allow the collection and storage of clinical samples for research without consent and require approval by an HREC if the sample is to be used subsequently for research. The HREC will require de-identification in nearly all cases to protect the privacy of the individuals whose genetic material is to be used.
- Require general consent for research use to be obtained at the time the tissue sample is obtained and require approval by an HREC if the sample is to be used subsequently for research. The HREC will require de-identification in nearly all cases.
- Require specific consent to be obtained from the individual each time a tissue sample is to be used for a research study, as well as approval of the research by an HREC.¹⁰²

19.76 The HGSA favoured the first option ‘as the benefits for research are considerable and the likelihood of harm to individuals is very small’. Obtaining informed consent every time a tissue sample is collected in routine hospital work was seen to be impractical.¹⁰³ The HGSA also noted that consent should later be sought from the patient, where possible.

19.77 In practice, consent to possible future research may be difficult to obtain in clinical settings. Dr Rosemary Balleine noted:

Pre-operative consent is neither practical in the context of busy clinical practice nor considerate of the patient's emotional well-being. Approaching patients post-operatively may be considered by some as intrusive, especially as researchers are frequently not the clinicians responsible for care of the patient. A reasonable approach may [be] to ask the attending clinician to obtain consent from the patient post-operatively, however clinicians may be disinclined to undertake this complicated and time-consuming process in support of research that they are not directly involved with.¹⁰⁴

19.78 Chapter 15 and Chapter 18 discussed procedures that are being developed within the medical research community for obtaining prospective consent for unspecified future research,¹⁰⁵ and recommended that the NHMRC provide new guidance to researchers about obtaining such consent.

19.79 Similar approaches might be taken at the time an individual is admitted to hospital, consents to a screening test for their child, or provides a pathology sample. For example, the Western Australia Research Tissue Network is developing a system where, upon admission to hospital, patients are asked to complete a one-page form in triplicate on which they indicate their consent or refusal to consent to research use of tissue samples taken in the course of treatment. One portion of this form is attached to

102 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

103 *Ibid.*

104 R Balleine, *Submission G035*, 14 January 2002.

105 These include the idea of ‘tick a box’ consent options—so that individuals can elect from a graduated set of consent options, including consent to unspecified future research: See Ch 15.

the patient's medical record and can be linked to any samples taken so that future researchers will be able ascertain whether consent to research was given.¹⁰⁶

Disclosure for law enforcement purposes

When parents of newborns consent to the blood test, the furthest thing from their mind is any measured judgement of the potential use by the state, through the police or otherwise, of their child's DNA.¹⁰⁷

19.80 Many submissions accepted that there are circumstances in which the police might reasonably require access to stored tissue samples, though most noted that these circumstances should be exceptional.¹⁰⁸ The Office of the Victorian Privacy Commissioner emphasised that samples should be disclosed only in the most serious cases as a last resort, and should not result in samples being added to a DNA database. The Office also submitted that disclosure of samples should require a court order and informed consent, although in some unusual cases it might be reasonable to waive the consent requirement.¹⁰⁹ Disclosure for the identification of human remains was regarded as more acceptable than for the identification of suspects.¹¹⁰

19.81 A few submissions suggested that disclosure of stored samples for law enforcement should not be allowed.¹¹¹ For example, the Department of Human Services South Australia stated that it does not disclose newborn screening cards to police as this is 'viewed as intrusive to privacy, especially considering that there is no informed consent given to such a use for Guthrie cards'. The Department also noted that there is usually another source of genetic material available in most circumstances where cards have been requested for this purpose.¹¹²

19.82 Some submissions favoured legislative constraints on the disclosure of samples for law enforcement purposes.¹¹³ For example, the Office of the Victorian Privacy Commissioner stated:

106 WA Research Tissue Network, *Consultation*, Perth, 28 October 2002.

107 Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003. Privacy Victoria agreed: Office of the Victorian Privacy Commissioner, *Submission G171*, 8 August 2002.

108 For example, Centre for Law and Genetics, *Submission G255*, 21 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002.

109 Office of the Victorian Privacy Commissioner, *Submission G171*, 8 August 2002. The Office of the Federal Privacy Commissioner agreed with these points: Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

110 For example, Centre for Genetics Education, *Submission G232*, 18 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

111 Department of Human Services South Australia, *Submission G288*, 23 December 2002; National Legal Aid, *Submission G314*, 19 February 2003.

112 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

113 Office of the Victorian Privacy Commissioner, *Submission G171*, 8 August 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; National Legal Aid, *Submission G314*, 19 February 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

The issue needs to be addressed transparently and by the appropriate authority, Parliament. ... It should be a matter for thorough public debate and considered parliamentary decision before ... collections of infant DNA or research volunteers' tissue are tapped for law enforcement purposes.¹¹⁴

19.83 Privacy NSW stated:

We consider that the disclosure of newborn screening cards for law enforcement should be regulated by law. In addition to protecting the interests of tissue donors, legal regulation is critical to secure public accountability and to promote public confidence in genetic screening programs and the banking of human tissue undertaken by hospitals and other medical institutions.¹¹⁵

19.84 The Office of the Federal Privacy Commissioner (OFPC) suggested that amendments could be made to the *Crimes Act 1914* (Cth) (*Crimes Act*) to provide legislative controls on disclosure.¹¹⁶ The OFPC highlighted perceived flaws in the use of MOUs to control disclosure of newborn screening cards in New South Wales and, in particular, the fact that the protocol was developed without public debate or parliamentary process and does not establish any accountability measures.¹¹⁷

19.85 Others suggested that legislation was unnecessary. The New South Wales Health Department and the New South Wales Police considered the MOU to be a sufficient response to existing concerns—although the New South Wales Police did not have objections to formalising the MOU through legislation.¹¹⁸

19.86 Other submissions suggested that legislation regulating, and legitimating, the use of newborn screening cards for law enforcement might discourage parents from allowing their children to be screened. The Office of the Victorian Privacy Commissioner stated:

Individuals may be less inclined to seek a genetic test or to provide a tissue sample if the genetic register or tissue bank becomes a pool of data into which police may routinely dip.¹¹⁹

19.87 Concern about disclosure of stored samples to police are related to broader concerns about police collecting samples outside the formal regulatory framework established under Part 1D of the *Crimes Act* (see Chapter 41). Seeking disclosure of stored samples, which is not specifically covered by these legislative provisions, could be a means of circumventing forensic procedures legislation. The NSW Legal Aid Commission observed:

114 Office of the Victorian Privacy Commissioner, *Submission G171*, 8 August 2002.

115 Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002.

116 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

117 Ibid. The OFPC also noted that the New South Wales MOU does not include any processes of review or limitations on its duration, leaving open the possibility of 'function creep'.

118 NSW Health Department, *Submission G303*, 13 January 2003; NSW Police Service, *Submission G306*, 22 January 2003.

119 Office of the Victorian Privacy Commissioner, *Submission G171*, 8 August 2002. The Western Australia Genetics Council agreed: WA Genetics Council, *Consultation*, Perth, 28 October 2002.

Apart from the general concern about the retention of this vast database of identified genetic material, and its use for purposes for which no consent has been given, it is a particular concern that law enforcement agencies could obtain access to this information, thereby bypassing the safeguards on collection of forensic information included in the forensic procedures legislation in the various jurisdictions.¹²⁰

19.88 In Chapter 41, the Inquiry recommends that the *Crimes Act* be amended to provide that, with the exception of crime scene samples, law enforcement officers may lawfully collect a genetic sample for law enforcement purposes only from the individual concerned under Part 1D of the *Crimes Act*, or with consent or a court order (see Recommendation 41–13).

Need for consistent policies and practices

19.89 In DP 66 the Inquiry proposed the development of nationally consistent policies and practices in relation to the collection, storage, use of and access to stored tissue samples.¹²¹ This proposal was well received and many submissions expressed support.¹²² The Commonwealth Department of Health and Ageing stated that:

Lack of clarity about sample retention periods and permissible secondary uses, such as for research and law enforcement, may undermine newborn genetic screening programs. For this reason, the Department supports the development and implementation of nationally consistent legislation and/or policies and practices governing the disclosure of genetic information and samples held on Guthrie cards.¹²³

19.90 Many submissions highlighted the need for community consultation on the issues surrounding the use of genetic samples and information held in human tissue collections, and for programs to increase public awareness.¹²⁴

120 New South Wales Legal Aid Commission, *Submission G282*, 24 December 2002.

121 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 16–1.

122 NSW Health Department, *Submission G303*, 13 January 2003; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; National Legal Aid, *Submission G314*, 19 February 2003; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; NSW Health Department, *Submission G303*, 13 January 2003; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

123 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

124 Centre for Genetics Education, *Submission G232*, 18 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002.

19.91 While the submissions supporting the proposal emphasised the desirability of a national response to the issues raised by human tissue collections,¹²⁵ there were also suggestions that this might slow the development, in particular jurisdictions, of improved policies and practices.¹²⁶

Inquiry's views

19.92 The handling of samples and information held in human tissue collections is regulated by a complex, fragmented and overlapping set of legislation and guidelines. In relation to *some* aspects of the handling of *some* genetic samples and information there is no clear need for further regulation because detailed rules have already been established and are observed.

19.93 An example is the current regulation of retention periods for pathology laboratory samples and records. As discussed above, NPAAC guidelines contain comprehensive and specific minimum requirements for retention of laboratory records and diagnostic materials, reflecting best laboratory practice.¹²⁷ Pathology laboratories must comply with NPAAC guidelines in order to be accredited and ongoing accreditation requires periodic assessment.¹²⁸ Laboratories also have to comply with federal, state or territory information and health privacy legislation. Such legislation generally requires that personal information, such as that in laboratory records, be destroyed or de-identified if it is no longer needed for the purpose for which it was collected.¹²⁹ In the circumstances of a complaint or investigation by a privacy regulator, the specific requirements of the NPAAC guidelines, which require retention of records for minimum periods primarily for review and confirmation of results, can be tested against more general legislative standards requiring destruction or de-identification when the records are no longer needed.

19.94 The implementation of many of the recommendations made elsewhere in this Report would strengthen the privacy protection and other regulation of genetic samples and information held in human tissue collections. For example, the Inquiry's recommendation that the Commonwealth, States and Territories enact new legislation to require laboratories to be accredited for genetic testing would ensure more complete compliance with NPAAC guidelines (see Chapter 11).

125 Genetic Support Council WA, *Submission G243*, 19 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002; National Legal Aid, *Submission G314*, 19 February 2003; Children's Cancer Institute Australia, *Submission G221*, 29 November 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

126 Children's Hospital at Westmead, *Consultation*, Sydney, 19 November 2002.

127 National Pathology Accreditation Advisory Council, *Guidelines for the Retention of Laboratory Records and Diagnostic Material* (2002), Department of Health and Ageing, Canberra.

128 See Ch 11.

129 For example, *Privacy Act 1988* (Cth) NPP 4.2; *Health Records Act 2001* (Vic), Health Privacy Principle 4.5.

19.95 Similarly, while the National Statement already governs the use in research of genetic samples and information held in human tissue collections, the Inquiry makes a range of recommendations intended to strengthen the protection provided by the National Statement. These include recommendations made in relation to mechanisms for achieving compliance with the National Statement, waiver of consent and obtaining consent to unspecified future human genetic research (see Chapters 14 and 15).

19.96 Importantly, while the handling of newborn screening cards is generally subject to Commonwealth, state and territory information and health privacy legislation, the Inquiry has recommended that new legislation should be enacted to ensure that minimum, legally enforceable, privacy standards apply to the handling of all genetic samples (see Chapter 8).

19.97 Certain aspects of the regulation of human tissue collections require further attention. In particular, submissions and consultations highlighted continuing professional and public concern about the long-term storage and secondary use, in research and law enforcement, of newborn screening cards. The Inquiry has concluded that these concerns require a regulatory response. Participation in newborn screening programs is vitally important for detecting certain treatable genetic conditions at an early stage and full participation in these programs should be encouraged. Lack of clarity about retention periods and permissible secondary uses may undermine newborn screening programs.

19.98 For this reason, the Inquiry recommends that Australian Health Ministers' Advisory Council (AHMAC) develop nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, newborn screening cards. These rules will need to be consistent with the minimum privacy standards set out in information and health privacy legislation, but should augment those legislative standards with more specific provisions.

19.99 As discussed above, a key area of concern relates to the disclosure of newborn screening cards for law enforcement purposes. Privacy legislation allows disclosure of personal information where it is reasonably necessary for the enforcement of the criminal law or of a law imposing a pecuniary penalty, or for the protection of the public revenue.¹³⁰ These provisions are permissive only—they permit agencies or organisations holding personal information to disclose it without breaching privacy legislation, but do not require disclosure. It is therefore open to those agencies or organisations holding newborn screening cards to restrict the circumstances in which they will disclose them. The MOUs developed in New South Wales and Victoria constitute an attempt to do so in those jurisdictions. However, there is no national consistency in the procedures by which police or other law enforcement agencies may obtain access to newborn screening cards.

19.100 The Inquiry recommends, in particular, that AHMAC, in consultation with state and territory Attorney-General's Departments and police services should develop nationally consistent rules governing disclosure of newborn screening cards for law

130 *Privacy Act 1988* (Cth) IPP 11.1(e); NPP 2.1(h).

enforcement purposes. These rules should provide for disclosure only with the consent of the person sampled or a person authorised to consent on his or her behalf, or pursuant to a court order, including a search warrant.

19.101 Another area of concern is in relation to the secondary use of newborn screening cards in research. While there have been some attempts to articulate policies on research use of these collections, the issue clearly requires further attention from state and territory health authorities, and the development of a national approach through AHMAC. There should be broad consultation to assess and define the potential research value of newborn screening card collections—for example, whether research use should be limited to epidemiological studies using de-identified samples. If collections of newborn screening cards are to be used commonly in research, health authorities need to review the consent processes they use, and may need to engage with their communities in a discussion about the acceptable research uses of newborn screening cards.

19.102 Newborn screening card collections constitute perhaps the best known and most extensive collections of human genetic samples. However, while newborn screening card collections are the subject of much of the detailed discussion in this chapter and were the focus of many of the concerns expressed in submissions and consultations, the need for nationally consistent rules in relation to other human tissue collections should be reviewed by AHMAC. The issues raised in relation to the secondary use in research and for law enforcement purposes also apply to other human tissue collections. The need for additional regulation of human tissue collections may depend in part on the implementation of recommendations made elsewhere in this Report—notably in relation to the Inquiry’s recommendation that new legislation be enacted to ensure that minimum, legally enforceable privacy standards apply to the handling of all genetic samples (see Chapter 8).

19.103 Accordingly, the Inquiry recommends that AHMAC, in consultation with the Human Genetics Commission of Australia (HGCA), the NHMRC and key professional bodies, should review the need for nationally consistent rules in relation to the collection, storage, use and disclosure of and access to other human tissue collections, including collections of pathology samples and banked tissue.

19.104 In developing these rules, it will be important to identify and balance the relevant ethical considerations, including those applying at individual and community levels. At the individual level, respect for the personal autonomy of parents and concern to benefit the health of newborn children can be in tension. In this context, the narrative experiences of families may be valuable. At a community level, the ethical considerations include the benefit of research and the enforcement of criminal law to the community. The development of these rules should be informed by an ethics of discussion.¹³¹

131 See the discussion of ‘narrative ethics’ in Ch 6.

Recommendation 19–1. The Australian Health Ministers’ Advisory Council (AHMAC), in consultation with the Human Genetics Commission of Australia (HGCA), the National Health and Medical Research Council (NHMRC) and key professional bodies, should develop nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, newborn screening cards. In particular, and in consultation with state and territory Attorney-General’s Departments and police services, AHMAC should develop nationally consistent rules governing disclosure of newborn screening cards for law enforcement purposes. These rules should provide for disclosure only: (a) with the consent of the person sampled or a person authorised to consent on his or her behalf; or (b) pursuant to a court order.

Recommendation 19–2. AHMAC, in consultation with the HGCA, the NHMRC and key professional bodies, should review the need for nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, other human tissue collections—including collections of pathology samples and banked tissue.

20. Ownership of Samples and the Human Tissue Acts

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Introduction

20.1 The existing regulatory framework to protect the privacy of human genetic information focuses on the rights of individuals to control the collection, use and disclosure of their genetic information. However, human tissue samples can also yield genetic information. The Inquiry considers that, in order to protect genetic privacy effectively, both samples and information must be adequately regulated. A uniform approach to the regulation of samples and information is preferable, to avoid complexity, inconsistency and further fragmentation of privacy laws.

20.2 In Chapter 8 the Inquiry recommended that the *Privacy Act 1988* (Cth) (*Privacy Act*), and similar state and territory health and information privacy legislation, should be amended to cover genetic samples, as well as the genetic information derived from them (Recommendation 8–2). Part of the reason for this recommendation was that if the *Privacy Act* does not cover genetic samples, there would be a major gap in the framework for protecting the privacy of the individuals from whom genetic samples are taken. Some other regime may have to be developed to fill the gap.

20.3 The Inquiry regards amendment of the *Privacy Act* as the most effective means of protecting genetic privacy in relation to samples. However this chapter considers two other possible approaches to regulating the handling of genetic samples and to providing the individuals from whom they are taken with greater control over what happens to the samples.

20.4 The chapter first discusses property rights in genetic samples and, in particular, whether the privacy of genetic samples and information could be more adequately protected by allowing any property rights, other than possession, to be exercised over genetic samples. The Inquiry considers that a property approach would be difficult to adopt, and that its drawbacks outweigh its benefits.

20.5 The chapter then examines whether the Human Tissue Acts should be amended to regulate comprehensively the collection, storage, use of, and access to, genetic samples. The Inquiry considers that amending the Human Tissue Acts would not adequately address the issue of sample privacy, although the Inquiry recognises the need for a general review of those Acts.

Ownership of human genetic samples

20.6 Reform of property law could provide individuals with the means to better protect the privacy of their genetic information. The exercise of information privacy rights and any property rights that may exist in human genetic samples need not conflict, in the same way that the right of access to medical records does not conflict with a medical practitioner's ownership of those records. The *Privacy Act* provides a legally enforceable right for patients to obtain access to their medical records held by private medical practitioners, notwithstanding that the records are the property of the medical practitioners who create and maintain them.¹

20.7 The idea of recognising property rights in human tissue, which includes the genetic material that may be extracted from almost all human cells, is not new. In the United States, property rights have been suggested as a possible means of protecting the privacy of genetic samples and information. For example, s 104(a) of the model Genetic Privacy Act (GPA) provides that 'an individually identifiable DNA sample is the property of the sample source'.² Patricia Roche, George Annas and Leonard Glantz, the authors of the GPA, explain that:

By establishing an individually identifiable sample as the property of the sample source, the GPA serves not only the interest of those who would want to maintain exclusive control over their DNA, but also enables those who desire to share or transfer such control to do so. This ability is particularly important to individuals who are concerned with preserving their own samples for the future use and benefit of relatives and descendants ... Owning one's own DNA sample allows transfer of control in accordance with property law principles.³

1 Compare *Breen v Williams* (1996) 186 CLR 71, which held that there was no general right of patient access to medical records at common law or in equity.

2 P Roche, L Glantz and G Annas, 'The Genetic Privacy Act: A Proposal for National Legislation' (1996) 31(1) *Jurimetrics* 1. In Australia, this would be known as a model Bill, since it has not passed through the legislative process.

3 G Annas, L Glantz and P Roche, 'Drafting the Genetic Privacy Act: Science, Policy and Practical Considerations' (1995) 23 *Journal of Law, Medicine and Ethics* 360, 363.

20.8 A version of the GPA, the *Genetic Confidentiality and Nondiscrimination Act* was introduced into the United States Congress in 1996. However, although it identified a DNA sample as property, it did not assign ownership of that property to anyone.⁴ A number of US states have enacted legislation similar to, or influenced by, the GPA.⁵

20.9 In 1995, Oregon became the first State in the United States to grant ownership rights in genetic samples to the individual from whom they were obtained, and to that person's children.⁶ However, the Oregon law was amended in 2001 to specify that genetic samples were *not* property, but that both genetic samples and information were private and must be protected.⁷ The amendments were made after the property approach was criticised as a disincentive to research—an argument that was also put to the Inquiry in the submissions and is discussed later in this chapter.⁸

20.10 At present, Australian legislation does not address the property status of genetic samples. Instead, the legal status of genetic samples is governed by the common law. However, the common law position on property rights in human tissue samples is not well developed, and there is no clear judicial statement on the issue. Reform of property law in this context is therefore difficult to achieve, particularly as the direction in which the common law develops cannot be dictated.

Legal status of genetic samples

20.11 The present state of the law suggests that human tissue samples are property in a limited sense, and in limited circumstances. The traditional position under the common law was that a human corpse could not be the subject of property rights. This rule gained support in a number of English cases and was generally accepted throughout the 19th century.⁹

4 No action was taken on the Act and a revised edition was introduced the following year: M Baram, 'The Laws of Genetics' (1997) 105 *Environmental Health Perspectives* 488.

5 P R Reilly, *State Show Keen Interest in Genetic Privacy*, Gene Letter, <www.genesage.com/professionals/geneletter/privacybills.html>, 7 February 2003.

6 *Oregon Genetic Privacy Act 1995* (US) § 4(1).

7 The provision was altered by §15 of Senate Bill 114 of 2001. Attempts were made to remove the property provision in 1999 through Senate Bill 937, but these measures failed. See also P Wentz, *Royal Flush*, *Williamette Week*, <www.wweek.com/html/leada033199.html>, 6 February 2003; A Onion, *Should You Own Your Own Genes?*, ABC News (USA), <abcnews.go.com/sections/scitech/DailyNews/geneprivacy010508.html>, 10 May 2001; Institute for Health Freedom, *Who Owns Your Genetic Information?*, <forhealthfreedom.org/Publications/Informed/WhoOwns.html>, 3 April 2001. The State of Georgia passed similar provisions, which have also subsequently been repealed.

8 N Zeps, *Submission G047*, 14 January 2002; Australian Academy of Science, *Submission G097*, 21 January 2002; Life Sciences Network, *Submission G129*, 19 March 2002.

9 The major cases are *R v Sharpe* (1857) 169 All ER 959; *R v Price* (1884) 12 QB 247 and *Williams v Williams* (1881) 20 ChD 659.

20.12 Since then, a number of cases have declared urine and blood samples to be capable of being stolen.¹⁰ These cases have been cited as support for the property status of samples, since only property can be the subject of theft.¹¹ Additionally, in 2000, the Supreme Court of Western Australia held that tissue samples were property within the meaning of a Supreme Court rule allowing inspection of ‘property’.¹²

20.13 Over the last century, the common law has shifted away from the original rule towards recognising limited ownership interests in one specific area—preserved samples of tissue held, generally, in hospitals and clinical laboratories, and laboratory samples that have been commercially developed, such as cell lines. The shift away from the rule against property in corpses began when the High Court of Australia held that it was possible for a person to acquire property rights over a corpse.¹³ Although the decision was not unanimous on this point, Griffith CJ took the view that the rule against property existed, but that it was subject to an exception. His Honour held that it was possible for human bodies and parts to become the subject of property rights where work or skill have been exercised to preserve them.¹⁴ This rule has been followed in subsequent cases, including the English case of *R v Kelly*,¹⁵ which held that preserved body parts taken from the Royal College of Surgeons were the property of the Royal College and that the unauthorised removal was theft.¹⁶

20.14 Under existing law, two elements are required for a sample to become property under this rule. First, the organisation or person using the tissue must have lawful authority to do so, such as a hospital has in relation to tissue taken for therapeutic purposes. Second, that organisation must apply some work or skill to the preservation of the sample. If both requirements are satisfied, the sample may be treated as property of the organisation.¹⁷

20.15 The recognition of property rights has implications for access, storage and use of such samples. However, the cases to date have dealt with only very limited fact situations. The courts have not produced any clear ruling on the particular property

10 *R v Rothery* [1976] Crim LR 691; *R v Welsh* [1974] RTR 478.

11 R Magnusson, ‘The Recognition of Proprietary Rights in Human Tissue in Common Law Jurisdictions’ (1992) 18 *Melbourne University Law Journal* 601, 617; G Dworkin and I Kennedy, ‘Human Tissue: Rights in the Body and its Parts’ (1993) 1 *Medical Law Review* 291, 300; P Matthews, ‘Whose Body: People as Property’ (1983) 36 *Current Legal Problems* 193, 223. Compare D Mortimer, ‘Proprietary Rights in Body Parts: The Relevance of Moore’s Case in Australia’ (1993) 19 *Monash University Law Review* 217, 244. On property status and theft see B Fisse, *Howard’s Criminal Law* (5th ed, 1990) Law Book Company, Sydney, 258–260.

12 *Roche v Douglas* [2000] 22 WAR 331. A similar conclusion on similar facts was made in *Pecar v National Australia Trustees Ltd* (Unreported, Supreme Court of NSW, Bryson J, 27 November 1996).

13 *Doodeward v Spence* (1908) 6 CLR 406. The case concerned an action for the return of a two-headed foetus preserved in a jar of alcohol.

14 *Ibid*, 414 (Griffith CJ).

15 *R v Kelly* [1998] 3 All ER 741.

16 Compare *Dobson v North Tyneside Health Authority* [1996] 4 All ER 474. This decision has been criticised and was not followed in later cases. See D Brahams, ‘Body Parts as Property’ (1998) 66(2) *Medico-Legal Journal* 45.

17 *Doodeward v Spence* (1908) 6 CLR 406, 414. This statement was cited with approval in *Pecar v National Australia Trustees Ltd (The Estate of Ivan Urlich deceased)* (Unreported, Supreme Court of NSW, Bryson J, 27 November 1996), 4; *R v Kelly* [1998] 3 All ER 741, 749.

rights that may be held over tissue samples, beyond a right of possession—the violation of which constitutes theft only in specific circumstances. It is not clear what other property rights exist in relation to tissue samples, though it could be argued that the common law has implicitly accepted the existence of other property rights in tissue, such as the right to use, by allowing continued possession by hospitals, laboratories and museums.¹⁸

Consequences of property rights

20.16 Property is often described as a ‘bundle of rights’, which includes the rights to use, transfer, manage and possess an object.¹⁹ This bundle of rights normally includes the right to the income generated by the object and the right to its capital value. Some of these rights may not be regarded as appropriate rights for individuals to have in respect of genetic material. For example, if full property rights existed in genetic material, its owner could sell it to the highest bidder. In place of the current system of altruistic donation of samples for research, a situation might develop whereby researchers would have to bid for access to genetic material.

20.17 There are other incidents of property that may be problematic in the context of human genetic information. Property can normally be subject to ‘execution’, that is, it can be seized to pay a debt pursuant to court order. However, it is unlikely that the legal system would countenance seizure of someone’s genetic material to satisfy a judgment debt.

20.18 Property rights normally are alienable, that is, they can be transferred to others. For example, if a hospital had property rights in a sample, it could transfer them to a pharmaceutical company for a fee. That company would then have the right to use the samples to produce a product and would be entitled to any income that was generated by this use.

Is a property approach appropriate?

20.19 Submissions made to the Inquiry differed on the benefits of an ownership regime for human genetic samples. A number favoured property rights in genetic material as a means of protecting genetic privacy, with some also suggesting solutions to the drawbacks of taking a property approach.²⁰

18 A more detailed analysis can be found in Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney Ch 17.

19 A Honore, ‘Ownership’ in A Guest (ed), *Oxford Essays in Jurisprudence* (1961) Clarendon Press, Oxford, 133.

20 A Johnston, *Submission G042*, 13 January 2002; Caroline Chisholm Centre for Health Ethics, *Submission G061*, 26 December 2001; D Pawlukowski, *Submission G067*, 15 January 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G106*, 26 February 2002; Womens Health Victoria, *Submission G076*, 3 January 2002.

Advantages of a property approach

20.20 A property approach to regulating human tissue has a number of advantages, including the following:

- Property rights can be asserted against others, who do not have the right to deal with the samples. Other persons are under a correlative duty not to interfere with an individual's rights in a sample, and are disabled from exercising rights over it.
- A property approach overcomes one of the major problems of the current consent-based approach to the use of genetic samples, namely the lack of on-going control. For example, if an individual gives consent for person A to use their tissue, and person A uses it in a manner inconsistent with that consent, the individual has a claim against that person. If, however, person A transfers the sample to company B who misuses it, the individual is not in a legal relationship with company B and generally has no claim against it.²¹
- A property approach promotes good record-keeping and best practice because researchers are legally required to act in accordance with the rights that others hold in those samples. Similarly, property rights promote consensual use of tissue samples.
- Property rights clarify the gift relationship between donors and researchers by defining which rights over the sample may be transferred, when and how. Property rights promote and regularise the transferability of samples within a commercial setting for the same reasons.
- A violation of property rights gives an individual the ability to bring a legal action for the return or destruction of their property. An individual could bring such an action if his or her genetic samples were being used in a manner contrary to those rights. If these rights had not been alienated, then a claim could be brought against any person who interfered with the samples.²²
- Property rights can be transferred to others. Rights to possess and use can be transferred by the individual to researchers, who will then be entitled to deal with the genetic samples in accordance with the right they have acquired.
- Property rights include rights to the income and the capital of an object. Allowing individuals to seek financial returns on the use of their tissue would enable them to share in the profits that are sometimes made from treatments that result from research.

21 However, there may be a claim for restitution or equitable relief.

22 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

Disadvantages of a property approach

20.21 There are, however, some significant problems with applying property principles to human genetic samples:

- Allowing people to exercise the rights to income and capital of human tissue might be regarded as allowing the human body to be commodified. This may alter community attitudes towards bodies and their parts, and as a result alter how communities perceive and treat living humans.
- Allowing people to exercise the rights to income and capital might also alter the current situation in which individuals freely donate their tissue.²³ Altruistic participation could be eroded.
- Sale of tissue samples would burden research by increasing costs, which would in turn be passed on to consumers.
- Individuals often discard tissue samples, intentionally or inadvertently, for example cheek cells left on dental floss.²⁴ A right of possession vested in the individual from whom the samples are taken might enable that individual to bring an action against others who deal with the samples, regardless of whether they were misusing them, on the basis of interference with the individual's right to possess the samples.
- By allowing an individual to transfer rights over their tissue samples to someone else, the individual's interest in what is done with the sample could be lost. The individual would then be precluded from preventing uses of the samples to which he or she objects.
- The recognition of property rights would also undermine the current system of ethical approval for research, where consent to use can be waived in some situations by a Human Research Ethics Committee (HREC).²⁵ It is questionable whether it would be lawful to waive consent where a person holds property rights over tissue.
- If genetic samples were regarded as property, de-identification would not extinguish the rights of the person from whom the sample was taken. De-identification is one of the current mechanisms used by researchers, with the approval of HRECs, to enable research to be carried out, while still protecting the privacy of individuals.

23 Coercion of the poor and the situation of the less wealthy selling parts of their bodies to the rich have also been raised as concerns. See T Murray, 'The Gift of Life Must Always Remain a Gift' (1986) (March) *Discover* 90.

24 H Kennedy, 'Bing's genes concern us all', *The Guardian* (London), 22 May 2002.

25 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra Ch 15; *Privacy Act 1988* (Cth) ss 95, 95A. On the waiver of consent to use tissue samples in research, see Ch 15.

- Property rights are difficult to apply to genetic material, which can be copied and reproduced.²⁶
- Property rights in tissue samples are currently grounded in the common law, which develops on a case-by-case basis in response to issues that come before the courts. For this reason, the common law cannot be relied on to produce a timely and comprehensive solution to these problems.

Views on property

20.22 DP 66 proposed that the common law right to possession of preserved samples should continue to be upheld. Responses to both this proposal and the general issue of property rights in tissue samples were varied.

20.23 Many submissions supported maintaining the status quo.²⁷ In doing so, a number warned of the consequences that might flow from attempting to alter the current common law position. The Law Society of New South Wales commented that any change ‘must be considered a fundamental shift in the law, and may have myriad unforeseen consequences’.²⁸ The Law Society recommended that no change be made to the law without further debate. The Queensland Government agreed, stating that ‘for the interim there is wisdom in maintaining the current position and practices in relation to ... preserved samples’.²⁹

20.24 Very few submissions disagreed with the efficacy of retaining the law as it currently stands. However, some presented alternative models, aimed at ameliorating the adverse consequences of possessory rights or of full ownership. These alternatives are described below.

20.25 One possible approach precludes individuals from holding property rights in their own genetic material, although hospitals and researchers would be able to have a proprietary interest in samples. This model circumvents problems with individual sale of tissue, while protecting and promoting research and aspects of clinical practice.

20.26 By upholding the possessory rights, this model would allow hospitals and researchers to maintain the integrity of their collections, and would protect samples from arbitrary interference. By not granting other rights (such as rights to the income) hospitals and researchers would be prevented from profiting from samples, and the culture of altruistic participation in research would be maintained. This approach allows for regulation through principles of consent to be maintained. Privacy NSW agreed on the efficacy of using informed consent principles to protect privacy.³⁰

26 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

27 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Queensland Government, *Submission G274*, 18 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002.

28 Law Society of New South Wales, *Submission G285*, 18 December 2002.

29 Queensland Government, *Submission G274*, 18 December 2002.

30 Office of the Privacy Commissioner (NSW), *Submission G118*, 18 March 2002.

Professor Loane Skene agreed with this approach, but advocated fuller property rights than possession for hospitals and researchers.³¹

20.27 A second approach, proposed by Genetic Health Services Victoria, is a modified version of the approach granting only possessory rights, described above. Tissue samples, such as the blood spots on newborn screening cards, would be owned by the health service that took the sample. However, that ownership would be subject to the requirements of National Pathology Accreditation Advisory Council standards and the *Privacy Act*.³² The Human Genetics Society of Australasia supported this approach.³³ The Australian Biospecimen Network proposed a similar approach, which would provide hospitals and others with full property rights over tissue to be exercised with regard to the notion of ‘moral ownership’.³⁴

20.28 As discussed above, property law is best understood as a ‘bundle of rights’. A number of approaches took advantage of this fact, and proposed prohibiting individuals from holding particular rights in tissue samples, which may be seen as problematic. For example, a third approach would allow individuals to hold property rights in their tissue to enable them to control its use, but would not give individuals the right to transfer these rights. Doctors and researchers would be able to use tissue samples under some form of lease agreement, after the presentation of a research proposal. It was suggested that lease agreements could be made publicly available and be overseen by an independent body.³⁵

20.29 Rights to income and capital could also be excluded from the ‘bundle of rights’ in order to address concerns about commodification, the erosion of altruistic participation in research, and the cost burden of having to pay for samples. UnitingCare NSW & ACT suggested allowing only limited property rights, in order to prevent commercial exploitation.³⁶

20.30 The Australian Academy of Science argued against individual property rights on the basis that genetic samples should be a community resource for health. The Academy made the point that if genetic samples were the personal property of the individual from whom they were taken, all research use would be encumbered by this interest.³⁷ The Life Sciences Network submitted:

We believe that health samples and records (maintained within a system which assures confidentiality about individuals) should be seen as part of our community resource for health, rather than as a form of personal property of the individual

31 L Skene, *Submission G017*, 16 November 2001.

32 Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

33 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

34 Australian Biospecimen Network, *Submission G238*, 19 December 2002.

35 A Johnston, *Submission G042*, 13 January 2002.

36 UnitingCare NSW & ACT, *Submission G052*, 14 January 2002. See also Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

37 Australian Academy of Science, *Submission G097*, 21 January 2002.

receiving care or treatment. The aggregated results of this knowledge bank confer a substantial community benefit.³⁸

20.31 Other approaches focused on the concept of custodianship. Dr Nikolajs Zeps of the Western Australia Tissue Bank suggested a model whereby tissue banks would be able to hold and use tissue as a custodian. The individual would retain the right to withdraw custodianship and could request the destruction or transfer of their samples as they wished:

Ideally the donor would delegate all responsibility for the use of their tissue to a Tissue Bank that distributes tissue under the auspices of [Human Research Ethics Committee] approval for those wishing to use tissue.³⁹

20.32 Each approach could also include a legal presumption that certain rights are transferred when samples are provided to overcome the problems that arise where tissue samples are discarded inadvertently. For example, it could be assumed that in providing a blood sample to a pathology laboratory, the individual transfers a right of possession. It could also be assumed that in some instances individuals have abandoned their rights over genetic samples, perhaps only on the condition that they are not misused.

Inquiry's views

20.33 As evidenced in other parts of this Report, the Inquiry considers it important to regulate the use of human genetic samples in order to ensure adequate protection of genetic information. In seeking to meet this goal, property rights have some clear benefits: they clarify the legal rights of donor and recipient; they facilitate on-going control by the donor until such time as the property is alienated; they enable the donor or other property owner to seek legal remedies for unlawful interference with propriety rights; and they enable a donor to share in the financial benefits that may accrue from use of the tissue.

20.34 However, the Inquiry has come to the view that the drawbacks of a property approach are considerable and outweigh the potential benefits at the present time. The recognition of property rights in human tissue has developed slowly in a piecemeal fashion under the common law, as new situations have been brought before the courts. As several submissions noted, there may be unforeseen consequences of extending property law to cover genetic samples, due largely to the strength of the rights that property law provides. As a result, property law may be a rather 'blunt instrument' for protecting a person's interest in his or her genetic samples.⁴⁰

20.35 The Inquiry recognises the need to avoid placing undue burdens on the conduct of research, given the considerable community benefit that is derived from the development of medical treatments and tests. Altruistic participation in research is

38 Life Sciences Network, *Submission G129*, 19 March 2002.

39 N Zeps, *Submission G047*, 14 January 2002.

40 R Magnusson, 'The Use of Human Tissue Samples in Medical Research: Legal Issues for Human Research Ethics Committees' (2000) 7(4) *Journal of Law and Medicine* 390, 394.

regarded as beneficial to the community as it reinforces a sense of unity and sharing.⁴¹ At present, virtually all research participation in Australia, including the use of tissue samples, occurs without remuneration of sample donors. Recognising an individual's right to sell tissue, as one incident of a property interest, would not only create an additional economic barrier to research, it would undermine the valuable system of community involvement in research.

20.36 Some alternative property models proposed in submissions addressed a number of specific concerns by removing various incidents of property from the 'bundle of rights' usually associated with property. However, once all the desirable limitations are imposed to obviate the adverse consequences of full ownership, the effect is very similar to the changes to the *Privacy Act* recommended in Chapter 8. This view was also expressed in submissions, with the Centre for Law and Genetics commenting that

if the proposal to extend the *Privacy Act* (and parallel state and territory legislation) to include genetic samples is accepted, then many of the arguments supporting the need to create property rights in genetic samples will be satisfied.⁴²

20.37 On balance, the Inquiry considers that the protection of genetic samples can be achieved more effectively by the changes to the *Privacy Act* recommended elsewhere in this Report.⁴³ In coming to this view, the Inquiry supports the circumstances in which the common law recognises a property right in preserved samples, and the process by which the courts develop the law incrementally in the context of specific cases. However, any legislative reform designed to extend the law of property to genetic samples would require a much broader debate within the community, in which the implications of an extension would need to be carefully weighed. This Inquiry has not been the occasion for such a debate.

Recommendation 20–1. The proprietary rights in preserved samples, which are currently enjoyed by hospitals and others under the common law, should continue to be upheld on a case-by-case basis. Legislation should not be enacted to confer full proprietary rights in human genetic samples.

Amendment of the Human Tissue Acts

20.38 DP 66 considered whether the Human Tissue Acts might be a useful vehicle for new regulations dealing with the collection and handling of human tissue samples and, in particular, the use of clinical samples for genetic research.⁴⁴

41 T Murray, 'On the Human Body as Property: The Meaning of Embodiment, Markets and the Meaning of Strangers' (1987) 20(4) *Journal of Law Reform* 1055.

42 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

43 *Ibid.*

44 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney Ch 17.

20.39 The Inquiry considers that amending the *Privacy Act* as recommended in Chapter 8 is a more effective means of protecting the privacy of genetic samples than amending the Human Tissue Acts. However, amendment of the Human Tissue Acts might be an option for protecting genetic samples if the *Privacy Act* is not amended as suggested in this Report.

20.40 In its 1977 report, *Human Tissue Transplants* (ALRC 7), the Australian Law Reform Commission noted that the common law was silent on property in human tissue removed during surgery or otherwise in the possession of a doctor or hospital. The Commission concluded, for the purposes of that project, that there was ‘no reason to endow such tissue with the attributes of property’.⁴⁵ As noted in DP 66, the current uses of tissue samples in genetic research were not envisaged at the time the Human Tissue Acts were enacted.⁴⁶

20.41 Following the recommendations in the Commission’s report, the Human Tissue Acts were enacted in all Australian jurisdictions.⁴⁷ As outlined in Chapter 18, the Human Tissue Acts provide for the consensual donation of blood, tissue and organs for transplantation, and for scientific, therapeutic or medical purposes. The Acts do not contain provisions dealing with the storage of samples, access to them, transfer to other researchers, or the future use of samples.

20.42 At present, the donation of tissue removed as part of medical treatment, such as sections of tissue excised during surgery or blood leftover from testing, is excluded from the coverage of the Human Tissue Acts.⁴⁸ There is no legislative requirement that consent be obtained to retain and use these samples. Consent to use of samples for further research must be obtained when access to the samples is sought, in accordance with the National Health and Medical Research Council’s *National Statement on Ethical Conduct in Research Involving Humans* (the National Statement).⁴⁹

20.43 In his submission, Dr Roger Magnusson referred to this omission in the Acts as a ‘loophole’, and suggested that it had facilitated the development of tissue banks in Australian hospitals and research institutions.⁵⁰ Another submission claimed that researchers obtain samples from tissue banks in order to avoid seeking consent directly from research subjects.⁵¹

45 Australian Law Reform Commission, *Human Tissue Transplants*, Report 7 (1977), AGPS, Canberra [13]. See also Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

46 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [17.45]–[17.46].

47 See *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1979* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1985* (Tas); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplant Act 1982* (WA); *Transplantation and Anatomy Act 1978* (ACT); *Human Tissue Transplant Act 1979* (NT).

48 *Human Tissue Act 1983* (NSW) s 34(1)(a), (b) and cognate state and territory legislation.

49 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra Ch 15.

50 R Magnusson, *Submission G039*, 10 January 2002.

51 *Confidential Submission G051CON*, 14 January 2002.

20.44 The quantity of samples currently stored in research databases and laboratory collections is a major concern for the protection of the privacy of genetic samples. Many of these samples, especially those that were originally taken for therapeutic purposes, are stored without the knowledge or consent of the person from whom they were taken.⁵²

Submissions and consultations

20.45 In DP 66 the Inquiry proposed that the Human Tissue Acts should not be used as the vehicle for regulating the collection, storage, access to or use of genetic samples, whether for the purposes of human genetic research or otherwise.⁵³ Most submissions agreed with this proposal. For example, the Centre for Law and Genetics submitted that:

we now accept the view of the inquiry that the inclusion of genetic samples in the *Privacy Act* would provide the optimal solution at the present time. If the proposal to include genetic samples is not accepted, then in our view the question of whether or not the Human Tissue Acts should be extended would need to be revisited.⁵⁴

20.46 The Victorian Institute of Forensic Medicine submitted that:

the need for legislative clarification of the status of tissue removed in situations and for uses not currently covered by the Human Tissue Acts needs to be debated in a wider context, taking account of the important issues raised by the Discussion Paper in relation to protection of genetic information.⁵⁵

20.47 The Inquiry is aware of moves to instigate a national review of the Human Tissue Acts.⁵⁶ The Department of Human Services South Australia suggested that the question of whether or not the Human Tissue Acts should be used to regulate the handling of genetic samples should be included in such a review.⁵⁷

20.48 There are a number of ways in which the Human Tissue Acts could be amended if a national review were undertaken, as suggested in these submissions. These include:

- comprehensive provisions dealing with consent to the storage and use of genetic samples in research;⁵⁸
- making tissue removed during medical procedures subject to the consent provisions and other requirements of the Acts;⁵⁹ and

52 These issues are discussed further in Ch 19.

53 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 17–2.

54 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

55 Victorian Institute of Forensic Medicine, *Submission G251*, 20 December 2002.

56 *Ibid*; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

57 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

58 Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; N Zeps, *Submission G047*, 14 January 2002.

- comprehensive provisions dealing with other aspects of the handling of tissue, including, for example, rules for the storage, use of and access to such tissue.

Inquiry's views

20.49 The Inquiry's preferred starting point for any comprehensive reform of the law relating to the collection, storage, use of, and access to, genetic samples is to build on existing information and health privacy legislation. As discussed in Chapter 8, this could be done by ensuring that privacy laws cover the handling of genetic samples, as well as the genetic information derived from them. Regulating genetic samples as well as information through the *Privacy Act* promotes uniformity and ensures that both are subject to the same regulatory controls.

20.50 If this approach is not adopted, the Human Tissue Acts may provide an alternative means of regulation. For example, amending the Human Tissue Acts could place the use of all human tissue samples under one regulatory scheme, removing confusion and promoting a consistent approach to the use of tissue in research. It could also give individuals a legal basis for complaint if they consider their tissue has been misused, and those who do not comply could be subject to criminal sanctions already provided for in the Acts.

20.51 However, on balance, the Inquiry considers that the Human Tissue Acts have a number of disadvantages, which make their potential extension problematic. One objection is that the storage and use of genetic samples would be regulated differently to the storage and use of genetic information derived from the samples. As a result, there may be overlaps, gaps or inconsistencies between the Human Tissue Acts and privacy laws.

20.52 Furthermore, the Human Tissue Acts were primarily intended to deal with the donation of blood, tissue and organs for transfusion, transplantation, and other therapeutic purposes; the removal of tissue after death; and the regulation of commerce in human tissue. To amend the Acts to regulate other aspects of the handling of human tissue may create unnecessary complexity in the existing legislative scheme.

20.53 The Human Tissue Acts have been reviewed over the last few years to deal with the retention and use of body parts after death. There may be practical reasons to refrain from further change until existing law reform processes have run their course.⁶⁰ As each State and Territory has enacted its own Human Tissue Act in substantially similar terms, any overhaul of the legislation might adversely affect harmonisation and

59 Womens Health Victoria, *Submission G076*, 3 January 2002; National Council of Women Australia, *Submission G095*, 31 January 2002; J Torrens, *Submission G029*, 11 January 2002.

60 B Walker, *Inquiry into Matters Arising from the Post-Mortem and Anatomical Examination Practices of the Institute of Forensic Medicine* (2001), Department of Health, New South Wales; South Australia Solicitor-General, *Report into the Retention of Body Parts after Post-Mortems* (2001), Department of Health Services, South Australia, Adelaide; NSW Health Department, *Review of the Human Tissue Act: Organ and Tissue Donation and Use and Post Mortem Examination* (1999), NSW Health, Sydney; Crown Solicitor for Western Australia and Commissioner for Health for Western Australia, *Final Report: Removal and Retention of Organs and Tissue Following Post-Mortem Examinations* (2001), Department of Health, Western Australia, Perth.

the smooth running of accepted processes for the collection and use of tissue in treatment and research.

20.54 A further argument against amending the Human Tissue Acts is that the Acts require consent to be obtained *prior* to the removal of tissue. To extend the requirement of prior consent to the collection of research samples that are removed during therapeutic procedures may be administratively onerous, potentially distressing to patients, and inconsistent with the present regulatory framework for the ethical conduct of research, based on the National Statement.⁶¹ The present regulatory framework provides for the waiver of consent in specified circumstances and gives guidance on the information that should be provided to individuals donating tissue for research. The system for ethical review of research proposals by HRECs allows a flexible approach to be taken on consent issues.

20.55 With these considerations in mind, the Inquiry is of the view that, until such time as the Human Tissue Acts are subject to a comprehensive national review, the regulation of the collection, storage, access to, or use of genetic samples (whether for the purposes of human genetic research or otherwise) should not rely primarily on amendments to the Human Tissue Acts.

Recommendation 20–2. Pending any comprehensive review of relevant laws, the regulation of the collection, storage, access to, or use of genetic samples (whether for the purposes of human genetic research or otherwise) should rely primarily on the *Privacy Act 1988* (Cth) as amended in accordance with the Recommendations in Chapter 8, rather than on amendment of the Human Tissue Acts.

61 See Part D.

Figure 1. Component parts of the human cell

(supplied by the Gene CRC <www.genecrc.org>)

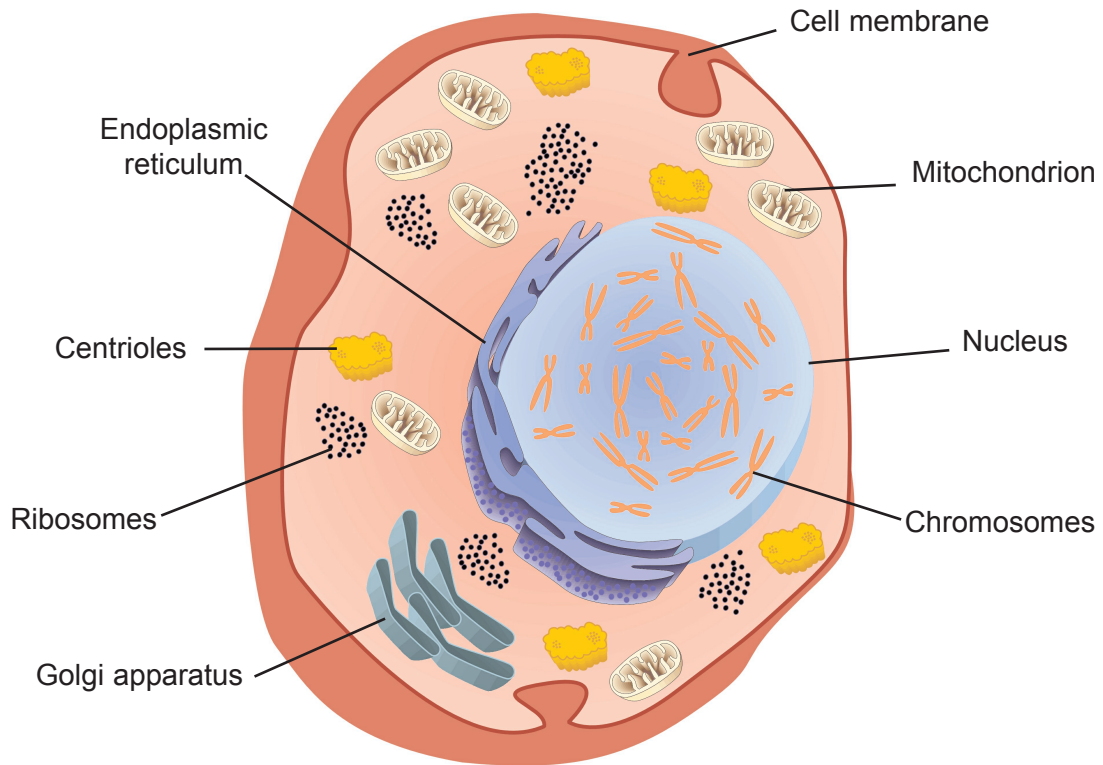


Figure 2. Human chromosomes

(supplied by the Gene CRC <www.genecrc.org>)

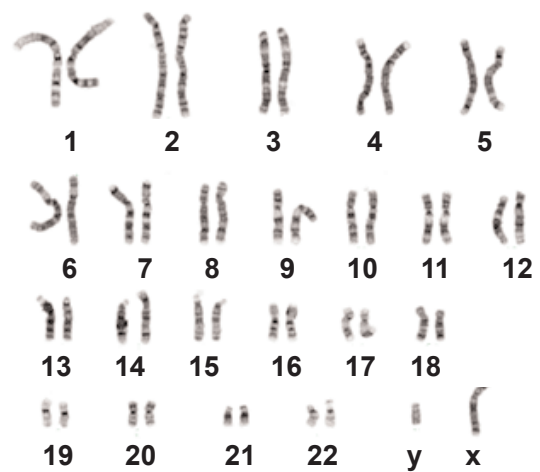


Figure 3. Each chromosome contains many genes

(supplied by the NSW Genetics Education Program <www.genetics.com.au>)

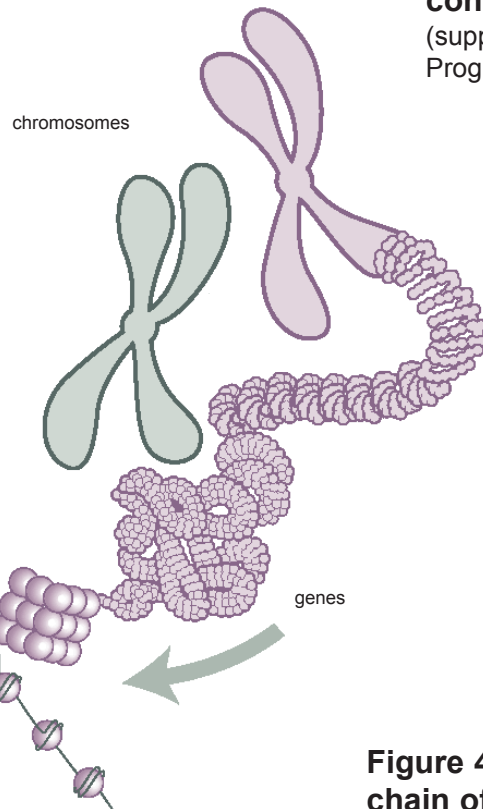


Figure 4. Each gene contains a chain of coded information

(supplied by the NSW Genetics Education Program <www.genetics.com.au>)

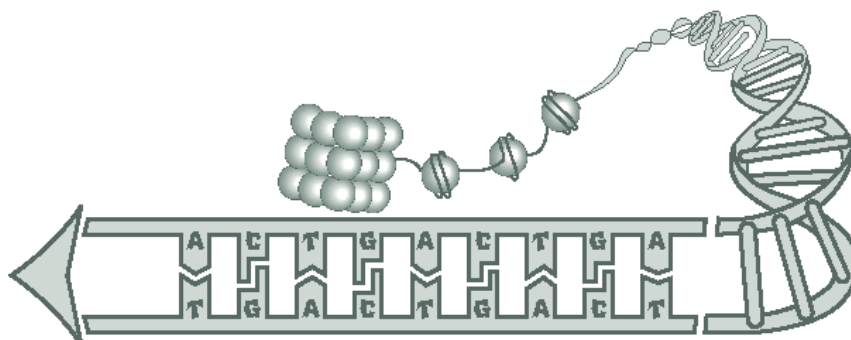


Figure 5. The DNA double helix, comprised of base pairs
(supplied by the Gene CRC <www.genecrc.org>)

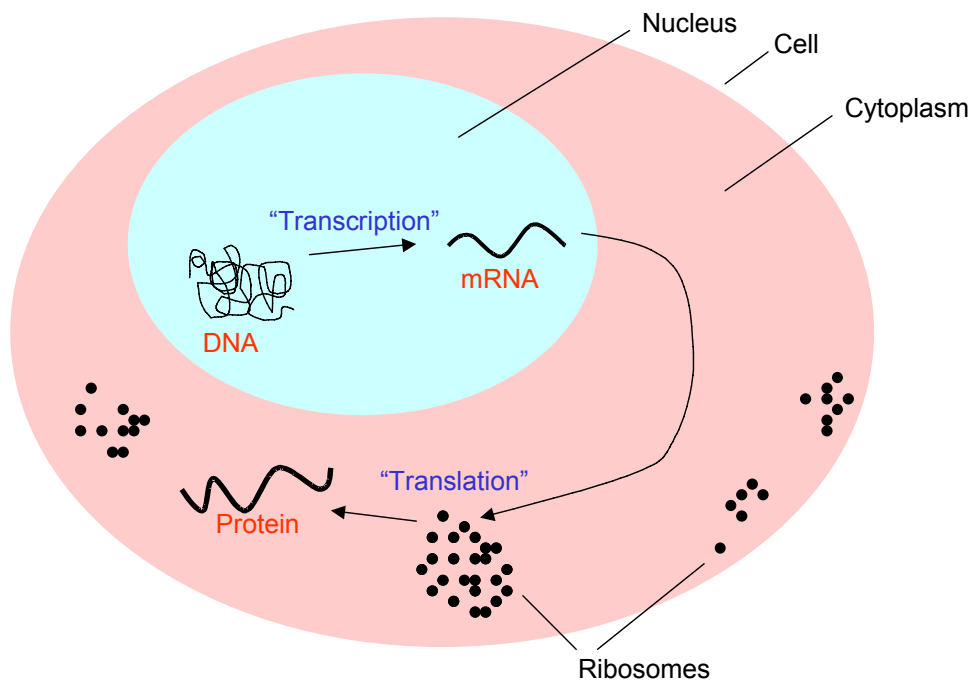
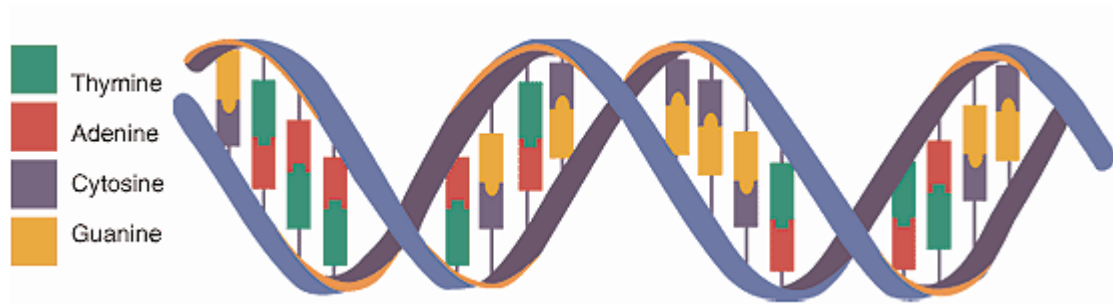


Figure 6. Protein synthesis
(supplied by David Abbott, NHMRC).
Genetic information in the DNA is transcribed to mRNA (messenger RNA) in the nucleus. mRNA then leaves the nucleus and moves into the cytoplasm where it associates with ribosomes and provides the information for the manufacture of proteins (translation).

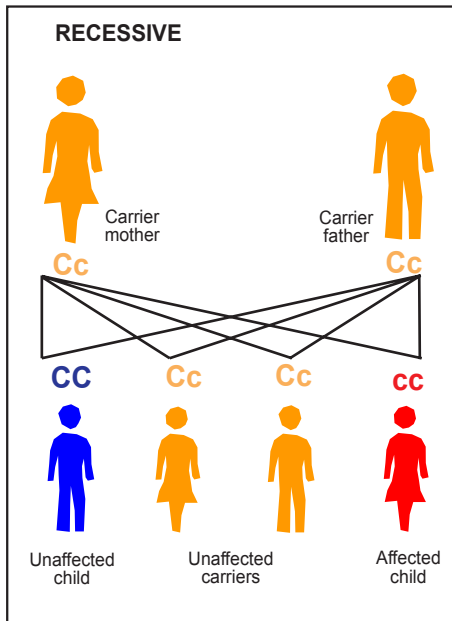


Figure 7, left. Inheritance patterns of recessive traits

(based on model supplied by the Gene CRC <www.genecrc.org>)

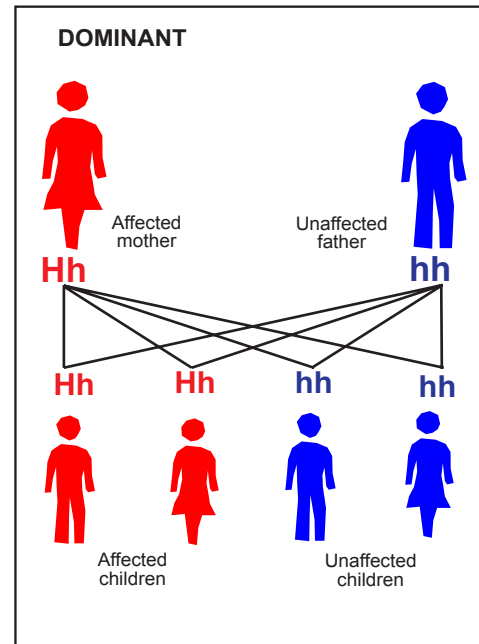


Figure 8, above. Inheritance patterns of dominant traits

(based on model supplied by the Gene CRC <www.genecrc.org>)

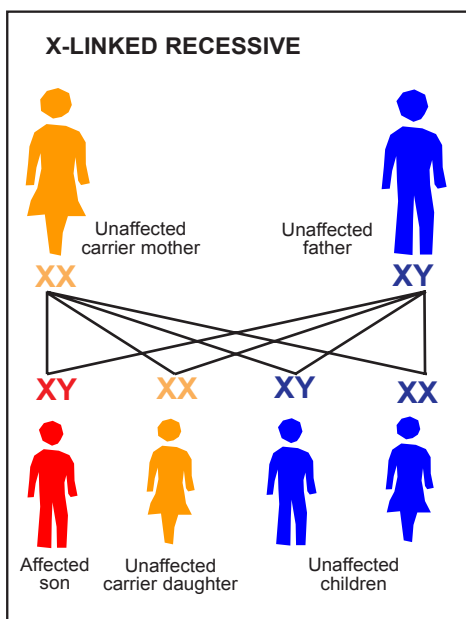


Figure 9, left. Inheritance patterns of X-linked traits

(based on model supplied by the Gene CRC <www.genecrc.org>)

Part F. Health Services

21. Health Professionals and Family Genetic Information

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Introduction

21.1 Genetic information may allow inferences to be drawn about persons other than the individual to whom the information most directly relates—most importantly about genetic relatives.¹ This leads to questions about how health professionals² should collect and deal with genetic information about genetic relatives, where that information is derived in the course of diagnosis, treatment or counselling.

21.2 This chapter examines issues related to the collection and disclosure of genetic information by health professionals, and individuals’ rights of access to genetic information about themselves, or their genetic relatives, held by health professionals. The chapter concludes that the *Privacy Act 1988 (Cth)* (*Privacy Act*) should be amended to broaden the circumstances in which health professionals may use or disclose genetic information to prevent threats to life, health or safety. The chapter also concludes that the *Privacy Act* should provide that individuals have a limited right to access genetic information about first-degree genetic relatives.

1 See Ch 3 and Ch 7.

2 In this Report, the term ‘health professional’ refers to those health professionals who can be expected to deal routinely with genetic information about their patients or clients, including clinical geneticists, other medical practitioners and genetic counsellors.

21.3 The collection and disclosure of family genetic information, and rights of access to such information, are central to the operation of genetic registers and to the conduct of genetic counselling. Issues raised by the use of genetic information in these contexts are addressed in Chapter 22.

Collection of genetic information by health professionals

21.4 The collection of family medical history is an established part of medical practice. When providing a health service, health professionals may need to collect family medical history in order to diagnose a patient's condition accurately. Information about the medical history of genetic relatives assists health professionals in providing effective health services to their patients. Such information may assist in diagnosis; the provision of medical advice about genetic risk to the patient or to present or future children; treatment or prevention; and genetic counselling. If this information is not collected the medical care or advice provided to the patient may be compromised.

21.5 This information may relate to the health, or age and cause of death, of individuals who are closely genetically related to the patient. It may also include social medical history; for example, information regarding marital status; health of spouse, children and other household members; and what social support is available.³

21.6 Family medical history takes on particular significance in the practice of genetic medicine.⁴ Genetic diagnosis is not always possible from a sample or information provided by one person and it may be desirable to test genetic relatives as well as the patient. Such testing may be necessary to establish inheritance patterns correctly, to confirm the mutation in at least one other affected member of the family (as part of developing a 'family-specific' genetic test) or to determine susceptibility for families at risk.⁵

21.7 Even if genetic relatives are not tested, the verification of a genetic diagnosis usually involves the provision of information from or about relatives. This is often referred to as the compilation of a 'family pedigree'.

The information to construct the 'family pedigree' may come from living relatives; death registers; autopsy results; cancer or other registers; medical records of relatives who have died; deposits of stored tissue; or other research studies.⁶

3 See *Privacy Commissioner Temporary Public Interest Determination No. 2001-1 2001* (Cth), 3.

4 Many submissions referred to the clinical importance of collecting medical information about the genetic relatives of patients. See G Suthers, *Submission G026*, 30 November 2001; *Confidential Submission G030CON*, 17 December 2001; Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

5 See L Skene, 'Patients' Rights or Family Responsibilities? Two Approaches to Genetic Testing' (1998) *6 Medical Law Review* 1, 5-6.

6 *Ibid.*, 6.

Application of privacy legislation to collection

21.8 The *Privacy Act* imposes constraints on the collection of genetic information by doctors and other health professionals about the genetic relatives of their patients.⁷ National Privacy Principle (NPP) 1 of the *Privacy Act* provides that an organisation must not collect personal information unless the information is necessary for its functions, and that it must collect personal information only by lawful and fair means and not in an unreasonably intrusive way.⁸ Individuals must be informed about various matters such as their access rights, the purposes of collection, and to whom the organisation usually discloses information of that kind.⁹ In general, an organisation must collect personal information about an individual only from that individual, rather than from any third party, unless it is not 'reasonable and practicable' to do so.¹⁰

21.9 NPP 10.1 of the *Privacy Act* provides that an organisation must not collect sensitive information (including genetic and other health information) unless the individual has consented. The principle then sets out the circumstances in which an organisation may collect sensitive information without consent. In some circumstances, the collection of family medical history by health professionals without the consent of the family members to whom the information relates would breach NPP 10.1.

21.10 This position was remedied by a Temporary Public Interest Determination (the Temporary PID) issued by the federal Privacy Commissioner on 21 December 2001,¹¹ and by final Public Interest Determinations (PIDs) issued on 15 October 2002.¹² In issuing the PIDs, the Privacy Commissioner concluded that the public interest in taking social, family or medical history outweighed to a substantial degree the public interest in full adherence to the NPPs because history taking is central to good health care and services. Strict compliance with NPP 10.1 would jeopardise public health and be an 'unrealistic curtailment' on the 'core and essential practices' of health providers.¹³

7 Patients themselves need not comply with the *Privacy Act* NPPs in their collection, use and disclosure of information about their genetic relatives provided this is for the purposes of their 'personal, family or household affairs': *Privacy Act 1988* (Cth) s 16E; *Health Records Act 2001* (Vic) s 13.

8 *Privacy Act 1988* (Cth) NPP 1.1–1.2.

9 *Ibid*, NPP 1.3.

10 *Ibid*, NPP 1.4. In order to determine whether it is 'reasonable and practicable' to collect information indirectly, the federal Privacy Commissioner has recommended balancing a number of factors. These include: whether it is possible to collect the information directly; whether a reasonable individual might expect information about them to be collected directly or indirectly; how sensitive the information is; the cost to an organisation of collecting directly rather than indirectly; the privacy consequences for the individual if the information is collected indirectly rather than directly; and what is accepted practice: Office of the Federal Privacy Commissioner, *Guidelines to the National Privacy Principles* (2001), OFPC, Sydney, 22.

11 The date the NPPs came into force under the *Privacy Amendment (Private Sector) Act 2000* (Cth).

12 *Privacy Commissioner Public Interest Determination No. 9 2002* (Cth); *Privacy Commissioner Public Interest Determination No. 9A 2002* (Cth). A Public Interest Determination (PID) may be issued by the Privacy Commissioner, on the application of an interested person, where an act or practice may breach the NPPs but the public interest in doing the act, or engaging in the practice, substantially outweighs the public interest in adhering to NPPs: See *Privacy Act 1988* (Cth) Pt VI. PID 9A gives PID 9 general effect to apply to all health service providers.

13 *Privacy Commissioner Public Interest Determination No. 9 2002* (Cth), 17.

21.11 The effect of the PIDs is to ensure that an organisation can collect social, family or medical history from an individual (a consumer) about another individual (a third party) without breaching NPP 10.1 where collection is

- necessary for the organisation to provide a health service directly to the consumer; and
- the third party is a member of the consumer's family or household, or the third party's information is otherwise relevant to the consumer's family, medical or social history.¹⁴

21.12 Unlike the Temporary PID, the final PIDs do not exempt organisations from their obligations to adhere to NPP 1.5. As a result, organisations remain obliged to take reasonable steps to ensure that third parties are informed about the collection of information.¹⁵

21.13 Issues concerning the collection of health information about genetic relatives will also arise under state and territory privacy legislation, which contains privacy principles similar to those in the federal *Privacy Act*. For example, the *Health Records Act 2001* (Vic) contains provisions requiring an individual's consent to the collection of health information about that individual.¹⁶ Under the Act, organisations must collect health information about an individual only from that individual (if it is reasonable and practicable to do so)¹⁷ and individuals must be informed about the circumstances of collection where health information about them is collected from someone else.¹⁸

21.14 Collection by health professionals of family medical history without the consent of family members may breach some or all of these provisions. In response, the Victorian government has promulgated regulations to permit health service providers to collect health information from a person about another individual where the information is reasonably necessary to ensure that health services are provided safely and effectively to the person.¹⁹

14 Ibid.

15 However, it may not be necessary to take such steps where the third party is already aware of the relevant matters or where steps could be taken but it is unreasonable to do so. The Privacy Commissioner has stated that deciding what steps are reasonable involves making a judgment based on the situation as a whole and balancing a number of possible factors including: a third party's expectations and existing knowledge about the collection of information; the practicalities of providing notice to the third party; conflicting legal obligations, such as legal professional privilege and the obligations of confidentiality on the organisation; and the costs of providing notice to a third party: Ibid, 9.

16 *Health Records Act 2001* (Vic) Health Privacy Principle 1.1(a).

17 Ibid, Health Privacy Principle 1.3. See also *Health Records and Information Privacy Act 2002* (NSW), Health Privacy Principle 3(1).

18 *Health Records Act 2001* (Vic) Health Privacy Principle 1.5. See also *Health Records and Information Privacy Act 2002* (NSW) Health Privacy Principle 4(2).

19 *Health Records Act 2001* (Vic) Health Privacy Principle 1.1(i); *Health Records Regulations 2002* (Vic) r 8. The information collected is exempt for the purposes of Health Privacy Principle 1.5, meaning that the organisation need not inform the third party about the collection.

21.15 Importantly, the PIDs and the Victorian regulations apply only to situations in which a health service is provided directly to the individual (the patient) from whom family medical history is sought. The information must also be collected directly from the individual being treated in order to come within the terms of these exemptions.

21.16 In some circumstances information about genetic relatives is collected in order to facilitate the diagnosis or treatment of someone other than the patient. This may be done to give genetic relatives an opportunity to become aware of their genetic risk and to access genetic counselling and medical advice. Information collected from patients may then need to be verified through contact with other sources, such as the treating doctors of those genetic relatives. The PIDs and the Victorian regulations do not authorise these forms of indirect collection of information about genetic relatives. These limitations are particularly relevant to the operation of genetic registers, as discussed in Chapter 22.

21.17 However, at least in relation to the collection of family medical history where this is necessary to provide a health service directly to the person providing the history, the PIDs have removed constraints on established medical practice that would otherwise exist. To this extent, the PIDs recognise the shared nature of family medical histories, including genetic information, and provide an appropriate framework for collection of this information.

Disclosure of genetic information to genetic relatives

21.18 Many submissions identified disclosure of genetic information to genetic relatives as an important issue for the Inquiry. In what circumstances should a patient or his or her doctor inform other members of the family about genetic information relevant to the latter's health or well-being? For example, it has been suggested that before patients are tested they should be advised to consider carefully the persons with whom the test result should be discussed.²⁰ If, following testing, the patient is unwilling to inform his or her relatives about results that may be important to them, should the doctor take steps to inform those relatives?

21.19 These issues have been referred to as 'a looming area of medico-legal controversy',²¹ and have generated a great deal of comment in Australia and overseas.²²

The overriding responsibility of the clinical geneticist remains with the patient and not to any other family members and certainly not to society because of the public health effects of the mutant gene. Nevertheless, the question of whether a mutant gene present in one member of a family constitutes clear-cut danger to others in the family,

20 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, NHMRC, <www.nhmrc.gov.au/issues/humangenetics.htm>, 19 February 2003, 34.

21 D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130.

22 See eg Ibid; L Skene, 'Patients' Rights or Family Responsibilities? Two Approaches to Genetic Testing' (1998) 6 *Medical Law Review* 1; T Lemmens and L Austin, 'The Challenges of Regulating the Use of Genetic Information' (2001) 2(3) *Isima: Canadian Journal of Policy Research* 26; E Pergament in M Rothstein, *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) Yale University Press, New Haven.

thereby justifying warning family members regardless of a patient's preference, has not yet been answered satisfactorily.²³

21.20 Some submissions suggested that there should be more latitude for disclosure to genetic relatives given that the adverse health consequences of some genetic conditions may be both serious and preventable.²⁴ Other submissions expressed the view that the position with regard to disclosure in such circumstances should at least be clarified.²⁵

21.21 This section examines existing constraints on disclosure to genetic relatives arising from the duty of medical confidentiality and the application of privacy legislation. The section also discusses the legal and ethical duties that health professionals may owe to family members.

21.22 The Inquiry has concluded that privacy legislation inappropriately constrains health professionals' decisions about the disclosure of clinically relevant information to genetic relatives. The Inquiry proposes reforms to remedy this situation, including amendments to the *Privacy Act* and the development of National Health and Medical Research Council (NHMRC) guidelines on this issue.

Assessing the need for reform

Disclosure and prevention of harm

21.23 In some circumstances, the disclosure of genetic information can prevent serious health consequences for genetic relatives by allowing the early detection and treatment of inherited genetic disorders. Issues surrounding the disclosure of information to genetic relatives may become increasingly important as further preventive measures become available to mitigate genetic risk.²⁶

[As] knowledge gained from genetic testing may enable effective prevention of some adverse outcomes ... restricting the information to the single individual tested may fail to offer the opportunity for preventative action in other at risk family members.²⁷

21.24 Developments in genetic medicine have implications for the extent to which the confidentiality of the doctor and patient relationship should be given primacy over other ethical considerations. It is easier to argue that the information should remain confidential when genetic information reveals an inherited susceptibility to a disease

23 E Pergament in M Rothstein, *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) Yale University Press, New Haven 92, 95.

24 Centre for Law and Genetics, *Submission G048*, 14 January 2002; F Macrae, *Submission G069*, 14 January 2002; G Suthers, *Submission G026*, 30 November 2001; R Magnusson, *Submission G039*, 10 January 2002.

25 New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; *Confidential Submission G051CON*, 14 January 2002.

26 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

27 J MacMillan, *Submission G015*, 19 November 2001.

that is not preventable than when denying access may place family members at increased risk of harm.²⁸

21.25 Clinical geneticists and others provided many examples of such situations in submissions to the Inquiry. Dr Graeme Suthers, the Head of the Familial Cancer Service in South Australia, referred to the following situation, based on an actual case (names have been changed).

Deidre has had breast cancer and has been shown to have an inherited mutation in the BRCA1 gene. Her mother, Marjorie, also had breast cancer and presumably carries the same mutation. Marjorie has a large extended family with many young women at risk of having the mutant gene and of developing early-onset breast cancer. Deidre had given me the address of her mother and had agreed that she (and other relatives) could be informed of the outcome of genetic testing. This would pave the way for genetic testing of Marjorie's unaffected relatives. However, on receiving the test result Deidre changes her mind and revokes permission for this information to be released to her relatives. At this stage none of her relatives are aware that she has had a genetic test. Whose rights should prevail—Deidre's right to the confidentiality of her test result, or Marjorie's right to be informed of a result she doesn't know about but which maybe life-saving?²⁹

21.26 The literature dealing with genetic information and confidentiality contains many other examples in which disclosure is clearly capable of averting significant health dangers.³⁰ Professor Loane Skene has concluded that disclosure to genetic relatives is arguably most justified in the case of familial adenomatous polyposis (FAP). Other situations may be identified where the benefits for genetic relatives of knowing they are at increased risk are merely speculative and may not, therefore, be capable of justifying a breach of confidentiality.³¹

21.27 In situations where there are benefits in informing genetic relatives, consent to do so may be obtained following discussion with the person tested. Existing ethical guidelines emphasise that when genetic information is to be shared with family members, the most appropriate person to make the initial contact is the individual who has undergone the genetic test.³² Submissions confirmed that it is standard clinical practice to request the individual's permission to pass on relevant genetic information to relatives.³³

28 Ibid.

29 G Suthers, *Submission G026*, 30 November 2001.

30 L Skene, 'Genetic Secrets and the Family: A Response to Bell and Bennett' (2001) 9 *Medical Law Review* 162, 169; W-C Leung, 'Results of Genetic Testing: When Confidentiality Conflicts with a Duty to Warn Relatives' (2000) 321 *British Medical Journal* 1464 referred to in R Magnusson, *Submission G039*, 10 January 2002.

31 For example, M Burgess, C Laberge and B Knoppers, 'Bioethics for Clinicians: 14. Ethics and Genetics in Medicine' (1998) 158(10) *Canadian Medical Association Journal* 1309 referring to a hypothetical example involving breast cancer where there is no guaranteed prophylaxis for breast cancer (but early detection and treatment may lead to a better outcome) and there are social and psychological risks associated with informing and not informing a patient's sisters.

32 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, NHMRC, <www.nhmrc.gov.au/issues/humangenetics.htm>, 19 February 2003, 49.

33 Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

21.28 Disclosure to genetic relatives by, or with the consent of, the patient is obviously desirable. However, during consultations the Inquiry was informed of circumstances in which a patient will neither disclose the information nor consent to the health professional doing so, such as where family relationships have broken down irretrievably. While some patients may actively object to, or obstruct, the disclosure of information to genetic relatives, non-disclosure may also come about because of lack of interest, denial or practical barriers to communication.³⁴ Dr Finlay Macrae, Head of Colorectal Medicine and Genetics at the Royal Melbourne Hospital, referring to genetic test results that confirm FAP, commented that:

Standard advice is that the affected and genotyped individual passes the information of the availability of predictive testing to at risk relatives. But some do not pass the information on to all relevant family members, for a variety of reasons. Avoidable deaths do occur because of this.³⁵

21.29 Other submissions and consultations emphasised situations in which there are strong ethical arguments for health professionals telling a genetic relative about the consequences for them of another person's genetic test results. This is especially so where there is high risk of a genetically based cancer that is treatable.³⁶

Where voluntary genetic testing reveals that a sibling, son or daughter may face a *substantial and quantifiable risk* of a serious disease or disability, in circumstances *where earlier interventions could alleviate or reduce the harm suffered*, then there may be strong moral reasons for breaching confidentiality. Similar considerations would apply, even where genetic testing reveals that a genetic relative is only a carrier of a harm-causing mutation.³⁷

21.30 Similarly, the Human Genetics Society of Australasia (HGSA) considered that, with regard to highly penetrant heritable genetic disorders

doctors should consider the situation of the person's relatives and try to ensure that they are informed if they are at increased risk of having inherited the disorder in question (or susceptibility to it).³⁸

The right not to know

21.31 While disclosure of genetic information has the potential to prevent harm to genetic relatives, decisions about disclosure should also take into account the fact that people have a 'right not to know' about their genetic risk.³⁹

34 National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002.

35 F Macrae, *Submission G069*, 14 January 2002. Two cases of deaths that may have been avoidable, if disclosure of genetic information had occurred, were reported in *The Age* newspaper: T Noble, 'Let Us Warn Patients of Gene Mutations, Say Doctors', *The Age* (Melbourne), 6 August 2002.

36 R Magnusson, *Submission G039*, 10 January 2002; Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002; National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002.

37 R Magnusson, *Submission G039*, 10 January 2002.

38 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

39 See Ch 7.

21.32 Privacy NSW submitted that the wishes of genetic relatives not to know their genetic information are better respected if the patient has control over disclosure and is appropriately supported in making a decision as to whether or not to disclose the information to others. The Office of the Federal Privacy Commissioner (OFPC) observed that:

in some families the person best qualified to make a particular decision regarding disclosure to a relative is another member of the family or even a friend of the family. Alternatively, it may be impossible for family members to communicate at all, let alone about the implications of genetic test results affecting one of the family.⁴⁰

21.33 The Cancer Genetics Ethics Committee of the Anti-Cancer Council of Victoria has observed that:

With a condition like FAP, in which virtually all who carry a gene mutation develop cancer, and in which the cancer may be prevented, the strong presumption should be that the relatives will be grateful for being warned. The same presumption should not be made in a cancer such as breast cancer, where the risk of developing cancer ... is less than 100% and there is no assurance of a successful medical intervention.⁴¹

Need for clarity

21.34 Many submissions considered that the law and ethical guidelines in this area should be made clearer. Dr Roger Magnusson submitted that where there is a contest between a patient's privacy and a risk of genetic harm to a third person, existing privacy laws are arguably deficient.⁴² He concluded that the current law on confidentiality and disclosure of genetic information is 'unreasonably opaque'⁴³ and suggested that it is one area 'where some "genetic-specific" privacy regulation would be welcome'.⁴⁴

21.35 Several submissions expressed concern about the uncertainty of existing law dealing with doctors' duties to genetic relatives⁴⁵ and about its possible future development through case law or legislation.⁴⁶ Associate Professor John MacMillan considered that, whatever the current position, in future it may be held by courts that doctors' duties to genetic relatives are not discharged simply by informing their patient about the implications of genetic information for the patient's relatives.⁴⁷

21.36 What is the legal position where the patient is unwilling to communicate with his or her relatives? In what circumstances does the law permit a doctor to take steps to inform genetic relatives about information relevant to their health? The law

40 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

41 Cancer Genetics Ethics Committee, *Ethics and Familial Cancers* (1997), Anti-Cancer Council of Victoria, Melbourne, Guideline 16.

42 R Magnusson, *Submission G039*, 10 January 2002.

43 *Ibid.*

44 *Ibid.*

45 G Suthers, *Submission G026*, 30 November 2001; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

46 J MacMillan, *Submission G015*, 19 November 2001; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

47 J MacMillan, *Submission G015*, 19 November 2001.

relating to these issues is discussed below, with reference to common law duties of confidentiality and the provisions of the *Privacy Act* and similar state and territory privacy legislation.⁴⁸

Duties of confidentiality

21.37 Doctors and other health professionals owe their patients a common law duty to maintain the confidentiality of information provided by patients.⁴⁹ Where a doctor breaches this duty, the doctor may be liable for damages in tort, contract or for equitable breach of confidence. In addition, a breach of confidence may constitute unsatisfactory professional conduct and form grounds for proceedings before medical registration authorities.⁵⁰

21.38 The common law duty of confidentiality may be breached where there is an unauthorised use of information that is subject to the duty. This will occur where the information is used for a purpose inconsistent with the purpose for which consent was expressly or impliedly given.⁵¹

21.39 In many circumstances, patients may be taken to have consented to disclosure of information, especially for purposes related to their own treatment, such as to other health service providers who assist the patient's doctor in providing optimal care. However, disclosure by the doctor to facilitate diagnosis or treatment of family members cannot ordinarily be implied and is likely to breach the duty of confidentiality, unless the disclosure is covered by some exception recognised by law.

21.40 Exceptions to the common law duty of confidentiality permit disclosure of the information in ways that would otherwise infringe the duty. One exception is where a patient consents to the disclosure. It is the patient to whom the duty is owed and so he or she can choose to permit information to be released, including to facilitate the

48 This discussion proceeds on the basis that relevant disclosures to genetic relatives involve information about an individual patient whose identity is apparent or can reasonably be ascertained from the information. In some circumstances it may be possible to inform genetic relatives about their genetic risk without disclosing information about a particular individual.

49 Duties of confidentiality may also arise as a term of the contractual relationship between medical practitioner and patient: *Parry-Jones v Law Society* [1969] 1 Ch 1, or as an incident of the fiduciary character of that relationship: *Breen v Williams* (1996) 186 CLR 71, 81 (per Brennan CJ). Similar duties of confidentiality can be imposed directly by legislation governing the use of medical information in publicly funded health services: *Health Administration Act 1982* (NSW); *Public Health Act 1991* (NSW); *Private Hospitals and Day Procedure Centres Act 1988* (Qld); *Health Services Act 1991* (Qld); *Health Act 1937* (Qld); *South Australian Health Commission Act 1976* (SA); *Public and Environmental Health Act 1987* (SA); *State Service Act 1984* (Tas); *Health Services Act 1988* (Vic) or by the application of statutory disciplinary standards of unethical conduct: *Duncan v Medical Practitioners Disciplinary Committee* [1986] 1 NZLR 513. Duties of confidentiality are also expressed in codes of professional ethics.

50 For example, in proceedings before the NSW Medical Board or Medical Tribunal under *Medical Practice Act 1992* (NSW).

51 J Hamblin and D Bell, 'Confidentiality' in J Golden and D Grozier (eds), *The Laws of Australia* (1994) Law Book Company Limited, Sydney, vol 20, [6]. In the United Kingdom, the English Court of Appeal, in *R v Department of Health, ex parte Source Informatics Ltd* [2000] 1 All ER 786, has held that the unauthorised disclosure of anonymised information is not a breach of confidentiality. See G Laurie, *Genetic Privacy* (2002) Cambridge University Press, Cambridge, 223–229.

diagnosis or treatment of a genetic relative. A second exception is where there is a statutory obligation to disclose information. This is regularly exercised in the compulsory disclosure of certain notifiable diseases or other conditions for which there is a statutory register. It also includes compulsion to disclose information in court proceedings. A third exception permits the release of confidential information where to do so is in the public interest. The possible application of the public interest exception to genetic information is of particular relevance, and is considered below.

The public interest exception

21.41 It has been stated that the public interest exception to the duty of medical confidentiality 'is notable for its extraordinary flexibility'.⁵² It can potentially be invoked to justify the disclosure of confidential patient information in a wide range of circumstances. Whether these circumstances might encompass disclosure of confidential information to genetic relatives is open to debate.

21.42 Some legal commentators believe that the public interest exception might be used in this way. On this view the public interest exception could cover cases where

the patient's medical condition presents an infection risk to others, where a patient's ill health renders him or her unfit to continue certain activities because others would be placed at risk, or where inherited genetic disorders should properly be disclosed to other family members.⁵³

21.43 However, there has been no reported Australian case in which disclosure of genetic information has been found to be justified, and other commentators have doubted whether the public interest exception would extend to the disclosure of genetic information. Professor Skene has concluded that the public interest exception may not always be sufficient to protect health professionals making disclosures of genetic risk to relatives because the harm is seldom imminent.⁵⁴ Dean Bell and Belinda Bennett state that the existing case law requires that disclosure must be confined to 'exceptional circumstances', where 'another's life is immediately endangered and urgent action is required'⁵⁵ or where there is a 'real risk and consequent danger to the public'.⁵⁶ They conclude that:

These limitations suggest that the public interest basis for disclosure to family members of a genetic condition has not, to date, been contemplated for a situation such as disclosure to a family member of genetic condition, both because such a condition will rarely, if ever, present an immediately life-threatening risk, and also because such disclosure would ultimately have to be disclosed to the family member

52 J Hamblin and D Bell, 'Confidentiality' in J Golden and D Grozier (eds), *The Laws of Australia* (1994) Law Book Company Limited, Sydney, vol 20, [31].

53 *Ibid.*, vol 20, [31].

54 L Skene, 'Genetic Secrets and the Family: A Response to Bell and Bennett' (2001) 9 *Medical Law Review* 162, 168–169.

55 D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) *Medical Law Review* 130, 149, citing *Duncan v Medical Practitioners Disciplinary Committee* [1986] 1 NZLR 513.

56 D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130, 149, citing *W v Egdell* [1990] 1 All ER 835.

rather than a responsible authority (although if the relative is not a patient of the doctors, it may be disclosed to that patient's treating doctor).⁵⁷

21.44 It has been suggested that the factors that a court might take into account in weighing up a public interest claim include the effects of disclosure on the willingness of individuals to take advantage of genetic tests in the future, and the likely public health outcomes of such a policy.⁵⁸ Roger Magnusson has stated that the scope of the discretion to disclose under the public interest defence to an action for breach of confidentiality is uncertain and probably requires resolution by the High Court.⁵⁹

A duty to warn?

21.45 In some circumstances it might be argued that a health professional has a positive 'duty to warn' third parties—even if doing so would infringe the patient's confidentiality. Such a duty might be derived from common law principles relating to the tort of negligence, based on the concept of a duty of care. Submissions questioned whether Australian law imposed such a duty on health professionals.⁶⁰

21.46 Cases in the United States, and in particular *Tarasoff v Regents of University of California*,⁶¹ have established that, where there is a foreseeable risk of significant harm to an identified individual, doctors and other health professionals may have a duty to warn those individuals. Some United States cases involved genetic risks.⁶² The cases differ as to whether the doctor's duty to warn can be satisfied by warning the patient that family members should seek medical care or whether a doctor has a duty to warn genetic relatives directly.⁶³

21.47 There is limited recognition in Australian law that a doctor may owe a duty of care to someone who is not a patient.⁶⁴ In *BT v Oei*,⁶⁵ the Supreme Court of New South Wales held that a duty of care was owed by a doctor to a patient's partner in advising the patient in relation to the need for an HIV test.⁶⁶ It has been suggested that a doctor or genetic counsellor may therefore owe a duty of care to third parties, which could be satisfied by explaining to the patient the implications of a genetic test for the

57 D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130, 149.

58 *Ibid.*, 148–149.

59 R Magnusson, *Submission G039*, 10 January 2002 contrasting the approaches taken by Gummow J in *Corrs Pavey Whiting & Byrne v Collector of Customs (Vic)* (1987) 14 FCR 434 and Kirby P in *Attorney-General (UK) v Heinemann Publishers Australia Pty Ltd* (1987) 10 NSWLR 86.

60 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; R Magnusson, *Submission G039*, 10 January 2002.

61 *Tarasoff v Regents of University of California* 551 P2d 334 (1976).

62 *Pate v Threlkel* 551 P2d 334 (1976); *Safer v Pack* 291 NJ Super 619 (1996), 677. These cases are discussed in D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130, 149–152 and L Andrews, 'The Genetic Information Superhighway: Rules of the Road for Contacting Relatives and Recontacting Former Patients' in B Knoppers (ed), *Human DNA: Law and Policy* (1997) Kluwer Law International, The Hague, 133, 136–138.

63 See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [18.51]–[18.53].

64 *BT v Oei* (Unreported, Supreme Court of NSW, Bell J, 5 November 1999).

65 *Ibid.*

66 *Ibid.*

future health of third parties, at least where disclosure by the patient could ameliorate the harm.⁶⁷

21.48 However, there is no legal authority for the imposition of a *Tarasoff*-style duty to warn in Australia. Professor Skene concluded in 1998 that arguments that doctors have a duty to warn genetic relatives are unlikely to be successful in Australia.⁶⁸ Similar conclusions have been reached by other commentators.⁶⁹

21.49 The Inquiry does not favour reforms that would impose a duty of care on doctors or other health professionals to warn genetic relatives about genetic information relevant to them. In appropriate circumstances, doctors should be able to disclose genetic information without incurring liability but they should have no duty to do so. The policy reasons for not requiring disclosure of genetic information to relatives have been summarised as including

the negative impact that such a breach of confidentiality will have on patients' willingness to seek genetic testing in the first place; the potential negative social, psychological, and financial impact that such information will have on the relative who receives the information; and the eugenic message that is conveyed by mandating such disclosures.⁷⁰

21.50 There are many practical difficulties in recognising a duty to warn. These include how to define the relatives to whom the duty is owed; determining how far health professionals are required to go in seeking to contact them; and the level of advice health professionals should give in order to discharge the duty.⁷¹

21.51 As discussed below, the Inquiry recommends amendment of the *Privacy Act* to broaden the circumstances in which health professionals may disclose genetic information to genetic relatives without the consent of their patients. While this reform

67 R Magnusson, *Submission G039*, 10 January 2002.

68 L. Skene, 'Patients' Rights or Family Responsibilities? Two Approaches to Genetic Testing' (1998) 6 *Medical Law Review* 1, 29–30. See also D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130, 149–154. Similarly, Graeme Laurie has concluded that 'the likelihood of a duty of disclosure being imposed in the United Kingdom is currently slight': G Laurie, *Genetic Privacy* (2002) Cambridge University Press, Cambridge, 270.

69 B McGivern, 'Tortious Liability for (Selected) Genetic Harm: Exploring the Arguments' (2002) 10 *Torts Law Journal* 41, 48.

70 L Andrews, 'The Genetic Information Superhighway: Rules of the Road for Contacting Relatives and Recontacting Former Patients' in B Knoppers (ed), *Human DNA: Law and Policy* (1997) Kluwer Law International, The Hague, 133, 138–139. Andrews also notes that '[s]imilar cautions are appropriate with respect to recontacting patients when new genetic information is available about them': L Andrews, 'The Genetic Information Superhighway: Rules of the Road for Contacting Relatives and Recontacting Former Patients' in B Knoppers (ed), *Human DNA: Law and Policy* (1997) Kluwer Law International, The Hague, 133, 139.

71 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002. Other submissions shared this perspective: Queensland Government, *Submission G274*, 18 December 2002; NSW Health Department, *Submission G303*, 13 January 2003. The AMA expressed concern that a duty to warn would result in many unnecessary breaches of patient confidentiality where the health professional fears legal repercussions if he or she does not disclose the information: Australian Medical Association, *Submission G212*, 29 November 2002.

could make it more likely that courts in the future may impose positive duties to inform genetic relatives, the Inquiry does not support the development of a duty to warn.⁷²

Application of privacy legislation to disclosure

21.52 The *Privacy Act* imposes constraints on the disclosure of genetic information by doctors and other health professionals to genetic relatives of their patients.⁷³ In particular, the *Privacy Act* appears to prohibit the disclosure of clinically relevant information to genetic relatives in circumstances where it is possible to argue there would be no breach of ethical or common law duties of confidentiality.⁷⁴

Disclosure to prevent 'serious and imminent' threat

21.53 Under the *Privacy Act*, disclosure of genetic information other than for the primary purpose of treating the person tested is generally permitted only with the consent of that person.⁷⁵ Where consent is not obtained, a health services provider may generally disclose personal information to a relative only if this is necessary to lessen or prevent a 'serious and imminent threat' to an individual's life, health or safety.⁷⁶ This formula may have been derived from existing case law relating to the duty of confidentiality,⁷⁷ but appears to leave less room for flexibility than is provided by the range of exceptions to the common law duty.

21.54 In most situations the consequences of someone not knowing about a genetic predisposition to illness may not be a sufficiently imminent threat to their life, health or safety to justify disclosure. The Office of the Federal Privacy Commissioner (OFPC) stated that:

The threat is 'imminent' if it is about to occur. This test could also include a threat posed that may result in harm within a few days or weeks. It is much less likely to apply to situations where the risk may not eventuate for some months or longer.⁷⁸

21.55 Legislation in the Australian Capital Territory, Victoria and New South Wales includes information privacy principles that also incorporate reference to 'serious and imminent' threat or risk.⁷⁹ The Australian Health Ministers' Advisory

72 The Commonwealth Department of Health and Ageing stated that it would be concerned to ensure the proposed legislative change would not give rise to a duty to warn in tort: Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

73 The relevant provisions of the *Privacy Act* (NPP 2) also apply to the 'use' of information. As the distinction between use and disclosure has no particular significance in the present context, the following discussion refers only to disclosure.

74 This position may also apply under state and territory privacy legislation.

75 See *Privacy Act 1988* (Cth) NPP 2.1 (a)–(b).

76 *Ibid* NPP 2.1 (e)(i).

77 For example, *Duncan v Medical Practitioners Disciplinary Committee* [1986] 1 NZLR 513, *W v Egdell* [1990] 1 All ER 835.

78 Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 19.

79 *Health Records (Privacy and Access) Act 1997* (ACT) Privacy Principle 10(1)(c); *Health Records Act 2001* (Vic) Health Privacy Principle 2.2(h); *Health Records and Information Privacy Act 2002* (NSW) Health Privacy Principle 11(1)(c).

Council Draft National Health Privacy Code, released in December 2002, retains this formulation.⁸⁰

Disclosure 'required or authorised by or under law'

21.56 The *Privacy Act* permits disclosure where 'required or authorised by or under law'.⁸¹ It appears to be accepted that 'law' may include the common law.⁸² However, it is not entirely clear whether NPP 2.1(d) permits a doctor to disclose confidential information where the disclosure is covered by the public interest exception to the common law duty of confidentiality.

21.57 In an Attorney-General's Department information paper, the Government acknowledged that the health profession had a strong respect for the confidentiality of health information and maintained sound privacy practices. The paper stated that the 'legislation is not intended to interfere with those professional values and standards'.⁸³

21.58 On the other hand, the Explanatory Memorandum to the Privacy Amendment (Private Sector) Bill 2000 (Cth) stated that this provision of the *Privacy Act* was intended to cover situations where a law 'unambiguously' requires or authorises the use or disclosure of personal information.⁸⁴ The application of the *Privacy Act* to disclosure of health information by doctors and other health professionals, in circumstances that may not breach common law or ethical requirements of confidentiality, may require clarification.

80 Australian Health Ministers' Advisory Council National Health Privacy Working Group, *Draft National Health Privacy Code* (2002), AHMAC, Canberra, National Health Privacy Principle 2.2(h)(i).

81 *Privacy Act 1988* (Cth) NPP 2.1(d). Similar issues are raised under state and territory privacy legislation. The ACT and Victorian legislation include information privacy principles that refer to disclosure required or authorised by law: *Health Records (Privacy and Access) Act 1997* (ACT) s 10(1)(d); *Health Records Act 2001* (Vic) Health Privacy Principle 2.2(c). The Victorian exception appears broader than the ACT or Commonwealth provision and refers to disclosure 'required, authorised or permitted, whether expressly or impliedly, by or under law'.

82 Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 20.

83 Commonwealth Attorney-General's Department, *Privacy Amendment (Private Sector) Act 2000: Information Paper* (19 February 2002), <www.law.gov.au/privacy/royalinfo.html>, 19 February 2002.

84 The Explanatory Memorandum went on to note that '[t]here could be some situations where the law requires some actions which, of necessity, involve particular uses or disclosures, but this sort of implied requirement would be conservatively interpreted': Explanatory Memorandum to the Privacy Amendment (Private Sector) Bill 2000 (Cth).

The Privacy Act and the duty of confidentiality

21.59 A disclosure that is permitted by the *Privacy Act* may none the less breach common law duties of confidentiality.⁸⁵ In general, legislation is presumed not to alter common law doctrines unless a contrary intention is expressed.⁸⁶ The *Privacy Act* does not express any intention to do so.⁸⁷

21.60 Further, the interests protected by duties of confidentiality do not coincide with those protected by privacy legislation. Duties of confidentiality generally arise in the context of specific relationships,⁸⁸ most relevantly between health professionals and their clients. Medical confidentiality rests on the premise that there is a public interest in patients being candid with their doctors. Statutory privacy rights are not so limited.

21.61 The ambit of the duty of confidentiality and the *Privacy Act* also differ. For example, duties of confidentiality apply only to information that is ‘confidential in quality’.⁸⁹ The *Privacy Act* applies to all personal information, whether of a confidential nature or not. In general terms, the common law duty of confidentiality may be breached where there is an unauthorised use or disclosure of the information covered by it.⁹⁰ The *Privacy Act* deals not only with the use or disclosure of personal information, but also with the collection, quality, security and transborder data flow of information, as well as individuals’ rights of access to information about them.

21.62 Finally, medical duties of confidentiality may extend beyond the death of the individual to whom the duty is owed,⁹¹ but constraints on the use and disclosure of personal information under the *Privacy Act* apply only to information about a living person.⁹² For these reasons, it seems unlikely that the *Privacy Act* was intended to codify or alter the common law relating to duties of confidentiality.⁹³

85 One submission to the Inquiry noted that ‘[a] latent complexity is whether the common law of confidentiality and privacy exists alongside the Act or whether it is a complete and exhaustive regime of privacy obligations and rights’: K Liddell, *Submission G141*, 23 March 2002.

86 See eg D Pearce and R Geddes, *Statutory Interpretation in Australia* (5th ed, 2001) Butterworths, Sydney, 145–151.

87 The only mention of the common law appears in s 91 of the *Privacy Act*. This provision states that, for the purposes of Pt VIII (which creates rights to recover damages for breach of certain obligations of confidence owed by Commonwealth officers), common law and equitable obligations of confidentiality continue to exist.

88 But not exclusively. For example, the High Court has held that confidentiality applies where the person acquiring information ought reasonably to consider their conscience bound—it is not dependent on a prior existing relationship: *Moorgate Tobacco Co. Limited v Philip Morris Limited* (1984) 156 CLR 415, 438; K Liddell, *Submission G141*, 23 March 2002.

89 *Commonwealth v John Fairfax & Sons Ltd* (1980) 147 CLR 39, 51.

90 That is, where the information is used for a purpose inconsistent with the purpose for which consent was expressly or impliedly given: J Hamblin and D Bell, ‘Confidentiality’ in J Golden and D Grozier (eds), *The Laws of Australia* (1994) Law Book Company Limited, Sydney, vol 20, [6].

91 *Ibid*, vol 20, [6]. L Skene and J Savulescu, ‘Who Has the Right to Access Medical Information from a Deceased Person? Ethical and Legal Perspectives’ (2000) 8 *Journal of Law and Medicine* 81, 82.

92 *Privacy Act 1988* (Cth), s 6 definitions of ‘personal information’ and ‘individual’.

93 This opinion is supported by the fact that recent Victorian and New South Wales health privacy legislation provides specific protection against actions for breach of confidence where access to information has been provided to the subjects of that information, but not in the case of other use or

21.63 The fact that a disclosure permitted by the *Privacy Act* may none the less breach common law duties of confidentiality is important in assessing the implications of amending the *Privacy Act* to broaden the circumstances in which health professionals may disclose genetic information. At present, the *Privacy Act* appears to be more restrictive of disclosure than the common law, depending on the uncertain scope of the public interest exception to the common law duty of confidentiality and the relevant provisions of the *Privacy Act*.

21.64 The Inquiry's recommended reform of the *Privacy Act* (Recommendation 21-1 below) may reverse this position with respect to the disclosure of genetic information, and does not foreclose the possibility that disclosure permitted by the Act may be actionable at common law.⁹⁴ The Inquiry leaves open the question of whether health professionals may, in future, require some form of statutory protection from actions for breach of confidentiality based on disclosure of confidential information in order to warn genetic relatives about their genetic risk.

Problems with the 'serious and imminent' test

21.65 The effect of NPP 2.1(e)(i) of the *Privacy Act* is that a health professional may disclose personal information to a genetic relative only if this is necessary to lessen or prevent a 'serious and imminent threat' to an individual's life, health or safety.⁹⁵

21.66 There are strong arguments that the 'serious and imminent threat' test may be too restrictive. The requirement that a threat be 'serious and imminent' is not likely to be met by many known genetic conditions—given that most will take time to manifest.⁹⁶ The fact that, in the case of some genetic conditions, the threat is not to a living individual but to future children, may also prevent genetic risks from satisfying this requirement.

21.67 The application of the 'serious or imminent threat' test to genetic conditions is considered to be problematic by Australian and overseas commentators.

The first aspect of this question is the degree of likelihood of having a genetic condition. That is, is a 50 percent likelihood of carrying a mutation of the BRCA1 gene sufficiently serious? The second element is the degree of likelihood that the genetic predisposition will lead to the actual physical manifestation of the disease. In other words, if having a particular mutation is associated with a 60 percent risk of

disclosure of health information: See *Health Records Act 2001* (Vic) s 86; *Health Records and Information Privacy Act 2002* (NSW) s 72(2). The Victorian Health Privacy Principles provide that health information may be used or disclosed for the investigation of unlawful activity and for law enforcement purposes only if such use or disclosure would not be a breach of confidence: See *Health Records Act 2001* (Vic) Health Privacy Principle 2.2(i)–(j).

94 On the other hand, permitting disclosure under the *Privacy Act* of genetic information to genetic relatives in a wider range of circumstances may make it more likely that, in future, courts may accept that disclosure of genetic information falls within the public interest exception to the duty of confidentiality.

95 *Privacy Act 1988* (Cth) NPP 2.1(e)(i).

96 D Bell and B Bennett, 'Genetic Secrets and the Family' (2001) 9 *Medical Law Review* 130, 143 referring to *Privacy Act* IPP 10, which also contains the words 'serious and imminent'; R Magnusson, *Submission G039*, 10 January 2002.

developing a particular condition then is this sufficiently serious? Does it matter that this represents the known lifetime risk, rather than some imminent health risk? Other questions include whether the notion of harm should refer only to an individual's health or also to situations of reproductive decision making?⁹⁷

21.68 Dr Finlay Macrae reserved special criticism for this constraint on disclosure. He stated that those who operate the Victorian FAP Register

feel disappointed and frustrated when their work is truncated by these ethical barriers to disclosure of life saving information—albeit not an imminent threat. The lack of imminency precludes more direct contact [with genetic relatives] at that stage according to the Privacy Laws, but does not preclude the development of cancer at a later stage which is not less lethal for its lack of imminency, and no less destructive within the family.⁹⁸

21.69 Another clinical geneticist argued that the right to individual genetic privacy 'should not include the right not to disclose the information to another who would be harmed by such non-disclosure' and that the obligation to disclose relevant information to genetic relatives should be more strongly emphasised.⁹⁹ The HGSA stated that, with regard to highly penetrant heritable genetic disorders, doctors should consider the situation of the person's relatives and try to ensure that they are informed if they are at increased risk.¹⁰⁰

Reform options

21.70 The Inquiry has considered a range of options to reform the application of the *Privacy Act* to disclosure to genetic relatives. These options include:

1. amending NPP 2.1(e)(i) to change the 'serious and imminent threat' test to a more permissive formulation; or
2. enacting a new NPP 2.1(e)(iii) to permit organisations to exercise a discretion, subject to guidelines issued by the NHMRC and approved by the federal Privacy Commissioner, to disclose an individual's genetic information to a genetic relative, where such disclosure could reasonably be expected to lessen or prevent serious harm to the relative,¹⁰¹ or any other individual.¹⁰²

97 T Lemmens and L Austin, 'The Challenges of Regulating the Use of Genetic Information' (2001) 2(3) *Isima: Canadian Journal of Policy Research* 26, 32–33.

98 F Macrae, *Submission G069*, 14 January 2002.

99 J MacMillan, *Submission G015*, 19 November 2001

100 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

101 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; NSW Health Department, *Submission G303*, 13 January 2003.

102 Other options include enacting a new NPP 2.1(e)(iii) to permit disclosure 'in accordance with rules established by competent health or medical bodies that deal with obligations of professional confidentiality' or recommending that a relevant organisation seek a public interest determination under the *Privacy Act*. Neither of these options was the subject of significant support or other comment in submissions or consultations.

21.71 The simplest reform is Option 1. This would involve amending NPP 2.1(e)(i) to permit use or disclosure where necessary to lessen or prevent a serious risk to an individual's life, health or safety, without the requirement that the threat be imminent. An advantage of this approach is that reform need not await the development of guidelines.

21.72 The proposal made to this effect in DP 66¹⁰³ attracted considerable support in submissions and consultations,¹⁰⁴ including from health professional and consumer organisations otherwise concerned to protect medical confidentiality. The Australian Medical Association (AMA) supported the proposal while stating that:

Consistent with the AMA's *Code of Ethics*, however, such disclosure must be clearly justified and, in general, breaching patient confidentiality should always remain 'the exception' and not become 'the rule'.¹⁰⁵

21.73 There was some opposition to the proposal.¹⁰⁶ For example, the Genetic Support Council WA did not support the proposal because

there is limited empirical information to support the view that there is a significant number of cases that fall into this situation. Furthermore, there is likewise little information to sustain the view that with proper counselling, individuals will not disclose such information to genetic relatives.¹⁰⁷

21.74 In DP 66 the Inquiry recognised that simply amending NPP 2.1(e)(i) to change the 'serious and imminent threat' test to a more permissive formulation would have implications beyond the context of genetic information, by permitting disclosure of *any* personal information where disclosure is necessary to prevent or lessen a serious risk to an individual's life, health or safety, even where the threat is not 'imminent'.

103 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 18–1.

104 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; I Robertson, *Submission G209*, 29 November 2002; Australian Medical Association, *Submission G212*, 29 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Submission G281*, 24 December 2002; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Australian Privacy Charter Council, *Submission G304*, 21 January 2003; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002.

105 Australian Medical Association, *Submission G212*, 29 November 2002.

106 Genetic Support Council WA, *Submission G243*, 19 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002.

107 Genetic Support Council WA, *Submission G243*, 19 December 2002.

DP 66 requested comment on the possible wider consequences of such a reform on disclosure of non-genetic health information.¹⁰⁸

21.75 Some submissions favoured Option 2, in part because of concerns about the effect of amending NPP 2.1(e)(i) for disclosure of other categories of personal information.¹⁰⁹ The OFPC favoured Option 2 and stated that there may need to be broader policy arguments to justify weakening existing privacy protection to the extent implicit in Option 1.¹¹⁰

21.76 The New South Wales Health Department, while recognising that the *Privacy Act* should permit disclosure of genetic information to genetic relatives in a broader range of circumstances, stated that

enacting a new NPP to permit organisations to exercise discretion, subject to NHMRC guidelines and approved by a privacy commissioner, may be a preferable option, as it would allow guidelines to be developed specifically for disclosure of genetic risks. Amending privacy legislation to remove the criteria of 'imminence' would allow for disclosure of medical information in a wide range of circumstances, and for a wide range of medical conditions and would weaken the privacy protections currently in place.¹¹¹

Threats to third parties

21.77 The intention of the proposal made in DP 66¹¹² was to permit disclosure where a genetic risk is not necessarily imminent, but failure to disclose could place the life, health or safety of a genetic relative at serious risk.¹¹³ In submissions and consultations it was suggested disclosure without consent might also be justified where disclosure of information to a genetic relative is capable of averting a significant health threat to someone other than a genetic relative.¹¹⁴

21.78 For example, where there is a confirmed case of familial Creutzfeldt-Jakob disease (CJD),¹¹⁵ it may be argued that health professionals should be entitled to disclose to first-degree relatives the fact that a family member carries the gene associated with CJD.¹¹⁶ While there is no treatment or cure for CJD, disclosure may prevent the transmission of CJD to others through the exercise of reproductive choices

108 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [18.110]. It has been suggested that removing the need for a threat to be 'imminent' may also be appropriate where the threat arises from mental health problems: Australian Medical Association, *Consultation*, Canberra, 22 November 2002.

109 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; NSW Health Department, *Submission G303*, 13 January 2003.

110 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

111 NSW Health Department, *Submission G303*, 13 January 2003.

112 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 18-1.

113 *Ibid* [18.108].

114 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Public Health Genetics Working Group, National Public Health Partnership, *Consultation*, Melbourne, 22 October 2002; WA Genetics Council, *Consultation*, Perth, 28 October 2002.

115 Familial CJD is a late-onset disorder with an autosomal dominant pattern of inheritance.

116 *Confidential Submission G168CON*, 12 July 2002.

by the relative or by warning health professionals about the risks of performing certain medical procedures.¹¹⁷

21.79 The existing language of NPP 2.1(e)(i) refers to threats to ‘an individual’s life, health or safety’. It is not necessary that the disclosure lessen or prevent a threat to the person to whom disclosure is made. The Inquiry does not recommend that NPP 2.1(e)(i) be amended to refer expressly to threats to genetic relatives but rather that it should continue to cover threats to any known individual.

Harm from reproductive choice

21.80 Disclosure of genetic information may be capable of averting a significant health threat to an unborn future relative by informing the parents’ reproductive choices such that children are not born with genetic disorders.¹¹⁸ The Inquiry does not favour permitting disclosure solely to avert harm that may result from the exercise of reproductive choices, as suggested in some submissions.¹¹⁹

21.81 The threat of harm through the exercise of reproductive choice is too remote to justify departure from existing privacy protection and duties of medical confidentiality. It is one thing to justify disclosure of information to prevent harm to a known living individual. It is another to attempt to justify it on the basis that an individual *might* be born with certain genetic or other characteristics in circumstances where the individual *might not* have been born if the parent had known certain facts.

21.82 In practice, difficult questions would inevitably arise about whether the prospect of giving birth to a child with a certain genetic condition is a sufficiently serious outcome to justify breaching privacy. Another relevant factor is that there is no community consensus on the ethical considerations surrounding individuals’ decisions about the use of contraception, pre-implantation genetic testing or the termination of pregnancy. Further, some people may not wish to know about relevant genetic risks.

Inquiry’s views

21.83 The Inquiry has concluded that there may be exceptional circumstances in which disclosure by health professionals of genetic information to genetic relatives without the consent of their patient should be permissible and that privacy legislation inappropriately constrains health professionals’ decisions about such disclosures.

21.84 Amending privacy legislation to broaden the circumstances in which health professionals may use or disclose genetic information might be criticised on the grounds that it could encourage breaches of duties of medical confidentiality. The first

117 Such as ophthalmic surgery or neurosurgery: Ibid. Familial CJD may be transmissible by surgical instruments, even when sterilised.

118 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Public Health Genetics Working Group, National Public Health Partnership, *Consultation*, Melbourne, 22 October 2002; WA Genetics Council, *Consultation*, Perth, 28 October 2002.

119 Genetic Health Services Victoria, *Submission G211*, 28 November 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002.

response to this concern is that, as discussed above, a disclosure permitted by the *Privacy Act* may none the less breach common law duties of confidentiality (or be grounds for professional disciplinary action).

21.85 In any case, it is the experience of clinicians and genetic counsellors that, in the vast majority of cases, patients are happy to involve their genetic relatives in the consultation or follow-up process to a genetic diagnosis.¹²⁰ Even if the *Privacy Act* (or common law) were to be reformed to be more permissive of non-consensual disclosure of information to genetic relatives, breaches of confidentiality would not be the norm.¹²¹

21.86 Another concern relates to the need to recognise that some people may not wish to know about their genetic risk. However, if the circumstances in which disclosure is permitted are limited to situations where it is necessary to lessen or prevent a serious risk, it is reasonable to assume that only rarely would individuals not wish to know about the risk. Recognition of a right not to know should not be a barrier to reform in this area but is an important matter to be taken into account in guidelines to assist health professionals to make decisions about disclosure, as discussed in the following section of this chapter.

21.87 The Inquiry recommends that the Commonwealth amend the *Privacy Act* so that use or disclosure of genetic information by a health professional is permitted where the health professional reasonably believes that the use or disclosure is necessary to lessen or prevent a serious threat to an individual's life, health, or safety, even where the threat is not imminent.

21.88 This recommendation could be implemented by either:

- amending NPP 2.1(e)(i) to change the 'serious and imminent threat' test to a more permissive formulation; or
- enacting a new NPP 2.1(e)(iii) to permit organisations to exercise a discretion, subject to guidelines issued by the NHMRC and approved by the federal Privacy Commissioner, to disclose an individual's genetic information to a genetic relative where such disclosure is reasonably believed to be necessary to lessen or prevent serious harm to any individual.

21.89 Consistent amendment is also needed to the equivalent Information Privacy Principle (IPP 11), which applies to doctors or other health professionals working for Commonwealth government agencies. The Inquiry also recommends that state and territory governments consider parallel amendments to state and territory privacy legislation (see Recommendation 7-1 to 7-3).

120 L. Skene, 'Genetic Secrets and the Family: A Response to Bell and Bennett' (2001) 9 *Medical Law Review* 162, 166.

121 See *Ibid*, 166.

Recommendation 21–1. The Commonwealth should amend the *Privacy Act 1988* (Cth) (*Privacy Act*) to permit a health professional to disclose genetic information about his or her patient to a genetic relative of that patient where the disclosure is necessary to lessen or prevent a serious threat to an individual's life, health or safety, even where the threat is not imminent.

Guidelines on disclosure to genetic relatives

21.90 One way to implement the recommendation above would be to enact a new NPP 2.1(e)(iii) to permit organisations to exercise a discretion to disclose genetic information to genetic relatives, subject to guidelines issued by the NHMRC and approved by the federal Privacy Commissioner. Even if this is not the chosen mechanism of reform, guidelines are desirable because it is not possible to provide sufficient guidance in legislation on how health professionals should approach decisions about the disclosure of genetic information to genetic relatives.

21.91 The circumstances in which it may be justifiable to disclose genetic information are not easily defined and may require detailed consideration of specific genetic conditions, including details about their penetrance and expression, the prevention and treatment options, and the extent to which the patient has been counselled to disclose relevant information directly.

21.92 The OFPC observed that an attempt to legislate exhaustively on permissible disclosure 'would be defeated by the multiplicity of situations which could arise and by future scientific advances'.¹²² In comparison, guidelines are capable of accommodating the impact of rapid technological advance in genetic medicine and taking account of the multiplicity of factors relating to risk, or perceptions of risk.¹²³ Other submissions suggested that the exceptions to confidentiality should be governed primarily by ethical codes, rather than prescribed by legislation.¹²⁴

21.93 A further reason that guidelines on the disclosure of genetic information by health professionals are desirable is to provide health professionals with additional protection from complaints or litigation. The fact that a disclosure was made in compliance with established guidelines would be evidence that it accorded with accepted standards of professional practice.

122 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

123 Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002.

124 Centre for Law and Genetics, *Submission G048*, 14 January 2002; M Dunne, *Submission G041*, 17 December 2001; Sydney IVF Limited, *Submission G062*, 14 January 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002.

21.94 Submissions to the Inquiry strongly favoured the further development of guidelines on disclosure of genetic information by health professionals.¹²⁵ The HGSA stated that it would welcome the development of ethical guidelines dealing with the application of privacy law to genetic information, with the Australian Health Ethics Committee's involvement.¹²⁶ The Centre for Law and Genetics stated:

The ethical justification of disclosure of serious disease to third parties is complex enough but in the area of genetics it has caused real concerns for health professionals. The degree of uncertainty and the developmental stage of the issues in this area prevent any more finite recommendations. This is an area that is appropriately dealt with by the development of guidelines that will allow both the clarification and the refinement of views in this area as well as the development finally of the guidelines themselves.¹²⁷

21.95 The Hereditary Bowel Cancer Group of the Cancer Council Victoria suggested that guidelines may even need to be 'gene specific'.

[T]he disease risk (penetrance x risk of inheritance of the gene) to the individual to whom the disclosure is relevant should, in the Group's judgement, be >25%, though some might argue for a lower figure. This risk would be influenced by other factors, including level of morbidity, mortality and preventability.¹²⁸

21.96 Clinical ethics committees might also have a role in giving guidance to individual health professionals in relation to the disclosure of information to genetic relatives.¹²⁹

125 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; Centre for Law and Genetics, *Submission G048*, 14 January 2002; Australian Medical Association, *Submission G212*, 29 November 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Submission G281*, 24 December 2002; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Australian Privacy Charter Council, *Submission G304*, 21 January 2003.

126 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

127 Centre for Law and Genetics, *Submission G255*, 21 December 2002.

128 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002. The AIS Support Group also highlighted the need for gene specific guidelines: Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

129 Some hospitals have clinical ethics committees, whose activities are not defined by the NHMRC. It has been suggested that such committees could provide guidance to clinicians on situations relating to, among other things, non-consensual disclosure of genetic information and the provision of pre-natal testing services: Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

Australian and overseas guidelines

21.97 Existing Australian codes of ethics do not deal with disclosure of genetic information to genetic relatives with any specificity, although an NHMRC information paper on human genetic testing recognised that:

there may be rare circumstances in which a health professional considers that the risk to the health of relatives is sufficiently large, serious, imminent and potentially preventable that consideration should be given to breaching the individual's confidentiality.¹³⁰

21.98 The AMA Code of Ethics recognises that exceptions to the duty of confidentiality 'may arise where the health of others is at risk'.¹³¹ The Code contains no guidance on the application of this provision.¹³² The Centre for Law and Genetics submitted that the Inquiry should ensure that exceptions to medical confidentiality recognised by codes of ethics properly reflect the family context of genetic information. In particular, the Centre suggested that the content of doctors' ethical duties should be revised by the AMA to take account of genetic information.¹³³

21.99 An indication of the possible content of guidelines on the disclosure of genetic information by health professionals can be obtained from guidelines developed elsewhere. An important example of such guidelines are those issued in 1983 in the United States by the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioural Research. The President's Commission proposed that disclosure to genetic relatives should take place only when

- reasonable efforts to elicit voluntary consent to disclosure have failed;
- there is a high probability both that harm will occur if the information is withheld, and that the disclosure of information will actually be used to avert harm;
- the harm that identifiable individuals would suffer would be serious; and
- appropriate precautions are taken to ensure that only the genetic information needed for diagnosis or treatment of the disease is disclosed.¹³⁴

130 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, NHMRC, <www.nhmrc.gov.au/issues/humangenetics.htm>, 19 February 2003, 51.

131 Australian Medical Association (NSW), *Code of Ethics* (1996) AMA [1.3.4].

132 In 1996, the Privacy Commissioner argued that the AMA Code of Ethics should be supplemented with a clearer set of guidelines on what threshold of risk must be satisfied for a disclosure to occur without the patient's permission; the steps that should be followed before making such a disclosure; and the safeguards that should apply to any such disclosures: Federal Privacy Commissioner, *The Privacy Implications of Genetic Testing* (1996), OFPC, Sydney, 18.

133 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

134 President's Committee for the Study of Ethical Problems in Medicine and Biomedical and Behavioural Research, *Screening and Counseling for Genetic Conditions: The Ethical, Social and Legal Implications of Genetic Screening, Counseling, and Education Programs* (1983), Government Printing Office, Washington DC, 44 cited in T Lemmens and L Austin, 'The Challenges of Regulating the Use of Genetic Information' (2001) 2(3) *Isuma: Canadian Journal of Policy Research* 26, 32. These guidelines were

21.100 In 1998 a sub-committee of the American Society of Human Genetics (ASHG) concluded that disclosure of genetic information by physicians to genetic relatives should be permitted in exceptional circumstances.

1. Disclosure should be permitted where: attempts to encourage disclosure on the part of the patient have failed; the harm is highly likely to occur and is serious, imminent, and foreseeable; the at-risk relative(s) is identifiable; and the disease is preventable, treatable, or medically accepted standards indicate that early monitoring will reduce the genetic risk.

2. The harm from failing to disclose should outweigh the harm from disclosure.¹³⁵

21.101 The ASHG statement includes an ‘imminent’ harm test. However, it is also clear from the context that the authors considered that disclosure may be justified in cases where the condition has not yet manifested but may be prevented.

21.102 In November 2001, a report of the Ontario Provincial Advisory Committee on the New Predictive Genetic Technologies recommended that further research be undertaken to determine whether disclosing genetic information to high-risk relatives against an individual’s wishes should be permitted. It was suggested that criteria to permit this disclosure should include the following:

- the interest in informing relatives strongly outweighs the interest in maintaining confidentiality;
- reasonable attempts to elicit voluntary disclosure are unsuccessful;
- there is a high probability of serious and irreparable harm to an identifiable person;
- the disclosure of the information will enable that person to prevent the harm and there is a high probability that the harm will occur if the information is withheld; and
- the disclosure is limited to the information necessary for the diagnosis and treatment of the third party.¹³⁶

21.103 In the United Kingdom, the Human Genetics Commission (HGC) has concluded that disclosure of genetic information without consent for the benefit of family members may be justified where the benefit of disclosure substantially outweighs the patient’s claim to confidentiality. The HGC stated that:

adopted in 1991 by the Science Council of Canada in its recommended guidelines for physician disclosure to third parties: Science Council of Canada, *Genetics in Canadian Health Care* (1991), Minister for Science (Canada), Ottawa, 72–73, cited in T Lemmens and L Austin, ‘The Challenges of Regulating the Use of Genetic Information’ (2001) 2(3) *Isuma: Canadian Journal of Policy Research* 26, 32.

135 American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure, ‘Professional Disclosure of Familial Genetic Information’ (1998) 62 *American Journal of Human Genetics* 474, 474–483. This paper also contains a summary of relevant international ethical standards and statements.

136 The Provincial Advisory Committee on New Predictive Genetic Technologies, *Genetic Services in Ontario: Mapping the Future* (2001), Ministry of Health Ontario, Toronto, 37.

Such disclosure should be on the proviso that (1) an attempt has been made to persuade the patient in question to consent to disclosure; (2) the benefits to those at risk is so considerable as to outweigh any distress which disclosure would cause the patient; and (3) the information is, as far as possible, anonymised and restricted to that which is strictly necessary for the communication of risk.¹³⁷

New NHMRC guidelines

21.104 The Inquiry has concluded that new guidelines on the disclosure of genetic information to genetic relatives are required. The guidelines will need to address the often complex ethical tensions that surround this issue.

21.105 The ethical tension has traditionally been between a principlist approach, which champions respect for individual choice, and utilitarianism, which champions beneficial outcomes for families and communities. Within principlist ethics there are differences of view about the weight to be attached to respect for persons and the individual's choice about disclosure, on the one hand, and to non-maleficence (acting so as to minimise harm), on the other hand. The experiences of families, provided in the form of narratives about how they have dealt with genetic information, are likely to give fresh ethical perspectives. In the development of the recommended guidelines, the status of traditional principles, the influence of family stories, and the increasing awareness of the community's shared genetic heritage, will need to be considered. This is an example of how the nature of genetic information can compel a re-examination of aspects of the ethos of society and the expression of its ethics.¹³⁸

21.106 The form these guidelines should take will depend on the mechanism by which Recommendation 21-1 is implemented. Ideally, there should be a mechanism for recognising such guidelines within the fabric of the *Privacy Act* to minimise the possibility that a disclosure complying with ethical guidelines may nevertheless breach the *Privacy Act*. Amending the *Privacy Act* to provide for disclosure subject to guidelines issued by the NHMRC and approved by the Privacy Commissioner would provide this recognition.

21.107 However, while desirable, complete consistency between the *Privacy Act* and professional ethical guidelines relating to confidentiality is not essential. Health professional ethical guidelines and legislative privacy principles perform different functions. In the context of medical practice, the former focus on promotion of appropriate professional standards of practice and the latter on the rights of individual patients to exercise control over information about them. It is possible for a professional disciplinary body to find that a disclosure of health information constituted unsatisfactory professional conduct on the part of a medical practitioner in circumstances where the *Privacy Act* would permit disclosure. Similarly, a breach of the *Privacy Act* may not necessarily constitute unsatisfactory professional conduct.

137 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 64.

138 See Ch 6.

21.108 The Inquiry has concluded that the NHMRC would be the most appropriate body to issue guidelines on disclosure to genetic relatives,¹³⁹ whether under an amended *Privacy Act* or in accordance with s 7 of the *National Health and Medical Research Council Act 1992* (Cth).¹⁴⁰ The NHMRC has issued guidelines in related areas, notably guidelines for genetic registers.¹⁴¹

Recommendation 21–2. The National Health and Medical Research Council (NHMRC), in consultation with the Office of the Federal Privacy Commissioner, should develop guidelines for health professionals dealing with disclosure of genetic information to the genetic relatives of their patients. The guidelines should address the circumstances in which disclosure to genetic relatives is ethically justified or required, and the need for patients to be counselled about the disclosure of information in these circumstances. The guidelines should be made pursuant to either new provisions of the *Privacy Act* (amended consistently with Recommendation 21–1) or s 7 of the *National Health and Medical Research Council Act 1992* (Cth).

Disclosure by government health authorities

21.109 DP 66 noted an alternative option for reform. Dr Roger Magnusson has suggested that a protocol could provide doctors and other health professionals with legal authority to disclose genetic information to a government health authority, as occurs in the case of HIV/AIDS.¹⁴² It would then be up to that health authority to take steps to inform the genetic relatives about their genetic risks. This option would not require an amendment to the *Privacy Act* or to state and territory privacy legislation because these Acts permit disclosure where ‘authorised by law’.¹⁴³

21.110 Under this approach, disclosure to genetic relatives should be permitted in circumstances where the test results carry clear implications for genetic relatives; early intervention could reduce the burden of disease in genetic relatives (or prevent

139 The Association of Genetic Support of Australasia and the HGSA submitted that there would be advantage in NHMRC and OFPC developing the guidelines jointly: Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002. The Genetic Support Network of Victoria noted that consumer groups should be consulted on any disclosure guidelines: Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

140 *National Health and Medical Research Council Act 1992* (Cth) s 13.

141 These guidelines include extensive guidance for genetic register staff on contacting the family members of registrants: National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra Ch 6. See also National Health and Medical Research Council, *General Guidelines for Medical Practitioners on Providing Information to Patients* (1993) NHMRC, Canberra.

142 For example, health legislation in New South Wales permits the disclosure of the identity of a patient (who poses an HIV transmission risk to third parties) to the Health Department, activating the Department’s public health powers, exercised in accordance with a Departmental protocol: *Public Health Act 1991* (NSW) s 17(3); *Public Health Regulation 1991* (NSW) s 7(2).

143 See *Privacy Act 1988* (Cth) NPP 2.1(d); *Health Records (Privacy and Access) Act 1997* (ACT) s 10(1)(d); *Health Records Act 2001* (Vic) Health Privacy Principle 2.2(c).

transmission to the next generation); the burden of the disease is a substantial one; and counselling to encourage voluntary disclosure has failed.¹⁴⁴

21.111 The Department of Human Services South Australia suggested that the Inquiry should give further consideration to the notifiable diseases model because 'it is not reasonable to ask a patient's doctor to approach relatives, where their patient does not desire this'.¹⁴⁵ The Law Institute of Victoria submitted a detailed proposal for the establishment of an appropriate body, with statutory powers and functions, that would develop guidelines for disclosure and determine, on a case-by-case basis, whether disclosure should be made and, if so, how.¹⁴⁶

21.112 The Inquiry does not favour these options. Such reforms would be cumbersome and resource intensive to implement, requiring the involvement of state government health authorities in order to permit sensible disclosure.¹⁴⁷ Over time, the medical significance of genetic diagnoses for relatives is likely to increase. Involving the state in dealing with decisions about disclosure seems unnecessarily bureaucratic. While this mechanism may be appropriate in view of the public health implications of non-consensual disclosure of an infectious disease like HIV/AIDS, it is not clear that such a restrictive approach is justified in the case of disclosure of genetic information.

Access rights and genetic information

21.113 The *Privacy Act* and other privacy legislation impose constraints on the disclosure of genetic information by doctors and other health professionals to the genetic relatives of their patients. Privacy legislation also provides individuals with rights to access information about themselves.

21.114 NPP 6 of the *Privacy Act* states that, subject to some exceptions, if an organisation holds personal information 'about' an individual, it must provide the individual with access to the information on request.¹⁴⁸ The Act provides a legally enforceable right for patients to obtain access to their medical records held by private medical practitioners.¹⁴⁹ However, because these records may also contain information about genetic relatives (such as their probable genetic status) genetic relatives may also seek access under NPP 6.

144 R Magnusson, *Submission G039*, 10 January 2002. The Queensland University of Technology also submitted that the model for HIV/AIDS testing should be considered a useful and appropriate starting point: Queensland University of Technology, *Submission G109*, 14 March 2002.

145 Department of Human Services South Australia, *Submission G288*, 23 December 2002.

146 Law Institute of Victoria, *Submission G275*, 19 December 2002.

147 However, health professionals involved with genetic registers might, in some circumstances, be appropriate sources of advice, or even decision making, in relation to disclosure of information to genetic relatives.

148 See also *Health Records Act 2001* (Vic) Health Privacy Principle 6.

149 Notwithstanding the decision of the High Court in *Breen v Williams* that there was no basis at common law or in equity to find a general right of patient access to medical records: *Breen v Williams* (1996) 186 CLR 71. State and territory health privacy legislation includes similar provisions: *Health Records Act 2001* (Vic) Health Privacy Principle 6; *Health Records (Privacy and Access) Act 1997* (ACT) s 10.

21.115 In these circumstances, where the information (such as in a family medical history) is ‘about’ a genetic relative who is not a patient, the obligation to provide access to the genetic relative under the *Privacy Act* may conflict with a health professional’s legal and ethical duties of confidentiality with respect to his or her patient.¹⁵⁰

21.116 NPP 6 provides that access may be refused to the extent that ‘providing access would have an unreasonable impact upon the privacy of other individuals’.¹⁵¹ Therefore, in some circumstances, a health professional may be entitled to refuse access to part of the records.¹⁵²

21.117 There are ways in which a health professional may be able to release familial genetic information without disclosing information about any specific person’s own genetic status, for example, by removing the other person’s identifying details.¹⁵³ Health professionals may have latitude, in dealing with requests for access, to recognise a distinction between familial and individual genetic information. Professor Skene has referred to this distinction in the following terms.¹⁵⁴

There is the fact that a mutation is in the family; and the fact that a particular person has tested positive for the mutation. The information that is familial is the first kind. A person’s own genetic status is personal information and should generally be kept confidential in the same way as information concerning the patient’s clinical or surgical history. Whether the person chooses to disclose his or her own genetic status to family members—or even chooses not to know it at all—is a matter for that person alone.¹⁵⁵

21.118 The HGSA suggested that health professionals may need more guidance on dealing with access by family members to genetic information held by them.

Medical practitioners would appreciate guidance on access by family members to genetic health information they hold. Having a separate file for each family member is appropriate and laboratories should not record the results of tests on multiple family members on the one report form. It would be very helpful to have advice on how to handle information in a file that has not been disclosed to the person tested eg. the result of a genetic test that the doctor knows reveals non-paternity because he/she also holds information on other family members or where a laboratory has tested multiple members of a family and records inconsistent results in its report. Also, while a senior next of kin should be entitled to access specific information about a deceased person that could be of use in the health care of other family members, it should not be

150 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [4.96].

151 *Privacy Act 1988* (Cth) NPP 6.1(c). Under the Victorian health privacy legislation, access may be refused if providing access would have an unreasonable impact on the privacy of other individuals and refusing access is accordance with guidelines issued or approved by the Health Services Commissioner: *Health Records Act 2001* (Vic) Health Privacy Principle 6.1(b).

152 The Victorian legislation also enables access to be refused where health information is ‘subject to confidentiality’ within the meaning of s 27 of the *Health Records Act 2001* (Vic).

153 Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 35.

154 See Ch 7 and L. Skene, ‘Genetic Secrets and the Family: A Response to Bell and Bennett’ (2001) 9 *Medical Law Review* 162.

155 *Ibid*, 166.

possible to access the whole medical record, which may contain information that the next of kin does not know about relatives, or paternity and other highly sensitive information.¹⁵⁶

21.119 The Inquiry has concluded that health professionals would benefit from further guidance on how to deal with requests for access to genetic information held by them.¹⁵⁷ This issue might usefully be considered in conjunction with the proposed development by the NHMRC of guidelines dealing with disclosure of genetic information by health professionals to the genetic relatives of their patients.

21.120 The discussion above deals with situations in which the information held by a health professional is clearly ‘about’ the genetic relative who is identifiable from the record. However, another situation must also be considered—that in which a genetic relative needs to obtain access to information that is clinically relevant to them but which is not ‘about’ them in terms of NPP 6. While it may be possible for an individual to argue that a genetic relative’s information is relevant to that individual’s health in its familial aspect, the rights under NPP 6 may not be asserted unless the information also identifies the individual seeking access.¹⁵⁸ The Inquiry has concluded that the *Privacy Act* should be amended to provide specifically that an individual has a right to access familial genetic information relating to his or her genetic relatives in limited circumstances.

21.121 Individuals sometimes need to obtain access to information relating to genetic relatives, either living or dead, in order to facilitate their own diagnosis and treatment. Yet the relatives (or the personal representative of a deceased relative) may decline to consent to the disclosure of this information. Privacy NSW suggested that the *Privacy Act* could be redrafted to

explicitly permit access to both health information and tissue samples of the deceased person by a genetic relative. Whilst it might appear unlikely that a next-of-kin would seek to legally challenge such access, the amendment would provide clarification for patients requiring access and the hospital or research institution which holds the genetic samples and health records.¹⁵⁹

21.122 The implementation of Recommendation 21–1 would permit health professionals to disclose a patient’s genetic information to a genetic relative, without the patient’s consent, where disclosure is necessary to lessen or prevent a serious threat to the life, health, or safety of an individual. Consistently with that position, genetic relatives should also be entitled to exercise rights of access on their own initiative.

21.123 The Inquiry considers that any such right of access should be limited to situations where the information is necessary to lessen or prevent a serious threat to their life, health, or safety¹⁶⁰—that is, in circumstances in which a health professional

156 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

157 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney [18.138].

158 See *Privacy Act 1988* (Cth) s 6(1), definition of ‘personal information’.

159 Office of the Privacy Commissioner (NSW), *Submission G257*, 20 December 2002.

160 Privacy NSW submitted that ‘[a]ccess should be restricted to only those relations for whom the information would be of clinical relevance’: *Ibid.*

would be permitted to disclose the information, under Recommendation 21–1, without any request for access.

21.124 The rights of access should be exercisable only in relation to familial genetic information about the siblings, parents or children of the individual (first-degree genetic relatives) on the basis that genetic information is likely to be of sufficient clinical importance to justify disclosure only if the family relationship is very close. Access should be provided by making the information available to the requester's nominated medical practitioner or genetic counsellor, who can explain the clinical relevance of the information obtained for the individual seeking it.

21.125 Where an organisation receives a request for access to genetic information about an individual's genetic relatives, the organisation should be obliged to first ask the genetic relatives whether they consent to the organisation providing access, unless it is impracticable to do so. Where consent is not given, access could be refused if providing access would have an unreasonable impact upon the privacy of any individual.¹⁶¹

21.126 The other reasons for refusing access set out in NPP 6 should also apply to requests from individuals seeking access to information about genetic relatives, including that the request for access is 'frivolous or vexatious' or that the information relates to legal proceedings.¹⁶²

21.127 The Inquiry recognises that this recommendation, if implemented, would represent a significant extension of the *Privacy Act*. For the first time, the Act would provide certain individuals with rights to access information about other people. However, this policy position is a corollary of the shared or familial nature of genetic information (see Chapter 7).

21.128 As in cases where disclosure by a health professional is capable of preventing a serious threat to the life, health or safety of an individual (see Recommendation 21–1), the Inquiry's recommendation may permit disclosure of genetic information by health professionals that would breach usual duties of confidentiality. However, disclosing information to those relatives when it is clinically relevant to them clearly differs from disclosing it to others. Obligations of confidentiality would remain in respect of the world at large.

21.129 Providing rights to access familial genetic information would not lead to significant or unjustifiable erosion of medical confidentiality. Decisions on access would be made by health professionals who may be expected to have a good understanding of the importance of preserving medical confidentiality and an ability to

161 As is already the case under NPP 6(1)(c). The Inquiry has recommended that the *Privacy Act* should protect information about deceased individuals (see Recommendation 7–6). However, an important factor in determining whether providing access would have an unreasonable impact upon the privacy of any individual is whether or not the individual is deceased. Where the information is primarily about a deceased individual the privacy impact of providing access is likely to be minor and less likely to prevent access.

162 See *Privacy Act 1988* (Cth) NPP 6.1(d), (e).

balance this interest against the clinical consequences of non-disclosure for the individuals involved.

21.130 In order for an individual to request access, he or she would already have to know that the genetic relative was being treated by the particular health professional to whom the request is directed. Health professionals who receive such requests would have ample grounds for refusing to provide access where the information they hold is not important to the health of the individual seeking it.

21.131 The existence of new statutory access rights (and an obligation on health professionals to ask patients to consider consenting to disclosure) may assist health professionals in dealing with conflicting ethical duties to patients and genetic relatives. It may also bring home to patients the fact that they should consider any ethical obligation to share genetic information with their families. Health professionals who chose to disclose information will be better protected from legal action and complaints in relation to the disclosure of familial genetic information.

Recommendation 21–3. The Commonwealth should amend the *Privacy Act* to provide that an individual has a right to access genetic information about first-degree genetic relatives where such access is necessary to lessen or prevent a serious threat to the individual’s life, health, or safety, even where the threat is not imminent. Where an organisation subject to the *Privacy Act* receives a request for access, the organisation should be obliged to seek consent, where practicable, before determining whether to provide access. The right of access should be exercisable only through a nominated medical practitioner or genetic counsellor and may be refused where providing access would have an unreasonable impact upon the privacy of the individual whose information is sought or other individuals. (See also Recommendation 8–4.)

Recommendation 21–4. In developing the guidelines referred to in Recommendation 21–2, the NHMRC should include advice to health professionals in dealing with requests for access to genetic information by the genetic relatives of their patients.

22. Genetic Registers and Family Genetic Information

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Introduction

Families have black sheep, missing sheep and dead sheep.¹

22.1 This chapter deals with the operation of genetic registers and asks whether the existing regulatory framework to protect the privacy of genetic information collected and held on genetic registers is adequate. The collection and disclosure of family genetic information is central to the operation of genetic registers and the conduct of genetic counselling. The familial or collective nature of genetic information raises special issues in this context, which are discussed below.

What are genetic registers?

22.2 The primary purpose of genetic registers is to operate as an effective way of identifying and contacting members of families who are at significantly increased risk of developing an inherited disorder or of having affected children.² Information on a genetic register will generally comprise genetic information about many genetically related people and may also contain genetic samples.

1 L Skene, 'The Genetics Debate: Why Doctors Must be Heard' (Paper presented at 2001 UMMS Lecture, 15 November 2001), quoting Professor Graham Giles, Director of the Cancer Epidemiology Centre, Anti-Cancer Council of Victoria, explaining the need for genetic registers: Lecture to Graduate Students in Law and Human Genetics, University of Melbourne, October 2001.

2 See National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra, 7.

22.3 Health professionals may need to collect information about an individual's family medical history in order to provide effective health care (see Chapter 21). In some ways, genetic registers may be seen as an extension of this established part of medical practice. Family pedigrees³ are documented in greater detail than would be usual in routine medical practice, and are linked to medical information, including genetic test results and tissue samples. The bringing together of pedigrees and medical information about multiple families constitutes a genetic register.

22.4 Each genetic register usually addresses one disorder or a closely related group of disorders. Genetic registers in Australia include those for Huntington's disease, familial adenomatous polyposis (FAP), Duchenne muscular dystrophy and Fragile X mental retardation.⁴

22.5 Genetic registers are designed to contribute to the provision of health care to family members by undertaking the systematic collection of accurate and up-to-date information over a long period; ensuring that family members have an opportunity to become aware of their risk and testing, prevention, treatment and reproductive options; and bringing together pedigree and medical information relating to individuals, nuclear families and branches of the family in order to construct a single large pedigree.⁵

22.6 Genetic registers may improve risk assessment, provide information about disorder expression in the family, prevent duplication of genetic testing, help validate genetic test results and facilitate research.⁶ While genetic registers may be used to facilitate research, they can be distinguished from human genetic research databases (discussed in Chapter 18) because genetic registers are used primarily in the provision of health care. The information on genetic registers may be used, along with information in other medical records,⁷ to assist in genetic counselling. Some registers assist health professionals to provide surveillance for inherited disorders such as familial cancer.

22.7 The National Health and Medical Research Council's *Guidelines for Genetic Registers and Associated Use of Genetic Material*⁸ (the NHMRC Guidelines for Genetic Registers) distinguish genetic registers from statute-based health data collections, including cancer registers. Each State and Territory maintains a statutory cancer register.⁹ The core function of these registers is to measure the incidence or

3 A family pedigree is a genealogical table showing lines of relationship and descent.

4 National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra, 7.

5 *Ibid.*, 17–19.

6 *Ibid.*, 17–19.

7 The NHMRC Guidelines for Genetic Registers note that the following are not considered to be genetic registers: medical records kept by health professionals, hospitals and clinical genetics units; records of research studies, research databases; public health surveillance data sets (such as statutory cancer registers); the results of genetic tests or collections of tissues held by laboratories and blood banks. See *Ibid.*, 8–9.

8 *Ibid.*, 8–9.

9 For example, in Victoria the Cancer Council of Victoria operates cancer registers established pursuant to the *Cancer Act 1958* (Vic) and in NSW the NSW Central Cancer Registry operates cancer registers established pursuant to the *Public Health Act 1991* (NSW).

prevalence of cancers in a defined population,¹⁰ rather than to assist directly in health service provision. This chapter does not deal with statute-based health data collections, the regulation of which is governed by the state and territory laws that establish them.

Existing regulation of genetic registers

22.8 Existing regulation of genetic registers is based on the NHMRC Guidelines for Genetic Registers.¹¹ In 1990, the Human Genetics Society of Australasia (HGSA) issued guidelines dealing with DNA banking, including the storage of genetic samples in association with the operation of genetic registers.¹² These guidelines may be supplemented by other, more specific, guidelines developed by clinical genetics services, and other organisations that operate genetic registers.

22.9 The collection, use and disclosure of genetic information on genetic registers may be subject to information and health privacy legislation, depending on where it is held. Privacy regulation of genetic registers differs according to the state or territory in which the register operates, whether the register is a public or private sector organisation, and the legal resolution of possible conflicts between federal, state and territory privacy laws.

22.10 Regulation of the handling of genetic information by genetic registers is primarily a state and territory matter. However, in the context of this federal Inquiry, this chapter focuses on federal privacy law and leaves it to the States and Territories to assess the implications of the Inquiry's recommendations for genetic registers operating under state and territory privacy regimes, in accordance with Recommendations 7-1 and 7-2.¹³

NHMRC Guidelines for Genetic Registers

22.11 The content of the NHMRC Guidelines for Genetic Registers was summarised in IP 26.¹⁴ In particular, the Guidelines deal in detail with consent to inclusion of genetic information on a register, and list information to be provided to a

10 Some registers have additional functions, such as providing population based cases for case-control or cohort epidemiology studies, or collection of information which can be used to monitor the effectiveness of the treatment and clinical management of cancers.

11 National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra.

12 Human Genetics Society of Australasia, *Guidelines for Human DNA Banking*, Guidelines, 1 July 1990.

13 A recent survey evaluating the compliance of data registers in respiratory medicine with privacy laws illustrates some of the complexities involved in applying privacy laws to voluntary (non-statutory) registers: R Magnusson and C Clarke, 'Data Registers in Respiratory Medicine: A Pilot Evaluating Compliance with Privacy Laws and the National Statement on Ethical Conduct in Research Involving Humans' (2002) 10 *Journal of Law and Medicine* 69. These complexities are equally applicable to the operation of genetic registers.

14 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [8.36]–[8.37].

person before they choose to participate.¹⁵ There are detailed provisions relating to obtaining consent from people other than the initial registrant, such as children and other nominated relatives and in relation to deceased persons.¹⁶ The Guidelines also contain detailed confidentiality provisions¹⁷ and guidelines on contacting other family members.¹⁸

22.12 The NHMRC Guidelines for Genetic Registers have no direct legal effect and do not provide any formal sanction for non-compliance. However, the Guidelines have a status similar to that of the NHMRC *National Statement on Ethical Conduct in Research Involving Humans*,¹⁹ as discussed in Chapter 14. Non-compliance may, therefore, influence National Health and Medical Research Council (NHMRC) advice and recommendations to government on any research funding related to the operation of a genetic register.²⁰ In addition, the Guidelines may be used as a standard in assessing complaints about the conduct of health professionals responsible for genetic registers and are likely to positively influence professional behaviour. As is the case with clinical practice guidelines, courts may approach the guidelines as a form of evidence of accepted standards of practice.²¹

Application of privacy legislation to genetic registers

22.13 The collection, use and disclosure of genetic information on genetic registers may be subject to information and health privacy legislation. The extent to which genetic registers are covered by existing legislation depends on whether they are maintained by federal or state public sector entities or by private sector organisations.

22.14 Genetic registers are most commonly hospital based or community based. Where registers are established and maintained as part of the services provided by a public hospital they may be covered by state privacy legislation such as the *Health Records (Privacy and Access) Act 1997* (ACT), the *Privacy and Personal Information Protection Act 1998* (NSW), *Health Records and Information Privacy Act 2002* (NSW), or the *Health Records Act 2001* (Vic).

15 This information includes: the fact that participation is voluntary; the aims of the register and how it may help the individual and relatives; how the register operates; relationships between registrants and register staff; policy about communicating new research information; registrant's role in introducing other family members to the register; what is on the register; how long it will be held; the sources from which register information is derived; procedures to ensure confidentiality; rights of access; and uses of register data and any stored genetic material: National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra, 17–19.

16 Ibid, 20–22.

17 Ibid, 23–27.

18 Ibid, 29–32.

19 National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra.

20 The NHMRC Guidelines for Genetic Registers mainly describe use of registers for diagnosis and treatment but use for research is contemplated: National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra [1.2(b)(ii)].

21 See J Pelly and others, 'Clinical Practice Guidelines Before the Law: Sword or Shield?' (1998) 169 *Medical Journal of Australia* 330; B Bennett, 'Setting the Standard? Clinical Practice Guidelines and Medical Negligence Litigation' (1997) 20 *University of New South Wales Law Journal* 707.

22.15 Genetic registers maintained by community based organisations, such as disease support groups, are covered by the private sector amendments to the federal *Privacy Act 1988* (Cth) (*Privacy Act*), as are registers maintained by private sector health professionals or organisations.

22.16 Some genetic registers are maintained by state and territory cancer councils. For example, the New South Wales Cancer Council maintains hereditary bowel cancer registers. Where such bodies are established for a public purpose under a law of a State they will not be covered by the *Privacy Act*²² but may be covered by state legislation.

22.17 In their submission, Professor Nick Saunders and Associate Professor Paul Komesaroff observed that a genetic register may take the form of a network of data sources that links information stored in a number of locations, such as hospital records, pathology laboratories and research institutes. They noted that

in such settings it is important that an individual is identified who assumes responsibility for ensuring security of the data and for discharging the other responsibilities normally associated with the conduct of a register.²³

22.18 This may also be important in order to establish which laws apply to the collection, use and disclosure of the information on the particular genetic register.

Is there any need for further regulation?

22.19 The Inquiry asked whether, in the specific context of genetic registers, federal, state and territory privacy laws provide an adequate framework for protecting the privacy of genetic samples and information.²⁴

22.20 One submission raised concerns about the privacy of information contained on a Huntington's disease genetic register.²⁵ The submission stated that identifying information had been transferred in the 1980s from a research institute to a Brisbane hospital, without the knowledge or consent of the individuals to whom the information related.

22.21 The *Privacy Act* would generally require consent to be obtained prior to such a transfer of health information.²⁶ However, if the organisations involved are state authorities,²⁷ the *Privacy Act* may not apply to the collection or disclosure of the

22 *Privacy Act 1988* (Cth) s 6C(1), 6C(3)(c).

23 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

24 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney, Question 9–2.

25 *Confidential Submission G126CON*, 12 March 2002.

26 Or, at least, prior to the collection of the information by the new organisation: see Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 47–48.

27 In terms of the *Privacy Act 1988* (Cth) s 6C(3).

information being transferred²⁸—emphasising the need for harmonisation of federal, state and territory privacy laws (see Chapter 7).

22.22 The Inquiry has not been made aware of any other specific concerns about the privacy of genetic samples and information held on genetic registers. The HGSA observed that, in fact, there are very few genetic registers in Australia at the present time and that ‘participation in such registers is with consent’. The HGSA considered that

professional ethics/codes of conduct and mechanisms for censure of health professionals, the Privacy Acts and associated penalties for breach, State privacy laws/regulations, guidelines from the NHMRC and other bodies, the risk management practices of hospitals and the common law appear to provide adequate protection for genetic information collected in the context of genetic registers.²⁹

22.23 A similar view was expressed by the Centre for Law and Genetics, which stated that it was not aware of any specific breaches of privacy in dealings with established genetic registers governed by the NHMRC Guidelines for Genetic Registers. The Centre considered that genetic registers and their operation should remain under the regulation of codes of ethical practice.³⁰

22.24 However, submissions have focused on a number of practical problems concerning the way in which the *Privacy Act* and other privacy legislation may overly constrain the operation of genetic registers and the conduct of genetic counselling. These concerns are discussed below in relation to the collection, de-identification, use and disclosure of information on genetic registers.

Collection of information on genetic registers

22.25 Issues concerning the collection of information for inclusion on genetic registers were raised in several submissions.³¹ For example, the HGSA referred to the collection of information needed to construct pedigrees and stated that

it is essential there be clarification on the use of family data by medical practitioners and other involved health professionals. The gathering of family information to construct a family tree, without the consent of the other family members, is essential to the diagnosis and risk assessment in families. The use of these data by registers is of great value in the management of other family members. The HGSA believes it should be made clear that obtaining family information in this context is considered an exception to the Privacy Act.³²

22.26 The Peter MacCallum Cancer Institute (PMCI) also expressed concerns that the *Privacy Act* could make the collection of pedigree information difficult.

28 Depending on where registers are established and maintained, state or territory privacy legislation may apply.

29 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

30 Centre for Law and Genetics, *Submission G048*, 14 January 2002.

31 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002;

32 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

It seems that a pedigree constructed from information provided by an individual will become part of his/her medical records, but cannot be verified or used during consultation with another family member. Does this mean a fresh pedigree must be drawn for every family member who attends a genetic clinic? Will the genetic risk of an individual be assessed only on the basis of the information provided by that individual rather than, as now, on a fully verified family history drawn from many sources? How are fully verified pedigrees to be obtained under the Act? Will there be a need to tag the sources of the information used to construct the pedigree so as to minimise the chances that information volunteered by one family member would be transmitted to another? This seems unworkable. At the very least there is a need for a clearer definition in the Paper of the genetic information that blood relatives and other family members *should* be able to supply and the character of the access to that information by clinicians and genetic clinics that is appropriate.³³

Collection of information and the NHMRC Guidelines

22.27 The NHMRC Guidelines for Genetic Registers note that potential registrants may be identified in different ways. They may contact the register themselves on the advice of health professionals or community based support groups. However, they may also be identified by register staff from the information provided by another registrant who identifies other family members.³⁴

22.28 The NHMRC Guidelines for Genetic Registers recognise that the collection and recording of family genetic information in a genetic register may involve a breach of the privacy of those family members who have not consented to be registrants. They state that registers should distinguish information that identifies people who are not registrants and not disclose it in identifiable form without the consent of the identified person. In general, the persons identified should be approached, in due course, so that their consent can be sought for inclusion of their information in the register.³⁵

22.29 However, in some circumstances it appears that collection of information for genetic registers, in ways that would comply with the NHMRC Guidelines for Genetic Registers, may nevertheless breach the *Privacy Act* or similar state and territory privacy legislation, as discussed below.³⁶

Application of privacy legislation to collection

22.30 National Privacy Principle (NPP) 1 of the *Privacy Act* provides that, in general, an organisation must collect personal information about an individual only from that individual, rather than from any third party, unless it is not 'reasonable and practicable' to do so.³⁷ Further, under NPP 10, an organisation generally must not collect sensitive information (including genetic and other health information) unless the individual has consented.

33 Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002.

34 National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra, 13–14.

35 *Ibid.*, 21–22.

36 The NHMRC Guidelines for Genetic Registers pre-date the commencement of relevant provisions of the *Privacy Amendment (Private Sector) Act 2000* (Cth).

37 *Privacy Act 1988* (Cth) NPP 1.4.

22.31 The federal Privacy Commissioner's *Guidelines on Privacy in the Private Health Sector* imply that in some circumstances consent may be obtained on an 'opt out' basis:

Where consent is required from individuals for the collection and use of data in relation to the establishment and maintenance of a disease register, it may sometimes be appropriate to give individuals the opportunity to opt out of being included on the register. The use of this approach by a health service provider would only be appropriate where individuals are clearly informed about the option to opt out and this is prominently presented and easy to adopt.³⁸

Collection from registrants

22.32 In some circumstances it may not be 'reasonable and practicable' to collect family medical history information directly from the genetic relatives, for example, because they are not contactable or are dead. However, in other circumstances the collection of family medical history information without the consent of those family members, for the purposes of a genetic register, might breach the *Privacy Act* because it is reasonable and practicable to obtain consent. The fact that consent may later be sought from these family members, whether by the registrant or staff of the genetic register, does not appear to alter this position.

22.33 As discussed in Chapter 21, Public Interest Determinations (PIDs) have been issued by the federal Privacy Commissioner to ensure that organisations can continue to collect family medical history information without breaching NPP 10.1 where the collection is

- necessary for the organisation to provide a health service directly to the consumer; and
- the third party is a member of the consumer's family or household, or the third party's information is otherwise relevant to the consumer's family, medical or social history.³⁹

22.34 The PIDs apply only to situations where a health service is being provided directly to the individual (the patient) from whom family history information is sought.⁴⁰ This criterion is likely to be satisfied where the information is being collected, for example, by the patient's treating doctor for the patient's own medical records.

22.35 However, where information is collected in order to facilitate the diagnosis or treatment of other, sometimes unknown, people and to ensure that genetic relatives have the opportunity to become aware of their genetic risk, or to access genetic

38 Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, xiii.

39 *Privacy Commissioner Public Interest Determination No. 9 2002* (Cth); *Privacy Commissioner Public Interest Determination No. 9A 2002* (Cth).

40 Recording health information may, in itself, constitute the provision of the health service: Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003. See also *Privacy Act 1988* (Cth) s 6(1) definition of 'health service'.

counselling and medical advice, the PIDs may not apply. This has important implications for the collection of third party information for inclusion on genetic registers.

Collection from other health professionals

22.36 Another issue concerning the collection of information for genetic registers relates to the verification of information provided by registrants. The NHMRC Guidelines for Genetic Registers state that confirmation of diagnosis may sometimes be obtained without consent where the relative is deceased or if there are circumstances in which it would be intrusive or potentially upsetting to seek consent.⁴¹

22.37 The Cancer Council Victoria Cancer Genetics Advisory Committee stated that where it is necessary to verify diagnoses to provide adequate risk assessment and screening recommendations, there needs to be a mechanism 'where public health genetic registers can undertake this verification process without fear of litigation'.⁴² However, privacy law may create problems for genetic registries where there is a need to verify diagnoses without consent.⁴³ The collection of information in this way may breach the *Privacy Act* and may not be authorised by existing PIDs.⁴⁴

22.38 Verification of diagnosis is vital to the operation of a genetic register.⁴⁵ However, some submissions had reservations about whether any genetic register should be permitted to verify health information with other health professionals without the consent of the individual involved.⁴⁶

22.39 The Cancer Genetics Advisory Committee noted that the legal position with regard to the disclosure of verifying information by statutory cancer registers, such as the Victorian Cancer Registry, may also need to be considered.⁴⁷ Such registers are not necessarily authorised to disclose verifying information to genetic registers.⁴⁸

De-identification of family information on genetic registers

22.40 The NSW and ACT Hereditary Bowel Cancer Register expressed a related concern about the application of the NHMRC Guidelines for Genetic Registers.⁴⁹ This concern relates to requirements to de-identify information where register staff have not

41 National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra, 22.

42 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

43 Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002.

44 Although the *Privacy Act* does not apply to the collection of personal information about a deceased individual. See *Privacy Act 1988* (Cth) s 6: an individual means a natural person. See, however, Recommendation 7–6.

45 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

46 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002.

47 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

48 Information held on the Victorian Cancer Registry may only be released 'for the purposes of medical research or the administration of cancer related public health programs' and with ethics committee approval: *Cancer Act 1958* (Vic) s 61A.

49 NSW and ACT Hereditary Bowel Cancer Register, *Submission G079*, 16 January 2002.

been able to obtain consent to retain identified information provided by a registrant about a relative.

22.41 The NHMRC Guidelines for Genetic Registers provide that, where consent is not obtained, register staff should remove identifiers from the information. The information that remains in the register, usually represented by a symbol in the pedigree, will be the position of the individual in the pedigree, the individual's gender, whether or not the individual was considered to have the genetic disorder and whether the individual is alive or dead.⁵⁰ The Guidelines also state:

At the time of constructing the pedigree there will be relatives of the registrant in whom register staff have no professional interest other than to confirm that the diagnosis provided by the registrant is correct. If the relative is deceased or if there are circumstances in which it would be intrusive or potentially upsetting to approach a living relative for consent, confirmation of diagnosis may be obtained without consent provided that identifiers are removed once the confirmed diagnosis has been recorded in the register.⁵¹

22.42 The Hereditary Bowel Cancer Register submitted that retention of identified information may be important in order to link affected families.

One of the unique functions of a central repository of genetic information is that information from families thought to be separate units may be linked so they are shown to be part of a single extended family ... The aggregation of families is an important role of the Hereditary Bowel Cancer Register because it:

- assists with identifying those at risk;
- provides information about disorder severity and manifestations in that family;
- prevents unnecessary genetic testing if another family member has already been tested;
- helps validate genetic test results;
- facilitates research.⁵²

22.43 The Hereditary Bowel Cancer Register stated that, because of the large number of families on genetic registers, linking is now done initially by computer searches of names. It submitted that de-identification of information about individuals who have not consented to be on a register 'would make it extremely difficult to link families'.⁵³

22.44 At least for the purposes of the *Privacy Act*, information on a genetic register may not be truly de-identified even where identifiers, such as name and date of birth have been removed. The information remains 'personal information'⁵⁴ because it is about an individual whose identity can reasonably be ascertained from the surrounding

50 National Health and Medical Research Council, *Guidelines for Genetic Registers and Associated Genetic Material* (2000), NHMRC, Canberra 22.

51 *Ibid.*, 22.

52 NSW and ACT Hereditary Bowel Cancer Register, *Submission G079*, 16 January 2002.

53 *Ibid.*

54 In terms of the definition in *Privacy Act 1988* (Cth) s 6(1).

information—the fact that the person is, for example, the father, mother, or sibling of another individual whose full details are on the register.⁵⁵

22.45 In DP 66, the Inquiry asked for comments on the de-identification requirements contained in the NHMRC Guidelines for Genetic Registers and whether any modifications were necessary.⁵⁶ In response, the Cancer Council Victoria Cancer Genetics Advisory Committee submitted that

[d]e-identification of pedigrees (where there is no consent to registration) substantially restricts the effective functioning of registers, such as the FAP register. It is precisely these family members who potentially would benefit most from timely intervention as a result of informed counselling through and by the register. Those registered are usually already within the health care system.⁵⁷

22.46 The Cancer Genetics Advisory Committee proposed that a PID for genetic registers should permit the collection and retention of information about identified individuals unless consent has been denied, in which case it should be de-identified.⁵⁸ Similarly, the Walter and Eliza Hall Institute of Medical Research observed that:

The new privacy laws can be cumbersome, as permission has to be obtained to record identifying features for every individual who is included in a pedigree. If permission is not obtained, the individual has to be deidentified. Sometimes this is absurd, as one could identify an anonymised individual on the basis of where they sit in a pedigree. The [PID] should be less prescriptive and allow for minimal identifying data to be placed on pedigrees, with the caveat that the pedigrees be only used for the purposes for which they were initially approved ...⁵⁹

22.47 The HGSA agreed that the requirements for the de-identification of information on genetic registers contained in the NHMRC Guidelines for Genetic Registers may cause problems and submitted that:

A distinction needs to be made between the situation in which consent has been refused and the situation in which there has been no response to a request for consent. The existing NHMRC guidelines remain appropriate in the former situation.⁶⁰

Use and disclosure of information on genetic registers

22.48 A related concern, raised by the PMCI and Dr Graeme Suthers,⁶¹ involves the use of family genetic information provided by one individual in the diagnosis or treatment of another genetically related family member who is attending the same clinical genetics service. Dr Suthers, the Head of the Familial Cancer Unit at the South

55 From the definition of personal information in the *Privacy Act s 6(1)*.

56 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Question 19–2.

57 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

58 *Ibid.*

59 Victorian Breast Cancer Laboratory — Walter and Eliza Hall Institute of Medical Research, *Submission G258*, 20 December 2002.

60 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

61 Peter MacCallum Cancer Institute, *Submission G104*, 20 February 2002; G Suthers, *Submission G026*, 30 November 2001.

Australian Clinical Genetics Service, provided the following example of such a situation:

Both Brendon and Bradley have been tested by me. As far as they know, they have no living relatives who would be at risk of having inherited this mutation. However, a long lost cousin, Claude, is seen by a doctor in another department of my hospital. Claude says that he had a relative called Bradley whose brother had early-onset bowel cancer, but he cannot remember Brendon's name. The further medical care of Claude would be greatly enhanced if the doctor seeing him could find out details of Brendon, Bradley, and the genetic testing that is now available. Should he ask me? Should the doctor have access to my database?⁶²

22.49 Dr Suthers noted that this issue often arises within the South Australian Clinical Genetics Service, which has a number of discrete, yet related, units sharing a common database for reasons of cost and to assist in managing familial disorders.

It is very common for one unit to have data about one end of a family, and another unit to have relevant information about another branch of the family. Should this information be shared?⁶³

22.50 Dr Suthers also noted constraints on the disclosure of such information to the treating doctors of other family members. Using the facts described above, Dr Suthers noted that:

If Claude's doctor was *not* part of our genetics service, he would not have access to our database, and I would not be able to release any information to him about Bradley and Brendon. He would not necessarily know that I had this information. In practice, we are the main repository of familial cancer information in the State, and Claude's doctor would probably call me. I could tell him non-identified information about the family and recommend that Claude be referred to our Service for genetic counselling and testing. But—as noted above—when a family has a limited number of affected people, it is impossible to avoid identifying the person with a familial mutation.⁶⁴

22.51 Under the *Privacy Act*, use or disclosure of health information for a secondary purpose is permitted if the secondary purpose is directly related to the primary purpose of collection⁶⁵ and the individual would reasonably expect the organisation to use or disclose the information for the secondary purpose⁶⁶ or where the individual has consented to the use or disclosure.⁶⁷

62 G Suthers, *Submission G026*, 30 November 2001.

63 *Ibid.*

64 *Ibid.*

65 In the *Guidelines on Privacy in the Private Health Sector*, the Office of the Federal Privacy Commissioner has indicated that the Commissioner would consider a reasonable interpretation of 'primary purpose' in the health context to be the 'main and dominant reason' a health service provider collects information: Office of the Federal Privacy Commissioner, *Guidelines on Privacy in the Private Health Sector* (2001), OFPC, Sydney, 12.

66 *Privacy Act 1988* (Cth) NPP 2.1(a).

67 *Ibid* NPP 2.1(b).

22.52 The requirement that a secondary purpose be directly related and within the reasonable expectations of the individual appears to restrict the use or disclosure of genetic information collected from one patient in the diagnosis and treatment of another.

22.53 However, there may be no reason why this cannot be addressed by routinely seeking consent from patients to the use or disclosure of their family genetic information for the diagnosis and treatment of other patients of the same clinic or hospital. Even broader consent could be sought to the use or disclosure of family genetic information in the treatment or diagnosis of other family members, including use by or disclosure to their treating doctors. Alternatively, in the situation described by Dr Suthers directly above, the clinic has the option of later seeking consent to a particular disclosure.

22.54 Dr Finlay Macrae, Head of Colorectal Medicine and Genetics at the Royal Melbourne Hospital, raised related issues concerning the application of privacy legislation to the disclosure of family genetic information in the course of genetic counselling. He stated that genetic counselling is impeded by the need to ensure that information on an accumulating pedigree is not shared with the family member being interviewed, in case the already collected information includes information ‘provided confidentially by others in the family’.⁶⁸ He noted that while, in theory, consent might be obtained for the disclosure of family genetic information to other family members

there may still be information about other [live family] members provided indirectly for the pedigree, and the status of consent for this disclosure is never clear, and potentially could lead to privacy difficulties.⁶⁹

22.55 Dr Macrae observed that:

Working pedigrees rarely include, on the pedigree, the source of the information (except if death certified or cancer register retrieved), so the usual practice is not to disclose anything other than the immediate family to the person being interviewed. The full impact of the pedigree pattern of inheritance is not then pictorially available to the family member being interviewed, and this deprives the counsellor of one of his/her most cogent educational tools to promote surveillance in the family. Displaying an anonymous pedigree is possible, but leads to confusion for both the counsellor and the family member (potentially) as names act as anchors to understanding the pedigree to the naive witness.⁷⁰

22.56 He suggested that this constraint on the disclosure of genetic information hampers effective counselling and causes more ‘overall harm than good’.⁷¹

68 F Macrae, *Submission G069*, 14 January 2002.

69 Ibid.

70 Ibid.

71 Ibid. Dr Macrae also noted that while the pedigree can be discussed with the patient (and verified against the patient’s own understanding) without the patient directly viewing the document, this ‘leads to an awkward interview where the counsellor is seen to be a controller of family information which “does not belong to [the counsellor]”’.

22.57 For a variety of reasons, pedigrees contain information which may include personal information about relatives who have not consented to having their information recorded. These relatives may have died, not been approached for consent, or have denied consent to have the information collected or disclosed. The Cancer Council Victoria Cancer Genetics Advisory Committee⁷² highlighted this concern and, in particular, the fact that current state legislation requires consent from a deceased's legal representative,⁷³ who is not always easy to identify or locate.

22.58 Some genetics services have developed detailed guidelines on how such information should be dealt with. The South Australian Clinical Genetics Service Familial Cancer Registry has developed *Confidentiality and Consent Guidelines* which, among other things, provide guidelines on the recording and release of pedigree information. These distinguish between situations where the relative (to whom the information relates): has died; is alive and consent has yet to be sought; is alive and consent has been sought but no response has yet been received; and where consent has been denied.⁷⁴ For example, the guidelines state that:

- Pending consent of a relative, identifying data may be recorded but not released to another party outside the Familial Cancer Registry and the Familial Cancer Unit; identifying data is not released to other services (eg laboratory services) of the South Australian Familial Cancer Service. Pending consent, confirmation of diagnoses may not be sought from other sources.
- Where consent has been denied, the following non-identifying data may be recorded on the database: gender, cancer diagnosis, and age at diagnosis. These data are not released to another party outside the Familial Cancer Registry and the Familial Cancer Unit.⁷⁵

Inquiry's views

22.59 The collection of family medical history information from registrants is essential to the operation of genetic registers. The Inquiry has concluded that privacy legislation may inappropriately constrain established practices for the collection of family medical history from registrants and that this should be remedied.⁷⁶

72 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

73 *Health Records Act 2001* (Vic) ss 3, 95(2).

74 G Suthers, *Submission G026*, 30 November 2001.

75 *Ibid.* The SA guidelines note that where consent is denied identifying details (ie name, date of birth, and address) must be deleted from the person's record in that family. However, 'a record of these people must be kept so that we know not to bother them again'.

76 In the UK, the Human Genetics Commission has recommended that further legislation is required to ensure that the holders of information about genetic relatives in a clinical context are specifically exempted from obligations in relation to the processing (obtaining and recording) of information under the *Data Protection Act 1998* (UK): Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 70–71.

22.60 A more difficult question is whether verification of such information should occur only with the consent of the genetic relative or next of kin (where the genetic relative is deceased). For example, as noted above, the NHMRC Guidelines for Genetic Registers state that confirmation of diagnosis may be obtained without consent where approaching the individual would be ‘intrusive or potentially upsetting’. This guidance is not consistent with the requirements of the *Privacy Act*.⁷⁷

22.61 The most obvious reform option at federal level would be the making of a new PID by the federal Privacy Commissioner to permit the operators of genetic registers to collect certain family medical information in ways that might otherwise breach the NPPs of the *Privacy Act*. A PID dealing with the operation of genetic registers may need to deal not only with the collection of genetic information, but also aspects of its use, disclosure or de-identification.

22.62 There was support in submissions for the development of a PID relating to the operation of genetic registers, at least to the extent that the *Privacy Act* applies to existing genetic registers.⁷⁸ However, submissions also expressed concern that the scope and content of such a PID be appropriately circumscribed.⁷⁹ The Cancer Council Victoria Cancer Genetics Advisory Committee stated:

[W]e would advocate that organisations applying for such a PID should first establish their credentials with the proposed [Human Genetics Commission of Australia]. The proposed HGCA would take into account the reasons why the organisation wishes to verify diagnoses (eg gene specific disease registers), the penetrance and seriousness (morbidity and/or mortality of the disease) and its treatability. ... the proposed HGCA would weigh the importance of such access to the organisation, alongside the potential benefit to the family and perhaps the community, on a disease by disease basis.⁸⁰

22.63 The Commonwealth Department of Health and Ageing suggested that

the privacy implications of extending a public interest determination to cover information on a genetic register (and prevent any breaching of the Privacy Act) should be further considered, provided it does not compromise the right not to know.⁸¹

77 *Privacy Act 1988* (Cth) NPP 1, NPP 10.

78 Law Institute of Victoria, *Submission G275*, 19 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003; Familial Cancer Service — Westmead Hospital, *Submission G295*, 6 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Office of the Health Services Commissioner Victoria, *Submission G307*, 17 January 2003.

79 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; I Robertson, *Submission G209*, 29 November 2002; Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

80 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

81 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

22.64 The Androgen Insensitivity Syndrome Support Group Australia submitted that genetic registers

should be exempt from privacy principles only where they relate to medical conditions that pose a serious risk to the health or life of genetic relative ... or only where specific permission has been given for the information to be obtained or disclosed.⁸²

22.65 The Inquiry recommends that organisations operating genetic registers should seek a PID under the *Privacy Act*. The primary intention of this recommendation is to ensure that, in appropriate circumstances, organisations can continue to collect and retain family medical history information from the registrants on genetic registers without the consent of the genetic relatives.

22.66 On the basis of the submissions received, the Inquiry has not been able to form a view on whether the PID should also permit genetic registers to collect health information from health professionals without the consent of their patients, for the purpose of verifying information provided by registrants. However, this issue should be examined as part of the PID process.

22.67 There may also be aspects of the use or disclosure of this information that require review in the context of a PID process.⁸³ For example, one issue that might be dealt with is disclosure of pedigrees by medical practitioners to genetic registers, in order to assist in identifying individuals at risk. Another is that disclosing information about genetic relatives on a genetic register, even in 'de-identified' form, could breach NPP 2 of the *Privacy Act*. As noted above, information on a genetic register may not be truly de-identified, even where identifiers, such as name and date of birth, have been removed.

22.68 In practice, the *Privacy Act* has limited coverage of genetic registers in Australia. The Inquiry understands that, with few exceptions, genetic registers are generally public hospital based. These registers will be governed by state or territory privacy legislation, where it exists. As noted above, and in accordance with Recommendations 7-1 and 7-2, state and territory legislatures and privacy regulators should consider amending their privacy regulation in accordance with Recommendation 22-1. The form and mechanism of such amendment will differ depending on the jurisdiction.⁸⁴

82 Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003.

83 Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002.

84 For example, in Victoria, the Health Privacy Principle applying to the collection of health information may be extended by regulation. See 'collection in the prescribed circumstances': *Health Records Act 2001* (Vic) Health Privacy Principle 1.1(i). In New South Wales the Privacy Commissioner may make written directions exempting organisations from complying with the Health Privacy Principles: *Health Records and Information Privacy Act 2002* (NSW) s 62.

Recommendation 22–1. Organisations operating genetic registers should seek a Public Interest Determination (PID) under the *Privacy Act 1988* (Cth), where applicable, to ensure that they can continue to collect family medical history information from registrants without breaching the National Privacy Principles. The PID process should review whether any other acts or practices of genetic registers, including those involving the use or disclosure of personal information, may justify exemption under the PID.

Recommendation 22–2. The National Health and Medical Research Council (NHMRC) should review the *Guidelines for Genetic Registers and Associated Use of Genetic Material* with particular regard to the de-identification of information. In conducting its review, the NHMRC should ensure that the Guidelines are consistent with privacy laws.

23. Genetic Counselling and Medical Education

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Introduction

Somebody told her she's not going to live forever and she's been preparing to die ever since.¹

23.1 Genetic test information may have profound medical and psychological implications for individuals. These implications will depend on the nature and context of genetic testing—the genetic condition being tested for and the reasons for testing. The results and implications of genetic testing will often be complex and difficult to understand. Some of the factors that create this complexity and uncertainty include the interaction between genes, the interplay between genetic and environmental causes of disease, different patterns of inheritance of genes, the degree of penetrance associated with particular genetic mutations, and the varying accuracy of genetic test results.² For these reasons, and to respond to the ethical implications of tests and results, it is important that individuals are provided with appropriate information about a genetic test and, in some cases, assisted in decision making through genetic counselling.

23.2 The provision of information about a genetic test and genetic counselling are not synonymous. The giving of information is primarily an educational process that utilises printed and audiovisual resources or explanation by a health professional, or both. This can be contrasted with genetic counselling, which encompasses both

1 From the screenplay of A Niccol, *GATTACA* (1997), Columbia Pictures.

2 For further information about these aspects of genetics and genetic testing, see Ch 2–3.

information giving and the discussion of the implications for the individual in a contextual framework that is unique to each person.³

23.3 This chapter discusses genetic counselling, the need for genetic counselling services, and issues related to its further development as a professional discipline. The chapter then examines the education and training of medical practitioners in clinical genetics, genetic counselling and related ethical issues.

Relevance to the Inquiry

23.4 The provision of appropriate information about genetic testing by medical practitioners to their patients and the provision of genetic counselling by appropriately trained health professionals may have an important, albeit indirect, role to play in protecting people from inappropriate discriminatory use of genetic information and in promoting privacy protection and ethical treatment and conduct.

23.5 For example, inappropriate discriminatory use of genetic information may arise where an employer seeks to rely on genetic information in making its employment decisions without fully comprehending the health implications of the information. Such misconceptions may be corrected by the provision of more complete or nuanced information by a medical practitioner or genetic counsellor. Similarly, inappropriate disclosure (or non-disclosure) of genetic information to an individual's relatives may be minimised where patients are properly advised about the implications for others of their test results.

23.6 Submissions and consultations have highlighted concerns about access to genetic counselling and about the inadequate knowledge of medical practitioners about genetics and related ethical issues.

Genetic counselling

23.7 The Human Genetic Society of Australasia (HGSA) defines genetic counselling as a communication process that deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family.

The process involves an attempt by one or more appropriately trained persons to help the individual or family (1) comprehend the medical facts including the diagnosis, the probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.⁴

3 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, NHMRC, <www.nhmrc.gov.au/issues/humangenetics.htm>, 19 February 2003, 30.

4 Human Genetics Society of Australasia, *Guidelines for Human Genetics Society of Australasia (HGSA) Training and Certification in Genetic Counselling*, <www.hgsa.com.au/policy/GTrainGenCoun.html>, 20 February 2003.

23.8 At present counselling is generally performed within a clinical genetics centre or a specialist service such as a familial cancer service. To ensure the effectiveness of genetic counselling, a multi-disciplinary team of professionals often work together. These teams may include clinical geneticists, other medical specialists, genetic counsellors, social workers and other health workers, as appropriate.

Genetic counsellors

23.9 In Australia, genetic counsellors are tertiary trained health professionals with specialist training in genetics and counselling. They are certified by the HGSA to provide genetic counselling in conjunction with a clinical geneticist (a medical practitioner who has undertaken specialist training in clinical genetics). Genetic counselling itself is a relatively new discipline in Australia—the first genetic counsellor received certification from the HGSA in 1991.

23.10 To commence training as a genetic counsellor requires tertiary qualifications, generally in a related discipline such as nursing, science, social work or education. To become an HGSA certified genetic counsellor requires accredited qualifications in both genetics and counselling (Part I) and two years full time equivalent supervised practice (Part II). The HGSA recommends that persons who have completed Part I of the training, and are working towards attaining Part II, should use the title Associate Genetic Counsellor. It also recommends that the title Genetic Counsellor should be used only after attainment of Part II of the certification. The HGSA Board of Censors for Genetic Counselling has established a post-certification program for the maintenance of professional standards for certified genetic counsellors.⁵

23.11 A growing number of persons have attained Part I of the HGSA training and are working in areas such as research and education. The Australasian Society of Genetic Counsellors, a special interest group of the HGSA, is discussing the use of the title of Genetic Associate for such persons.

Anticipating future need for genetic counselling

23.12 In a submission to the Inquiry, Dr Jennifer Kromberg of the Queensland Clinical Genetics Service remarked:

Genetic tests include prenatal tests, neonatal tests (eg newborn screening), predictive and diagnostic tests. A large proportion of the population will require one or more of these tests sometime during their life. Physicians cannot meet the demand. Therefore if consent to testing is to be adequately informed properly trained Genetic Counsellors who are registered in a regulated profession must be available.⁶

23.13 As genetic medicine and testing technology develop, there will be an increasing need for genetic counselling and related services. This demand has been recognised by similar inquiries overseas. In Canada, the November 2001 report of the Ontario Provincial Advisory Committee on New Predictive Genetic Technologies noted that:

5 New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002.

6 J Kromberg, *Submission G060*, 11 January 2002.

Since genetic testing often produces complex results, testing must be part of broader integrated multidisciplinary genetic services that incorporate genetic assessment and counselling, quality testing, psychosocial support and follow-up services, including surveillance, prevention and treatment. Every effort should be made to integrate genetic services into current health care.⁷

23.14 The Committee referred to the potential growth in the number and volume of genetic tests and their accompanying costs. The Committee made recommendations to increase the recruitment and training capacity for genetic service health care providers.⁸

23.15 Resourcing of all aspects of genetic services in the United Kingdom is currently being examined by the Department of Health, as it is expected that the increasing demand for genetic counselling will not be met by the current or projected resources.⁹

23.16 In Australia, submissions and consultations to the Inquiry highlighted concerns about increasing demand for genetic counselling.¹⁰ Submissions confirmed that steps need to be taken now to plan for this increased need.¹¹ For example, the Centre for Genetics Education submitted that:

There is wide discrepancy in access to services throughout Australia that requires urgent addressing. It is also essential that the services are resourced appropriately, enabling genetics education of the community and professionals and access to and support for continuing education and training.¹²

23.17 Similarly, the HGSA submitted that a national approach to clinical genetics services is essential to ensure equity of access, development of outreach services and the provision of culturally appropriate genetic counselling for the Indigenous community.¹³

7 The Provincial Advisory Committee on New Predictive Genetic Technologies, *Genetic Services in Ontario: Mapping the Future* (2001), Ministry of Health Ontario, Toronto, 7.

8 *Ibid.*, 7, 56–60.

9 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 52.

10 *Confidential Submission G051CON*, 14 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Thyroid Australia Ltd, *Submission G020*, 28 November 2001; Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002; *Confidential Submission G030CON*, 17 December 2001; Queensland Government, *Submission G274*, 18 December 2002. See also S Metcalfe and others, 'Needs Assessment Study of Genetics Education for General Practitioners in Australia' (2002) 4 *Genetic Medicine* 71, 71–77.

11 Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Queensland Government, *Submission G274*, 18 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

12 Centre for Genetics Education, *Submission G232*, 18 December 2002. The Genetic Support Network of Victoria highlighted the need for more counselling services in regional and rural areas: Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

13 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

However, it will be important to take a broad view in addressing the [need] for more genetic counselling services. The need should be addressed by improving resources for genetic education as well as increasing the number of substantive and training positions for both genetic counsellors and clinical geneticists.¹⁴

23.18 The funding of genetic counselling services is a closely related issue, which requires consideration. At present, genetic counselling is mainly provided within the state and territory public health system, for example, by genetic counsellors working in public hospital based clinical genetics services. The Medicare Benefits Schedule (MBS) does not cover genetic counselling by HGSA certified genetic counsellors. Some genetic counselling provided by medical practitioners may be covered by consultation fees. The Queensland Government submitted that the present lack of MBS coverage for genetic counselling may need review.¹⁵ It has also been suggested that any problems in relation to access to counselling are now more likely to arise from a shortage of paid positions for counsellors in the public health system than from a shortage in the number of people training as counsellors.¹⁶

23.19 The Inquiry has concluded that, as genetic medicine and testing technology develop, there will be an inevitable increase in the need for genetic counselling services in Australia. Strategies should be developed now to assess and respond to this increased need.

23.20 The Commonwealth Department of Health and Ageing is aware of the rapid growth in the field of genetic testing and the potential need for many genetic tests to be accompanied by counselling.¹⁷ The Department is currently working with States and Territories through the Australian Health Ministers' Advisory Council (AHMAC) on a number of issues relating to the provision of presymptomatic and diagnostic genetic testing.¹⁸ In February 2002, the National Public Health Partnership, a sub-committee of AHMAC, established a Public Health Genetics Working Group. In May 2002 an AHMAC Advisory Group on Human Gene Patents and Genetic Testing was established. The AHMAC Advisory Group will advise and make recommendations to AHMAC on matters relating to the planning, management, regulation, provision and delivery of human genetic testing and screening services, for the purposes of the diagnosis, prevention and treatment of human disease and the improvement of human health.¹⁹

14 Ibid.

15 Queensland Government, *Submission G274*, 18 December 2002.

16 Children's Hospital at Westmead, *Consultation*, Sydney, 19 November 2002.

17 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

18 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

19 In view of the establishment of the AHMAC Advisory Group, the National Public Health Partnership agreed that the Public Health Genetics Working Group would be disbanded and future National Public Health Partnership activity in the area of public health genetics would be undertaken in response to any reference from the AHMAC Advisory Group: National Public Health Partnership, *Public Health Genetics Working Group*, Department of Human Services (Victoria), <www.dhs.vic.gov.au/nphp/genetics/index.htm>, 20 February 2003.

23.21 The Inquiry considers that AHMAC or National Public Health Partnership working groups may be appropriate forums within which national approaches to the provision of genetic counselling services could be pursued.

Recommendation 23–1. As a matter of priority, the Commonwealth, States and Territories should develop strategies to assess and respond to the need for increased and adequately resourced genetic counselling services.

Development of genetic counselling as a profession

23.22 Submissions and consultations indicated strong support for the involvement of Commonwealth, state and territory governments in the further development of genetic counselling as a profession.²⁰

23.23 DP 66 noted that, unlike medical practice, nursing or psychology, genetic counselling is not a registered health profession. There is, therefore, no equivalent of provisions in medical practice acts that make it an offence for persons who are not registered medical practitioners to claim to be, or hold themselves out as, medical practitioners.²¹ There is no prohibition on any person, however qualified, holding themselves out as a genetic counsellor or offering genetic counselling services.²² Nor are there formal sanctions for breach of ethical or professional standards in genetic counselling.²³

23.24 Some submissions suggested that genetic counselling should be a registered health profession.²⁴ Associate Professor John MacMillan of the Queensland Clinical Genetics Service submitted that:

20 Centre for Law and Genetics, *Submission G255*, 21 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Haemophilia Foundation Victoria, *Submission G201*, 25 November 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

21 See eg *Medical Practice Act 1992* (NSW) s 105; *Medical Practice Act 1994* (Vic) s 62.

22 Subject, for example, to the provisions of trade practice legislation, which prohibits misleading or deceptive conduct in trade or commerce: *Trade Practices Act 1974* (Cth) s 52; *Fair Trading Act 1987* (NSW) s 42 and cognate legislation in other States and Territories.

23 However, as some genetic counsellors are registered health professionals, such as nurses or psychologists, unethical conduct as a genetic counsellor may have consequences under the regulatory frameworks applying to those registered health professions.

24 J Kromberg, *Submission G060*, 11 January 2002; J MacMillan, *Submission G015*, 19 November 2001; Centre for Genetics Education, *Submission G232*, 18 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002.

Regulation of genetic testing to the public hospital and public health care systems offers the opportunity to ensure it is appropriately available to all who need it and that full informed consent is obtained by an appropriately trained health professional. I believe that such individuals should be part of a profession whose practice is governed by a licensing process or registration board. This would require that Genetic Counsellors (who are not medically trained) become registered with the Professional Registration Boards in each state under an appropriate system. These professionals would also be required to have professional indemnity insurance. Failure to regulate this 'new profession' could have serious adverse consequences for patients and families through the provision of misleading or incorrect information concerning the results and interpretation of genetic tests.²⁵

23.25 Dr Kromberg stated that the public needs to have access to expert genetic counsellors, but it also needs to be protected against untrained people who may set themselves up as genetic counsellors.

Genetic Counsellors are being trained at post-graduate level to provide Genetic Counselling services in Australia. However they are not yet recognised as a health profession and professional recognition and regulation are essential. Such regulation would contribute to the progression and much needed expansion of the profession, protect the public, and lead to the development of a standardised, high quality and ethical service.²⁶

23.26 The Centre for Genetics Education submitted that:

It is unacceptable that currently any health professional can call themselves a genetic counsellor without any sanction. This is unacceptable given the complexity of genetic information and the potential damaging outcomes if information is incorrect or incorrectly interpreted.²⁷

23.27 The Royal Australian and New Zealand College of Obstetricians and Gynaecologists submitted that:

In clinical practice our Fellows often need to seek the assistance of Genetic Counsellors for the counselling and support of patients in our care. Currently a number of Ultrasound and IVF practices employ nurses, social workers and scientists who have a genetic counselling graduate diploma. It is essential that this profession is recognised as a health profession and a registration system is developed for its supervision.²⁸

23.28 The HGSA also submitted that genetic counsellors should be required to be registered and to be indemnified for their practice in order to 'ensure adequate oversight of appropriate practice and enable genetic counsellors to develop their role'.²⁹ The Association of Genetic Support of Australasia (AGSA) expressed the view

25 J MacMillan, *Submission G015*, 19 November 2001.

26 J Kromberg, *Submission G060*, 11 January 2002.

27 Centre for Genetics Education, *Submission G232*, 18 December 2002. The Centre noted recent recognition of genetic counselling professional training standards by the Psychotherapy and Counselling Federation of Australia and stated that this may assist in the development of genetic counselling as a recognised, and ultimately registered, health profession.

28 Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Submission G281*, 24 December 2002.

29 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

that because counsellors are not registered and have no medical indemnity, a greater (and unnecessary) load is placed on clinical geneticists.³⁰

23.29 In the United Kingdom, the Association of Genetic Nurses and Counsellors is pursuing statutory registration of genetic counsellors through the Health Professions Council.³¹ The Health Professions Council is an independent regulatory body responsible for setting and maintaining standards of professional training, performance and conduct of the 12 health care professions that it regulates. The Council also ensures that registration of professionals is linked to continuing professional development.³²

23.30 The primary purpose of registering health professionals is said to be the protection of the public.³³ The public interest is protected through the powers granted to health registration boards to recognise qualifications and assess the character of persons seeking registration, to discipline health professionals found guilty of unprofessional conduct, and to suspend or place conditions upon those whose capacity to practice is impaired.³⁴

23.31 AGSA stated that registration of genetic counsellors would be desirable in that it would encourage:

- Greater recognition of the profession
- Genetic counsellors to be responsible for their own practice (with the backup of indemnity insurance)
- Development of agreed minimum standards of practice that are universally accepted
- Improvement of professional standards
- Genetics units to be more proactive in training of counsellors, with the view to achieve finite goals put forward by a registering body
- Achieving a certain level of independence for genetic counsellors in practice, without compromising professional standards
- Relief of part of the clinical burden on geneticists, so that they may be used in other capacities, though this is not a replacement for ongoing medical supervision of genetic counsellors

30 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

31 J Kromberg, *Submission G060*, 11 January 2002. See also Association of Genetic Nurses and Counsellors, *Registration*, <www.agnc.co.uk/registration.htm>, 20 February 2003.

32 Health Professions Council, *About Us*, <www.hpcuk.org/docs/about_us.htm>, 2 July 2002. The HPC is the successor body to the Council for Professions Supplementary to Medicine, established under the *Professions Supplementary to Medicine Act 1960* (UK).

33 See K Breen, V Plueckhahn and S Cordner, *Ethics, Law and Medical Practice* (1997) Allen & Unwin, Sydney, 77.

34 *Ibid.* As at May 2001, all Australian jurisdictions regulated doctors, nurses, dentists, optometrists, physiotherapists, pharmacists, chiropractors and psychologists. However, only some jurisdictions register other health professions such as dental technicians, osteopaths, occupational therapists, radiographers and Aboriginal health workers: R Deighton-Smith, B Harris and K Pearson, *Reforming the Regulation of the Professions National Competition Council Staff Discussion Paper* (2001) National Competition Council, 4.

- Greater international recognition and acceptance of locally trained and registered genetic counsellors, which would encourage an exchange of expertise in the field, and benefit the profession in general.³⁵

23.32 Regulation of health professions has important competition policy implications. The National Competition Council has noted that overly restrictive or anti-competitive regulation can impose major and unnecessary costs on consumers and may contribute to the severe shortages of many health professionals, particularly in rural and regional Australia.³⁶

23.33 The Queensland Government noted that the integration of genetic counselling into established health professions should not be overlooked in a drive to establish genetic counselling as a recognised health profession. It stated that ‘registration is only one of a number of potential options to ensure appropriate standards are in place to protect the public’.³⁷

23.34 The Inquiry has reached no concluded view on whether a statutory registration scheme for genetic counsellors is desirable. A comprehensive assessment of the advantages and disadvantages of a registration scheme for genetic counsellors would be a major task—and one unsuited to the context of the present Inquiry.

23.35 However, in the Inquiry’s view, Commonwealth, state and territory governments need to examine closely all options for the further development of genetic counselling as a health profession. This examination should focus on measures to recognise the importance of genetic counsellors in building a bridge between emerging genetic science and the psychosocial needs of individuals who are, or may come to be, affected by genetic disorders. In this context, the development of clear standards of professional ethics would be desirable. There is also a need to protect the public from substandard genetic counselling services and, in particular, from persons who are not appropriately trained, qualified and supervised, holding themselves out to be genetic counsellors.

Recommendation 23–2. The Commonwealth, States and Territories should examine options for the further development of genetic counselling as a recognised health profession, including the use of certification, accreditation or registration systems for genetic counsellors.

35 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

36 National Competition Council, *Reform of the Health Care Professions*, Community Information Brochure, 2000.

37 Queensland Government, *Submission G274*, 18 December 2002.

Delivery of genetic counselling services

23.36 The need for genetic counselling will vary depending on the particular genetic test involved and the context of testing. Genetic counselling may be required both before and after predictive or presymptomatic tests,³⁸ following a positive genetic carrier test, and following an abnormal result on a prenatal diagnostic or screening test.³⁹ However, where the testing is purely diagnostic, the provision of appropriate information prior to testing may be sufficient.⁴⁰ If the result is positive, discussion of implications for relatives is essential. Some predictive tests may need skilled multi-disciplinary teams to deal with different aspects of the patient's medical and social care.⁴¹ For example, internationally accepted protocols for presymptomatic testing for Huntington's disease include provision of psychosocial support.

23.37 Madelyn Peterson and Dr Jennifer Kromberg noted that, while Queensland Health currently funds the provision of psychosocial support in Huntington's disease testing, funding is not available to support individuals referred for predictive tests for other adult-onset neurodegenerative diseases or familial cancers, which raise similar psychosocial issues. They noted, for example, that genetic testing for familial cancer genes 'generates potentially life-altering information about self, whether the result is positive or negative'. Such genetic tests will potentially necessitate decisions about prophylactic surgery, chemo-prevention trials, dietary and other health modifications, additional medical surveillance and insurance or privacy issues.⁴²

23.38 Other genetic tests may have fewer or less complex implications for the individuals tested and their families. Some predictive testing, where the penetrance of the gene is low, may be considered analogous to other tests used in clinical practice, such as the measurement of blood pressure or serum cholesterol,⁴³ and may not necessitate counselling.

23.39 The United Kingdom's Human Genetics Commission (HGC) has provided a useful comparison of tests of low and high 'informational impact' or significance for the individuals being tested—who therefore will have different needs in relation to

38 As discussed in Ch 4, predictive testing is also called presymptomatic testing where an individual's family medical history suggests that he or she may have the genetic disorder but symptoms of it are not yet manifest.

39 HGSA guidelines state the diverse reasons why a person may seek genetic counselling: Human Genetics Society of Australasia, *Guidelines for Human Genetics Society of Australasia (HGSA) Training and Certification in Genetic Counselling*, <www.hgsa.com.au/policy/GTrainGenCoun.html>, 20 February 2003, 1.

40 See National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: An Information Paper*, NHMRC, <www.nhmrc.gov.au/issues/humangenetics.htm>, 19 February 2003, 30.

41 A multidisciplinary team is the ideal for a comprehensive genetics service: J Kromberg, *Submission G060*, 11 January 2002.

42 M Peterson and J Kromberg, *Submission G108*, 19 February 2002.

43 R Zimmer and C Cook, *The Nuffield Trust Genetics Scenario Project: Genetics and Health: Policy Issues for Genetic Science and their Implications for Health and Health Services* (2000), The Stationery Office, London, 53.

counselling.⁴⁴ The HGC has stated that there are a large number of tests that are of low informational impact:

This includes 'genetic' tests which result in limited information about inheritance (for example ABO blood grouping tests) or those which give information about acquired 'somatic' mutations (for example tumour genotyping in a pathology laboratory) which does not necessarily have any impact for genetic relatives. However, even here judgement is required, as ABO testing could readily reveal non-paternity and tumour genotyping may have important implications for treatment options and prognosis.

At the other end of the spectrum there are a number of genetic tests that might be considered to have high significance (or informational impact). There are genetic tests for conditions that potentially have a major impact on the health of the patient and possibly for the patient's blood relatives. They also have wider implications in that the information that they disclose could affect the patient's financial and employment prospects as well as affecting their social relationships. Examples of tests in this category include tests for late-onset genetic disorders or for fetal abnormality ...⁴⁵

Access to medical genetic testing and counselling

23.40 While genetic testing for medical purposes usually requires a referral from a medical practitioner,⁴⁶ there are currently no uniform protocols regarding referral.⁴⁷ At least where there is no centralised clinical genetics service, protocols for dealing with referrals may vary between laboratories. Some may place more emphasis than others on ensuring that testing is linked with access to genetic counselling, or provide more interpretative information about genetic test results than others. In this situation, the laboratory may have to act as a 'gatekeeper', assessing whether the requesting medical practitioner has understood the patient's need for information or counselling prior to testing. If not, the laboratory may have to discuss the matter with the practitioner or even decline to provide testing.

23.41 The absence of clear protocols on access to genetic testing services through health professionals may leave open the possibility of genetic testing in inappropriate circumstances. For example, the Australian Huntington's Disease Association (NSW) expressed concern that some people at risk for Huntington's disease

undergo presymptomatic testing without accessing a recognised genetics service and therefore can have the predictive test for HD without the comprehensive support and expertise of a genetic counsellor and geneticist.⁴⁸

44 Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (2002), London, 51.

45 Ibid, 51.

46 National Pathology Accreditation Advisory Council, *Standards for Pathology Laboratories* (1998), Department of Health and Family Services, Canberra, Standard 5—Specimens.

47 However, the Inquiry understands that, in general, laboratories do not accept requests from general practitioners for sensitive types of genetic tests without referral to a genetic counsellor and clinical geneticist. Further, the HGSA has produced a range of policies and guidance notes relevant to requests for genetic testing: Human Genetics Society of Australasia, *HGSA Policies*, <www.hgsa.com.au>, 20 February 2003.

48 Australian Huntington's Disease Association (NSW), *Submission G054*, 14 January 2002.

23.42 A related concern is that medical practitioners may not make appropriate referrals for genetic counselling.⁴⁹ One genetic counsellor suggested that systems for referral to genetic counselling may need to be developed so that requests for certain types of genetic test automatically generate a referral.⁵⁰

23.43 Although some submissions suggested that the ability to order genetic tests for medical purposes should be limited to certain categories of medical practitioner,⁵¹ this approach was generally rejected. For example, while the HGSA was critical of the current level of genetics understanding in the medical profession, it stopped short of recommending restrictions on the ability of medical practitioners to order genetic tests.

At present, most health professionals do not have the expertise to interpret the results of many genetic tests, whether done for diagnosis, carrier detection or predictive testing. This situation is not unique to genetic tests and applies in other specialised areas of medicine. It would be inappropriate to restrict the ability of health professionals to order diagnostic, carrier or prenatal genetic tests.⁵²

23.44 In relation to the delivery of genetic counselling by medical practitioners, the Department of Health and Ageing stated:

While primary care providers such as General Practitioners can provide a basic level of information and advice about genetic testing, there will be more complex cases in which it may be more appropriate to refer patients to genetic counsellors who can provide a more specialised service. The Department recognises the risks relating to the provision of incorrect information to patients by unqualified counsellors. However, it considers that it is important to ensure that primary health care and other medical professionals continue to be able to provide basic level counselling without the requirement to be formally registered as genetic counsellors.⁵³

23.45 The Department also noted that the AHMAC Advisory Group on Human Gene Patents and Genetic Testing is considering guidelines on the appropriate delivery of counselling services and the qualifications of those service providers and will consult with the HGSA and other stakeholders.⁵⁴

Classification of genetic tests

23.46 One option that could provide an enhanced level of oversight for ordering sensitive tests and ensure better access to genetic counselling is a scheme for classifying genetic tests, or categories of genetic tests. For example, it might be considered appropriate to develop protocols so that predictive testing of late onset

49 Association of Genetic Support Australasia Inc, *Consultation*, Sydney, 11 February 2002; Genetic Support Network of Victoria, *Consultation*, Melbourne, 18 March 2002.

50 Genetic Counsellor, *Consultation*, Townsville, 3 April 2002.

51 For example, the Neurofibromatosis Association of Australia submitted that 'In our opinion medical practitioners should be required to undergo specific training before being able to order genetic testing or interpret the results for patients. In the ideal situation, only medically trained clinical geneticists should be authorised to order such tests and interpret their results': Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002.

52 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

53 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

54 Ibid.

neurological disorders (such as Huntington's disease) can only be ordered by, or in collaboration with, a clinical genetics service.

23.47 The development of a classification scheme has been examined in the United States by the Secretary's Advisory Committee on Genetic Testing (SACGT).⁵⁵ SACGT developed a classification proposal that was subjected to pilot testing and public consultation. After considerable internal and public review, SACGT concluded that 'fundamental, irresolvable questions' had been raised about 'the feasibility of categorising tests for oversight purposes based on a limited set of elements in a simple, linear fashion'.⁵⁶

23.48 The Queensland Clinical Genetics Service highlighted how the evaluation of genetic tests is complex and time intensive—and needs to involve assessment of the ethical, legal and social implications of a test, as well as its scientific validity.⁵⁷

23.49 An example of an evaluation methodology is that used in the United States in a project carried out by the Foundation for Blood Research,⁵⁸ which evaluated emerging genetic testing by looking at analytical validity, clinical validity and clinical utility, as well as ethical, legal and social implications. Applying such models can be contrasted with the generally *ad hoc* process by which genetic tests may be introduced in the Australian health care system.⁵⁹ Systematic evaluation is carried out only if a test is proposed for listing on the MBS, in which case the safety, effectiveness and cost effectiveness of testing will be assessed by the Medical Services Advisory Committee.⁶⁰

23.50 Despite the possible practical difficulties, in DP 66 the Inquiry proposed further development of genetic testing and counselling practice guidelines by the Human Genetics Commission of Australia (HGCA), in consultation with the HGSA, state clinical genetics services, and other interested organisations.⁶¹ DP 66 noted that such guidelines could indicate, for example, that certain types of genetic test may be ordered only where genetic counselling has been provided, or by medical practitioners with specific qualifications, or both. Guidelines could also emphasise the importance

55 See Secretary's Advisory Committee on Genetic Testing, *Enhancing the Oversight of Genetic Tests: Recommendations of the SACGT* (2000), National Institutes of Health, Baltimore; Secretary's Advisory Committee on Genetic Testing, *Development of a Classification Methodology for Genetic Tests: Conclusions and Recommendations of the Secretary's Advisory Committee on Genetic Testing* (2001), National Institutes of Health, Bethesda.

56 Secretary's Advisory Committee on Genetic Testing, *Development of a Classification Methodology for Genetic Tests: Conclusions and Recommendations of the Secretary's Advisory Committee on Genetic Testing* (2001), National Institutes of Health, Bethesda.

57 Queensland Clinical Genetics Service, *Consultation*, Brisbane, 11 November 2002.

58 J Haddow and others, *Genomics and Disease Prevention: Public Health Perspectives*, Centers for Disease Control and Prevention, <www.cdc.gov/genomics/info/perspectives/files/testACCE.htm>, 20 February 2003.

59 Queensland Clinical Genetics Service, *Consultation*, Brisbane, 11 November 2002.

60 See Medicare Services Advisory Committee, *Genetic Test for Fragile X Syndrome: Assessment Report*, Department of Health and Ageing, <www.health.gov.au/msac/pdfs/msac1035.pdf>, 20 February 2003.

61 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 20–3.

of using skilled multidisciplinary teams in the management of some genetic conditions and the need to identify when psychosocial support is required.

23.51 This proposal received broad support in submissions.⁶² The Genetic Support Council Western Australia emphasised the need for

appropriate involvement of community/consumer representation to ensure genetic counselling is responsive to the needs of genetic support groups and individuals with genetic conditions or genetic predispositions.⁶³

23.52 The Association of Genetic Support of Australasia (AGSA) emphasised that, in order to perform this role, the HGCA would need to include ‘geneticists, genetic counsellors, genetics support group representatives and molecular scientists’.⁶⁴ The HGSA confirmed that it would be pleased to be involved with the proposed HGCA in developing genetic testing guidelines in accordance with the recommendation below.⁶⁵

Recommendation 23–3. The Human Genetics Commission of Australia (HGCA) should develop genetic testing and counselling practice guidelines, in consultation with the Human Genetics Society of Australasia, state clinical genetics services, and other interested organisations. These guidelines should identify genetic tests, or categories of genetic tests, that require special treatment in relation to procedures for ordering testing and ensuring access to genetic counselling. (See also Recommendation 5–3.)

Genetics education and training

23.53 The appropriateness of decisions to order genetic testing or counselling ultimately depends on the training and understanding of the referring practitioner about genetics. Submissions and consultations highlighted concerns that medical practitioners, including some specialists, are not sufficiently knowledgeable about

62 Centre for Genetics Education, *Submission G232*, 18 December 2002; Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002; Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Australian Huntington’s Disease Association (NSW), *Submission G054*, 14 January 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002.

63 Genetic Support Council WA, *Submission G243*, 19 December 2002; The Genetic Support Network of Victoria also emphasised the need to involve genetic support groups in the development of HGCA guidelines: Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

64 Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002.

65 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

ordering genetic tests, interpreting the results of genetic tests, and providing or facilitating the provision of genetic counselling for patients.⁶⁶

23.54 The Commonwealth Department of Health and Ageing stated that there is a need to better educate health professionals about the ethical and scientific principles involved in genetic testing and information.⁶⁷ It supported proposals to enhance graduate and postgraduate training in clinical genetics, genetic counselling and related ethical issues and continuing professional development programs for health professionals and genetic health service providers.⁶⁸

23.55 The Medical Practitioners Board of Victoria submitted that there is clearly a need to better educate health professionals about the ethical principles involved in genetic testing and information, given the rapid developments in this area. The Board noted that many doctors, especially older practitioners, are unlikely to be sufficiently informed in this area.⁶⁹

23.56 The New South Wales Genetics Service Advisory Committee noted that, without additional training, some medical practitioners may have difficulties in interpreting the results of some complex predictive genetic tests and in providing appropriate counselling and support.⁷⁰ Similarly, the HGSA submitted that there is insufficient understanding in the medical profession, outside specialised genetics services, of the wider implications of genetic testing.⁷¹

There is a need to improve health professionals' understanding of the ethical principles involved in predictive testing of currently healthy individuals for later onset disorders, and of the practical consequences that flow from those principles in terms of information giving, counselling and support.⁷²

23.57 Submissions from genetic support groups also raised questions about the genetics knowledge of medical practitioners in specific contexts.⁷³ Thyroid Australia Ltd submitted that:

66 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002; New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002; *Confidential Submission G051CON*, 14 January 2002; Human Genetics Society of Australasia, *Submission G050*, 14 January 2002; Thyroid Australia Ltd, *Submission G020*, 28 November 2001; Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002; *Confidential Submission G030CON*, 17 December 2001; F Richards, *Submission G044*, 14 January 2002; Medical Practitioners Board of Victoria, *Submission G155*, 10 April 2002; N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

67 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002. See S Metcalfe and others, 'Needs Assessment Study of Genetics Education for General Practitioners in Australia' (2002) 4 *Genetic Medicine* 71, 71–77.

68 Commonwealth Department of Health and Ageing, *Submission G313*, 6 February 2003.

69 Medical Practitioners Board of Victoria, *Submission G155*, 10 April 2002.

70 New South Wales Genetics Service Advisory Committee, *Submission G094*, 25 January 2002.

71 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

72 *Ibid.*

73 Thyroid Australia Ltd, *Submission G020*, 28 November 2001; Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002; Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002.

After dealing with close to 2,000 thyroid patients nationwide, we find that many doctors (particularly general practitioners) quite commonly do not test thyroid patients (especially those suffering from hypothyroidism) for antibodies, and these patients are therefore blissfully unaware of the genetic implications of their conditions and believe that they can be 'cured'. They are also unaware of the risks other members of their families face in developing autoimmune thyroid conditions or other autoimmune conditions. This, I believe, is a sad state of affairs. We frequently are faced with the dilemma of telling people that their thyroid problems are probably genetic in nature, suggesting to them that they contact their doctors for the necessary antibody tests to verify this. I believe it is not the role of a support group to perform this function, but that it falls to doctors to do so.⁷⁴

23.58 AGSA had similar concerns and submitted that medical practitioners need to increase their genetics knowledge base.⁷⁵ The results of a member survey conducted by the Neurofibromatosis Association of Australia led it to conclude that

there is a need for the better education of health professionals into the full implications of genetic testing and the information generated by it. This needs to extend to education on the impact on the whole family (including extended family), not just the person being tested.⁷⁶

23.59 Submissions also noted that medical practitioners may be insufficiently aware of relevant resources available to patients affected by genetic conditions, including genetic counselling, clinical genetics services and genetic support groups.⁷⁷ One submission from a patient noted that following diagnosis by her general practitioner

there was no time for counselling about the implications [of the genetic condition] or how to discuss this with family members, implications re life insurance etc. There was no mention of the State's Clinical Genetics Service. The GP is very thorough in general and there has been no other cause for complaint at all.⁷⁸

23.60 The submission from AGSA highlighted the important role of genetic support groups and that more medical practitioners need to be aware of this role.

After diagnosis of a genetic condition, we believe it is important that the family is referred to AGSA or a similar genetic support group. We believe we are the 'third arm' of genetic services – which is just as vital. AGSA provides the essential information to families on what to expect – day-to-day, from families coping with the same condition. ... AGSA has received feedback from many people who have never been referred to us, and have found out about our services through their own research. AGSA is often asked "where have you been?" and "why has nobody ever told us about you?"⁷⁹

74 Thyroid Australia Ltd, *Submission G020*, 28 November 2001.

75 Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002.

76 Neurofibromatosis Association of Australia Inc, *Submission G121*, 18 March 2002.

77 *Confidential Submission G030CON*, 17 December 2001; Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002.

78 *Confidential Submission G030CON*, 17 December 2001.

79 Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002.

23.61 Submissions consistently suggested that the main focus of additional genetics education and training for medical practitioners should be on developing the ability to recognise the complex implications of genetic testing and the consequent need for specialised advice and counselling, rather than on skilling-up medical practitioners to provide genetic counselling services themselves.⁸⁰ For example, Genetic Health Services Victoria stated that:

Whilst it is important to increase genetic education at all levels it is not possible to train all doctors to provide genetic counselling. Doctors should be well informed about the availability of genetic services and how to access them and have the ability to explain carrier states, inheritance patterns, the distinction between diagnostic testing and screening programs and the pros/cons of established screening programs. People considering predictive testing should be referred to Genetic Services.⁸¹

Implementing new education and training programs

23.62 In the course of the Inquiry, a broad consensus emerged that the answer to many of the concerns raised by new and future uses of genetic information lies in education. In relation to medical practitioners specifically, the Inquiry received many comments about the need for additional education and training programs relating to clinical genetics and ethics.⁸²

23.63 The issues involved in developing appropriate genetics education and training programs are complex. For example, while some submissions focused on the need for specialised advice and counselling, some genetic conditions can and should be handled by general practitioners. It would be counterproductive to duplicate the present medical establishment by expecting genetic counsellors to deal with all genetic disorders if, as seems likely, a growing number of common conditions come to be identified as having a genetic component. Genetic counsellors cannot be expected to have detailed knowledge of the clinical implications of many genetic conditions; nor can medical practitioners be expected to provide full counselling services. Most genetic counsellors deal with a full range of genetic disorders and there may be a risk of inappropriate counselling because of a lack of knowledge about the clinical condition.⁸³

23.64 Among the approaches to education and training that should be examined are programs to assist medical practitioners in identifying the genetic conditions that should be referred to specialist genetics clinics, and programs by which medical practitioners may enhance their knowledge and skills in genetics and genetic counselling.⁸⁴ The Centre for Genetics Education submitted that it is important to avoid

80 F Richards, *Submission G044*, 14 January 2002; Association of Genetic Support of Australasia, *Submission G135*, 19 March 2002; Medical Practitioners Board of Victoria, *Submission G155*, 10 April 2002; Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

81 Genetic Health Services Victoria, *Submission G211*, 28 November 2002.

82 The need to address genetics education has been recognised in inquiries overseas. See The Provincial Advisory Committee on New Predictive Genetic Technologies, *Genetic Services in Ontario: Mapping the Future* (2001), Ministry of Health Ontario, Toronto, 7.

83 R Trent, *Correspondence*, 20 July 2002.

84 *Ibid.*

‘compartmentalisation of genetics in clinical terms’ and that all practitioners need to be educated to apply genetics to their field of speciality.⁸⁵

23.65 The Department of Health and Ageing submitted that, to address general practitioner education and training needs nationally

it would be necessary to further develop education and training resources for general practitioners to access voluntarily as part of their quality assurance and continuing professional development requirements. An increased emphasis on genetics education in both the undergraduate medical curriculum and in postgraduate training programs would also be required.⁸⁶

23.66 Professor Nick Saunders and Associate Professor Paul Komesaroff submitted that training of health professionals in ethics, the implications and uses of genetic testing, and genetic counselling, should be an integral part of all undergraduate courses and vocational education programs, and should be an important focus for continuing education programs.⁸⁷

23.67 The HGSA agreed that ‘the problem of an inadequately skilled workforce’ needs to be addressed by undergraduate and postgraduate education. The HGSA also referred to the need for laboratories to provide interpretation of test results in their reports, as well as the need for consultation between those who order a test and health professionals who can interpret the results properly.⁸⁸

23.68 In Australia, the bodies responsible for undergraduate medical education include the Australian Medical Council (AMC) and the Committee of Deans of Australian Medical Schools (CDAMS). The functions of the AMC include review and accreditation of courses conducted by medical schools in Australia and New Zealand leading to basic medical degrees.⁸⁹ Sufficient coverage of genetics could be included as a standard for accreditation.

23.69 From 2002, the AMC will be responsible, with the support of the specialist medical colleges, for the accreditation of programs of postgraduate specialist medical education and training, and continuing professional development programs.⁹⁰ In relation to continuing medical education, the Australian Medical Association submitted that:

85 Centre for Genetics Education, *Submission G232*, 18 December 2002.

86 Commonwealth Department of Health and Ageing, *Submission G150*, 15 April 2002.

87 N Saunders and P Komesaroff, *Submission G084*, 9 January 2002.

88 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

89 The AMC is a national standards body for medical education established by Commonwealth and State Health Ministers. Its other functions include: examination of overseas trained doctors for general (non-specialist) registration; advice to State and Territory Medical Boards on uniform approaches to the registration of medical practitioners; advice to Commonwealth and State Health Ministers on matters relating to the registration of doctors: Australian Medical Council, *Submission G065*, 15 January 2002.

90 Ibid.

responsibility for improving the knowledge of medical practitioners in this rapidly expanding area lies with the Colleges. They should be encouraged to give this area high priority in their educational activities.⁹¹

23.70 The Committee of Presidents of Medical Colleges (CPMC) provides a forum for interaction between the Colleges, AMC and CDAMS, including on issues concerning educational standards and professional training. The CPMC provides advice to government departments and health ministers on request.

23.71 There was substantial support for the Inquiry's proposals with regard to enhancing genetics education and training programs.⁹² Submissions noted the importance of emphasising the ethical, legal and social implications of genetics in the development of education and training programs⁹³ and of providing mechanisms for community and consumer input.⁹⁴

23.72 CDAMS highlighted the need for 'vertical integration' of programs—that is, the integration of undergraduate medical education with postgraduate and continuing training.⁹⁵ CDAMS also emphasised that its preferred approach to the development of the medical curriculum is to avoid, where possible, adding new stand-alone units or subjects to overcrowded curricula and instead to

design over-arching frameworks that articulate the essential knowledge, skills, behaviours and attitudes required of students, and which provide a range of options for their integration into pre-existing curricula structures at each medical school. A curriculum framework should be flexible enough to allow it to be adapted to the particular educational approaches and systems at individual medical schools.⁹⁶

23.73 The Inquiry has concluded that attention to the further development of genetics education is required across all phases of professional medical education. This conclusion is dictated by the increasing importance of genetics in clinical practice. It also emerges from specific concerns expressed to the Inquiry about the genetics

91 Australian Medical Association, *Submission G091*, 29 January 2002.

92 Anglican Diocese of Sydney, *Submission G256*, 20 December 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Royal Australian and New Zealand College of Obstetricians and Gynaecologists, *Submission G281*, 24 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Committee of Deans of Australian Medical Schools, *Submission G200*, 18 November 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Genetic Support Network of Victoria, *Submission G236*, 23 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Androgen Insensitivity Syndrome Support Group Australia, *Submission G290*, 5 January 2003; NSW Health Department, *Submission G303*, 13 January 2003; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002. The Centre for Law and Genetics stated that the proposed HGCA would not be 'constituted with expertise in the area of educational design and development' and should, therefore, be given responsibility for ensuring that other bodies address issues of genetic training and counselling within a specified time period: Centre for Law and Genetics, *Submission G255*, 21 December 2002.

93 Genetic Support Network of Victoria, *Submission G236*, 23 December 2002.

94 Genetic Support Council WA, *Submission G243*, 19 December 2002.

95 Committee of Deans of Australian Medical Schools, *Submission G200*, 18 November 2002.

96 *Ibid.*

knowledge of medical practitioners and the need for additional education and training programs relating to clinical genetics and ethics. These programs need to be addressed, in an integrated way, to those who are in the process of receiving basic medical education in medical schools, to postgraduates, and to general practitioners and specialists, through continuing professional development programs.

23.74 The Inquiry is also of the view that the HGCA should work collaboratively with key professional and educational bodies to design and enhance education and training programs aimed at improving genetic health services provided by medical practitioners as well as other health professionals.

Recommendation 23–4. The HGCA should work with the Australian Medical Council, the Committee of Deans of Australian Medical Schools, and the Committee of Presidents of Medical Colleges to develop an integrated approach to medical education and training in human genetics. This approach should ensure that present and future medical practitioners are appropriately trained and equipped in clinical genetics and in the use of relevant genetic counselling and genetic services.

Recommendation 23–5. The HGCA should work collaboratively with key professional and educational bodies to design and enhance education and training programs aimed at improving genetic health services provided by medical practitioners and other health professionals.

24. Population Genetic Screening

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Introduction

Healthy people attend screening programmes. It is therefore important that the screening programme does not cause ill-health.¹

24.1 Population genetic screening involves testing large numbers of people, who usually have no disease symptoms, for their genetic status in relation to a particular gene or condition. Population genetic screening programs are based on the assumption that it is desirable for certain populations to be tested and for individuals to be given information about their genetic status.²

24.2 Genetic screening tests are performed on individuals who are not necessarily known to have an increased risk of developing a particular genetic condition and who have not sought medical attention.³ By contrast, other forms of medical genetic testing generally take place after individuals seek professional treatment because they, or a

1 Nuffield Institute for Health, *Quality Management for Screening: Report to the National Screening Committee* (2000), University of Leeds, Leeds, 42.

2 House of Commons Science and Technology Committee, *Human Genetics: The Science and its Consequences* (1995), House of Commons, London [81].

3 National Screening Committee, *First Report of the National Screening Committee* (1998), Health Departments of the United Kingdom, 12.

relative, have symptoms. The United Kingdom National Screening Committee has observed that:

Screening is different from most other forms of health care. For most health care, the patient comes to the clinician, who will offer the treatment available on current knowledge. This may be limited, and it is legitimate for the clinician to say this. In screening, the health service is saying to apparently healthy people, 'come to us, go through this procedure, and there will be a subsequent benefit'. Under those circumstances, it is imperative that the service is able to demonstrate that benefit will follow, and that the collective benefit will outweigh the side effects or the harm from the screening programme.⁴

24.3 Population genetic screening can be distinguished from some other types of population screening, such as programs offering mammograms to detect breast cancer, because genetic screening is not always intended to lead directly to prevention or treatment. For example, genetic screening for carrier status is intended to inform participants in making reproductive choices and, therefore, to prevent genetic conditions arising in a subsequent generation.

24.4 This chapter discusses the types of population genetic screening programs that can be undertaken and the guidelines that have been developed to regulate them. Population genetic screening programs raise a range of privacy, ethical and other issues, which are also discussed. At present, most population genetic screening programs are subject to limited specific regulation or policy guidance.

24.5 The Inquiry has concluded that there is a need for consistent national standards for the development and implementation of population genetic screening programs. The Inquiry recommends the development of national standards by the Australian Health Ministers' Advisory Council (AHMAC).

Population genetic screening programs

24.6 Population screening programs are established to offer individuals the opportunity to obtain more information about their current and future health. Armed with this information, they can make better-informed choices about health care, lifestyle and reproduction. The Health Council of the Netherlands has commented that population genetic screening programs seek 'to enable people to achieve greater autonomy and to decide upon a course of action that is acceptable to them'.⁵

4 Ibid, 14.

5 Health Council of the Netherlands: Committee Genetic Screening, *Genetic Screening* (1994), Health Council of the Netherlands, The Hague, quoted in European Society of Human Genetics, *Population Genetic Screening Programmes: Principles, Techniques, Practices, and Policies* (2000), ESHG, Birmingham, 8.

24.7 Screening programs are an expanding aspect of health care. As the cost of genetic testing decreases and the range of available tests increases, it is likely that population genetic screening will become feasible for a greater number of conditions.⁶

24.8 Screening can be undertaken in two ways:

- pro-active screening, where large populations are systematically targeted and offered screening tests; or
- opportunistic screening, where tests for an unsuspected disorder are offered by doctors when a person has sought medical advice for another reason.⁷

24.9 Population genetic screening programs differ from each other in scope and objective, and these factors will determine which population is screened, the tests offered, and the time at which screening takes place. These factors will also determine the implications and possible uses of the information generated by the screening program. Most programs share three general aims: to decrease the prevalence of a disorder, to reduce the health effects of a disorder, and to inform individuals about their reproductive choices.

24.10 Population genetic screening can give individuals information about their health that they would not otherwise have sought, and this may motivate them to seek treatment or make lifestyle changes. By identifying illness that would otherwise have gone undiagnosed, or have been detected too late for effective treatment, genetic screening can improve individual health in some cases. Screening results can also inform individual reproductive decisions and, because choices may reduce the likelihood of children being born with certain genetic disorders, screening programs can decrease the incidence of these disorders in the population.

24.11 Population screening also has research value, enabling researchers to collect information about the prevalence and incidence of particular disorders in the community. Sometimes these research objectives overlap with objectives related to the health of individuals.⁸ An example, discussed below, is the current project for screening Jewish children in Sydney private schools for Tay-Sachs disease carrier status.

24.12 Population screening may be conducted on a large scale by offering tests to entire populations or to a sub-set of individuals within a population. Screening is directed either to the widest population that can be reached (mass screening) or to a defined group (selective screening).

6 National Screening Committee, *First Report of the National Screening Committee* (1998), Health Departments of the United Kingdom, 9.

7 *Ibid.*, 12.

8 National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002.

Mass screening

24.13 Mass screening programs test entire populations or community groups in order to identify a condition or conditions suitable for treatment or prevention.⁹ Most participants will be asymptomatic and will be unaware that they might have a genetic disorder. Mass genetic screening is most appropriate for detecting treatable but often under-diagnosed conditions, and diseases such as cancer and heart disease where an individual's risk can be reduced by monitoring, medication and lifestyle changes.¹⁰ Newborn screening programs are an example of mass genetic screening.

Selective screening

24.14 Genetic screening of large populations can be time-consuming and expensive. Selective genetic screening is a more economical method of detecting individuals who carry a genetic mutation. The screening of groups with a higher prevalence of the mutation is more likely to identify affected individuals than screening the general population. Fewer resources are therefore expended on screening people who are not affected.

24.15 Selective screening programs are directed at population sub-groups that are distinguished by a common characteristic. Groups may be defined by:

- a higher risk of carrying a gene related to disease—for example, African populations have a relatively high prevalence of sickle cell anaemia;
- shared symptoms—for example, people with high cholesterol levels might be screened for genetic causes of familial hypercholesterolaemia, a disorder that causes elevated cholesterol levels and can contribute to the development of heart disease;
- phase of life—for example, newborn children or adolescents may be screened for certain genetic conditions; or
- workplace—for example, a company might screen all its employees for a genetic mutation that increases their risk of disease when working in a particular work environment.

24.16 Selective screening programs that target individuals who may be at increased risk for a genetic disorder often focus on community, racial or ethnic groups in which a disorder is more prevalent, or on individuals with a family history of an inherited disorder—for example, screening of families with a history of familial adenomatous polyposis, a form of colorectal cancer.

9 R Trent, *Molecular Medicine: An Introductory Text* (2nd ed, 1997) Churchill Livingstone, 193.

10 House of Commons Science and Technology Committee, *Human Genetics: The Science and its Consequences* (1995), House of Commons, London [97].

24.17 Cascade screening is a form of selective screening that seeks to identify and test relatives of people diagnosed with a genetic condition. Like other forms of selective screening, cascade screening is a more efficient and cost-effective method of identifying affected individuals than mass screening, and may be conducted through genetic registers. Genetic registers and family genetic information are discussed in Chapter 22.

24.18 Selected population groups defined by shared symptoms or disabilities that have a variety of causes may be screened to determine if, in some cases, the condition results from genetic causes. An example is the screening of individuals with learning disabilities to detect those with Fragile X syndrome, a genetic condition that causes mental retardation.¹¹ Screening of high-risk families for Fragile X is conducted in all Australian States.¹²

24.19 Screening programs can be targeted at populations at different stages of life: pre-implantation,¹³ prenatally,¹⁴ at birth, during early childhood or during adulthood. The stage at which screening is conducted has implications for issues of consent, counselling and the availability of treatment, which are discussed later in this chapter. For example, screening children for late-onset genetic disorders is generally regarded as unacceptable.¹⁵ Genetic screening of adolescents and adults is usually performed to identify carrier status or predict future disease. For example, the Tay-Sachs program discussed below screens adolescents for carrier status before they start families. Adult screening tests include haemochromatosis, breast cancer susceptibility and familial hypercholesterolaemia. In general, screening of adults and adolescents is recommended only for conditions that are reasonably treatable.¹⁶

24.20 Populations exposed to particular work environments might also be suitable for screening. For example, individuals with a predisposition to sickle cell anaemia may be more likely to develop the condition if exposed to carbon monoxide or cyanide. An employer might screen to determine which employees are not suited to tasks where exposure to these substances may occur. The use of genetic screening and other genetic testing in employment is discussed in Part H. Other contexts where selective screening

11 Nuffield Council on Bioethics, *Genetic Screening Ethical Issues* (1993), Nuffield Council on Bioethics, London, 15.

12 National Public Health Partnership, *An Overview of Public Health Surveillance of Genetic Disorders and Mapping of Current Genetic Screening Services in Australia* (2002), National Public Health Partnership, Canberra App 4, Table 8.

13 Screening can be performed through pre-implantation genetic diagnosis—testing cells from an embryo created through in-vitro fertilisation (IVF).

14 Screening at this stage is usually conducted only to detect serious disorders. This form of screening is not discussed in detail in this Report. See Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, IP 26 (2001), ALRC, Sydney [1.36]–[1.40].

15 AHEC guidelines state that ‘presymptomatic testing of children for adult onset disorders for which there is no preventative strategy or treatment ... is not considered ethical. Such testing would remove from the child the possibility of deciding, on reaching adulthood, whether or not to have the test in question, and the timing of testing’: National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: an Information Paper* (2000), NHMRC, Canberra [3.4.3.2]. Genetic testing of children and access to insurance is discussed in more detail in Ch 26.

16 House of Commons Science and Technology Committee, *Human Genetics: The Science and its Consequences* (1995), House of Commons, London [83].

might be used include immigration, sport, and law enforcement. Genetic testing in these contexts is discussed elsewhere in this Report.¹⁷

Types of population genetic screening programs

24.21 The genetic tests used in population genetic screening programs can detect:

- current conditions that have a genetic cause and may have an available treatment (screening for current conditions);
- the existence of a disorder before the onset of symptoms, or the existence of a genetic mutation that predisposes to a disorder (predictive screening); or
- carrier status for a genetic condition (carrier screening).

24.22 Each type of genetic screening test is discussed below, with reference to Australian examples.

Screening for current conditions—Newborn screening

24.23 Screening for current conditions can be used to detect conditions very early in life in circumstances where prompt treatment may be effective. Such screening may also be used to identify symptomatic individuals where the genetic cause of their symptoms has not yet been established.

24.24 Screening soon after birth, or during early childhood, has three major aims:

- to enable prompt detection of treatable genetic conditions;
- to detect inherited conditions, allowing parents to make reproductive decisions before having more offspring; and
- to clarify the causes of an infant or child's condition.

24.25 In all States and Territories, newborns are screened for a number of genetic conditions, including phenylketonuria (PKU).¹⁸ Newborns affected by PKU are unable to break down the amino acid phenylalanine and, untreated, the disorder causes significant brain damage within months. Early detection allows newborns to be placed on a special diet that will prevent brain damage and ensure normal development.

24.26 The nature of the genetic conditions tested for in newborn screening programs is such that testing cannot be delayed until the child is older. The conditions require immediate medical or dietary intervention to preserve the life or health of the infant. All newborns are tested unless parents refuse consent. Parents are provided with

17 See Ch 38 (sport); Part J (law enforcement).

18 Practice varies somewhat among the States and Territories but, as a general matter, newborns are tested for phenylketonuria (PKU), hypothyroidism, galactosaemia, cystic fibrosis and certain other metabolic conditions. The storage, use and disclosure of newborn screening cards, including for law enforcement purposes, are discussed in Ch 19.

leaflets and other information about the nature and value of the test for neonatal health and in practice consent is rarely refused.¹⁹ Tests are carried out by taking a small sample of blood via a heel-prick, and the blood is stored on blotting paper cards. A sample newborn screening card is reproduced at the end of this chapter.

24.27 Other screening programs detect current conditions for which there is limited treatment in order to confirm a diagnosis or to facilitate informed reproductive choice. For example, as noted above, all States conduct early childhood screening for Fragile X syndrome, targeting individuals with intellectual disabilities. The identification of a child with Fragile X is often followed by cascade screening of relatives²⁰ and can help prevent families from bearing more children who will have the disorder.

24.28 Screening programs can also provide peace of mind to parents by clarifying the cause of a child's condition. The symptoms of Duchenne muscular dystrophy (DMD), another genetic disorder with limited treatment options, are often not correctly diagnosed for some years.²¹ In relation to the possible benefits of screening for DMD, one parent of a child with DMD noted that, had she known the reason for his symptoms earlier:

I could have done more for him ... We would never have labelled him lazy or a dreamer. We could have started steroid treatment and other therapies earlier and to the best advantage. More importantly, he could have had a name for his difference and understood the weakness he felt.²²

Carrier screening—Tay-Sachs disease

24.29 Carrier testing is used to detect individuals who carry a recessive gene that could be passed on to their children. As discussed in Chapter 2, if two carriers of a recessive gene have children, the child may receive copies of the defective gene from each parent and develop the genetic condition. For X-linked recessive conditions, male children who receive one copy of the defective gene may develop the condition.

24.30 Carriers generally do not suffer from the disorder caused by the genetic mutation they carry and thus are often unaware of their status and of the fact that they may pass a genetic condition on to their children. Carrier screening programs seek to alert individuals to their carrier status so that they are able to make informed decisions about reproduction. These may include choosing not to have children, having children through sperm or egg donation, adoption, or pre-implantation genetic diagnosis.

19 L Skene, 'Access to and Ownership of Blood Samples for Genetic Tests: Guthrie Spots' (1997) 5(2) *Journal of Law and Medicine* 137, 138.

20 National Public Health Partnership, *An Overview of Public Health Surveillance of Genetic Disorders and Mapping of Current Genetic Screening Services in Australia* (2002), National Public Health Partnership, Canberra, 54.

21 Duchenne Muscular Dystrophy is not the subject of any population genetic screening program in Australia but has been screened for in other countries: See D Bradley, E Parsons and A Clarke, 'Experience with Screening Newborns for Duchenne Muscular Dystrophy in Wales' (1993) 306 *British Medical Journal* 357.

22 D Robins, *Submission G154*, 10 April 2002.

24.31 An example of selective carrier screening is a program that screens students at a number of private Jewish schools in Sydney for Tay-Sachs disease (TSD) carrier status. In the general population, only about one in 300 people are carriers for TSD, but in the Ashkenazi Jewish community the rate is closer to one in 30. TSD causes delayed development followed by neurodegeneration, blindness and eventually death, usually by the age of five.²³

24.32 Under the program, students are educated about the implications of carrier status and about the disease in general, and given the opportunity to take a voluntary screening test. The program employs a gene trustee who holds results until the student requests them.²⁴ If a student elects to receive his or her test results, and the results are positive, the student is contacted by a genetic counsellor.²⁵ The program aims to reduce the incidence of TSD within the Jewish community by enabling individuals to make more informed reproductive decisions.

24.33 The project also has research aims. The organisers collect and analyse data about the uptake of testing, the effect of educating participants about the condition, the psychosocial implications of screening, and participants' reactions to test results.²⁶

24.34 Population genetic screening programs for carrier status, particularly those targeted at higher risk populations, have had considerable success in reducing the incidence of some genetic disorders. For example, a national screening program for carriers of β -thalassaemia in Sardinia has reduced the incidence of the disorder from one in 250 live births in 1974 to one in 4000 in 1996.²⁷

Predictive genetic screening—Haemochromatosis

24.35 Chapter 2 discussed the importance of penetrance, a measure of the likelihood that a person carrying a genetic mutation will develop the disease associated with it.²⁸ Penetrance is relevant to population genetic screening because screening often targets asymptomatic individuals. From a health funding perspective, screening for only slightly penetrant genetic mutations may not be cost-effective because most individuals who are tested will not develop the disease. Also, if healthy people are offered genetic tests that may suggest they are predisposed to a disorder, this information should be accurate enough to enable them to take action, rather than merely creating doubt and anxiety.

23 L Burnett and others, 'The Tay-Sachs Disease Prevention Program in Australia: Sydney Pilot Study' (1995) 163 *Medical Journal of Australia* 298.

24 See Ch 18.

25 K Barlow-Stewart and others, 'Development and Evaluation of a Genetic Screening Program for Tay-Sachs Disease and Cystic Fibrosis for Australian Jewish High School Students' (2002) *Journal of Medical Genetics*.

26 Ibid.

27 J Dalton and L Krishnamurti, 'The Newborn Screen: An Underutilized Tool for Screening and Counselling for Thalassaemia' (2001) 31(3) *CURA Reporter* 19, 21.

28 Ch 2.

24.36 Predictive genetic screening is mainly undertaken to inform a larger proportion of the community whether or not they have a condition or a predisposition to a condition, and this in turn may enable them to make health decisions that may slow or treat the condition. Predictive test results might also influence a participant's reproductive decisions.

24.37 An example of population genetic screening for predisposition to disease is HaemScreen, a pilot genetic testing program conducted by the Murdoch Childrens Research Institute in a number of Melbourne workplaces.²⁹ Participants agree to be tested for genetic predisposition to haemochromatosis, a condition that causes the body to accumulate excess iron leading to irreversible organ damage.³⁰ Part of the rationale behind HaemScreen is to target a section of the population that is less likely to visit their doctor. Screened individuals who are found to be at risk of developing the condition may then take preventive measures, such as giving blood regularly.

Current regulation and guidance

24.38 The conduct of population genetic testing programs is subject to the same forms of regulation that govern genetic testing generally, including information and health privacy legislation, the law relating to consent to medical treatment and medical testing, and other laws and guidelines regulating the medical profession.

24.39 As population genetic screening programs are largely conducted through state and territory government health authorities, the collection, storage, use and disclosure of information derived from them will be subject to state and territory information and health privacy legislation, where it exists.³¹ The *Privacy Act 1988* (Cth) (*Privacy Act*) will also apply to some screening programs, for example those conducted by Australian Capital Territory public health authorities.

24.40 Australia does not have a national policy statement on population genetic screening. However, some guidance has been provided by the National Health and Medical Research Council (NHMRC)³² and the Royal Australian College of General Practitioners (RACGP).³³ Internationally, the World Health Organization (WHO) has developed widely accepted ethical standards for population genetic screening.³⁴

29 Although it may be argued that the program should be categorised as screening for a current condition. Some of those screened will not only have the genetic predisposition, but also existing excess iron levels.

30 Haemochromatosis may be connected with carrying one or more copies of a genetic mutation that has been linked with the condition: M Worwood, 'Early Detection of Genetic Hemochromatosis: Should All Young Adults Be Offered the Genetic Test?' (2000) 4 *Genetic Testing* 219, 219–223.

31 See Ch 7.

32 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: an Information Paper* (2000), NHMRC, Canberra.

33 Royal Australian College of General Practitioners, *Guidelines for Preventive Activities in General Practice* (2002), RACGP, Melbourne.

34 World Health Organization, *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services* (1997), WHO, Geneva. See also European Society of Human Genetics, *Population Genetic Screening Programmes: Principles, Techniques, Practices, and Policies* (2000), ESHG, Birmingham, 5–8; M Worwood, 'Early Detection of Genetic Hemochromatosis: Should All Young Adults Be Offered the Genetic Test?' (2000) 4 *Genetic Testing* 219, 224–225.

24.41 In relation to newborn screening, the Human Genetics Society of Australasia (HGSA) and the Paediatrics Division of the Royal Australasian College of Physicians (RACP) have released a joint policy statement (the HGSA/RACP policy statement).³⁵ Some state health departments have also produced guidelines on newborn screening.³⁶ These statements and guidelines are discussed below.

General population genetic screening guidelines

24.42 The NHMRC deals briefly with population genetic screening in its information paper, *Ethical Aspects of Genetic Testing*.³⁷ The paper defines a ‘screening test’ and outlines the contexts in which screening might be undertaken and the objectives of screening programs. The paper states that written consent should be obtained for screening tests and counselling is recommended following an abnormal result. The paper notes that the information and counselling needs of screening program participants differ from those who have had experience of a disorder in their own families.

Those undergoing a screening test will often have little understanding of the disorder in question, as they may never have met an affected person or had the opportunity to learn about the condition. Further, there is a relatively small chance that the test result will be abnormal, and if the test result is abnormal, the chance that they will develop the disorder may be only a little or moderately increased.³⁸

24.43 The RACGP’s *Guidelines for Preventive Activities in General Practice* includes a chapter devoted to genetic screening.³⁹ The guidelines state that there is no evidence to support routine genetic screening of all patients, but notes that this may change in the future.⁴⁰ Doctors should only screen individuals who appear to be at a higher risk for a genetic condition, after a detailed medical history has been taken.⁴¹ The guidelines list general factors that may suggest the presence of genetically determined disease and note that some ethnic and racial groups are at increased risk of certain genetic conditions.

35 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *Policy Statement on Newborn Screening*, 1 June 1999.

36 NSW Health Department, *Guidelines for Newborn Screening* (2001); Department of Human Services Victoria, *Guidelines for Newborn Screening* (2001).

37 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: an Information Paper* (2000), NHMRC, Canberra. Any research activities associated with population genetic screening are governed by the NHMRC National Statement: National Health and Medical Research Council, *National Statement on Ethical Conduct in Research Involving Humans* (1999), NHMRC, Canberra. The regulation of human genetic research is discussed in Part D.

38 National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: an Information Paper* (2000), NHMRC, Canberra, 19.

39 Royal Australian College of General Practitioners, *Guidelines for Preventive Activities in General Practice* (2002), RACGP, Melbourne, Ch 4.

40 *Ibid.*, 12.

41 *Ibid.*, 12.

24.44 Ethical standards for population genetic screening have been developed internationally. In 1968, WHO released general guidelines for the conduct of population screening programs.⁴² These guidelines were not specifically directed at genetic screening, and were developed before carrier testing was common. A fundamental presumption of the 1968 WHO guidelines was that conditions screened for should be treatable. This presumption does not apply to all forms of genetic screening, for example, carrier testing. In 1997 the general guidelines were augmented by specific guidance on genetic screening and testing.⁴³ These additional WHO guidelines state that:

- genetic screening should be voluntary (with the exception of newborn screening where early diagnosis and treatment will benefit the newborn);
- genetic screening should be preceded by adequate information about the purpose and possible outcomes of the screening and potential choices to be made;
- anonymous screening for epidemiological purposes may be conducted after notification of the population to be screened, but results should not be disclosed to employers, insurers, schools or others without the individual's consent, in order to avoid possible discrimination;
- in rare cases where disclosure may be in the best interests of the individual or for public safety, the health provider may work with the individual towards a decision by him or her;
- test results should be followed by genetic counselling, particularly where they are unfavourable;
- if treatment or prevention exists or is available, this should be offered with a minimum of delay.⁴⁴

24.45 The European Society for Human Genetics (ESHG) has stated that any new population genetic screening program should be voluntary and have defined health goals, a defined target population, laboratory controls, and measures to ensure the confidentiality of results and the provision of education. The ESHG also stated that population genetic screening programs should be evaluated through pilot programs before being introduced and that these programs should evaluate the test, its clinical validity and its acceptability, uptake rates, the use of the test results in decision making, the psychosocial consequences of the test, the utility of the screening process and total program costs.⁴⁵

42 J Wilson and G Jungner, *The Principles and Practice of Screening for Disease*, 34 (1968), World Health Organization.

43 World Health Organization, *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services* (1997), WHO, Geneva.

44 Ibid, Table 4. The new guidelines also emphasise the need to avoid stigmatisation of groups with higher incidences of a disorder.

45 European Society of Human Genetics, *Population Genetic Screening Programmes: Recommendations* (2000), ESHG, Birmingham, IV-1, IV-5, and section IV generally.

24.46 In the United Kingdom, a national body was established in 1996 to assess and oversee population screening programs, including those for genetic disorders. The National Screening Committee (NSC) advises government on all aspects of screening policy, the implementation of new screening programs and the evaluation of current programs. Since 1996, the National Health Service has been directed not to implement new population screening programs until they have been reviewed by the NSC. The NSC assesses screening programs against detailed criteria based on the WHO standards. The NSC emphasises that screening should involve more than just the offer of a test—it should educate the public and allow individuals to make free and informed health decisions.⁴⁶

Newborn screening guidelines

24.47 The HGSA/RACP policy statement on newborn screening programs recommends that genetic disorders should be screened for only where:

- there is benefit for the individual from early diagnosis;
- the benefit is reasonably balanced against financial and other costs;
- there is a reliable test suitable for newborn screening; and
- there is a satisfactory system in operation to deal with diagnostic testing, counselling, treatment and follow-up of patients identified by the test.⁴⁷

24.48 The HGSA/RACP policy statement provides that participation in the program should not be mandatory and the personal information of participants should remain private.⁴⁸ The statement also recommends which disorders should be screened for, and with what priority.⁴⁹

24.49 The New South Wales Health Department and the Victorian Department of Human Services have also developed guidelines for newborn screening programs.⁵⁰ The New South Wales guidelines emphasise the need for early detection and treatment of the genetic disorders covered by the program. The guidelines cover:

- the conditions to be screened for and procedures for testing, including the timing of the test, taking of the blood, and transfer of samples to the laboratory;
- the information to be provided to parents, and obtaining verbal consent from parents or guardians (consent must be preceded by a documented discussion about the testing process and refusals must be in writing and signed);

46 National Screening Committee, *First Report of the National Screening Committee* (1998), Health Departments of the United Kingdom, 12, 17.

47 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *Policy Statement on Newborn Screening*, 1 June 1999, 2.

48 *Ibid* [3.4], [3.6], [4.4].

49 *Ibid*, 6.

50 NSW Health Department, *Guidelines for Newborn Screening* (2001); Department of Human Services Victoria, *Guidelines for Newborn Screening* (2001).

- follow-up and re-testing procedures where results indicate a disorder;
- compliance with relevant privacy legislation and the New South Wales Health Department's Information Privacy Code of Practice; and
- the nomination by hospitals of a newborn screening liaison officer.⁵¹

24.50 The Victorian guidelines are similar, but include an additional requirement that parents who wish to refuse testing are referred to a newborn screening counsellor and required to sign a written statement of refusal stating that they understand the potential risk to their baby.⁵²

24.51 The Inquiry understands that newborn screening in the Australian Capital Territory follows the New South Wales guidelines. South Australia (which also conducts screening of Tasmanian and some Northern Territory newborns) and Western Australia follow the HGSA/RCPA policy statement.⁵³

Issues and problems

24.52 Population genetic screening programs give rise to a range of ethical, privacy and discrimination-related concerns. Individuals can experience anxiety on receiving test results, particularly if the information means they will be faced with difficult choices, such as the possible termination of pregnancy. Consent and counselling issues are important. Screening may reveal the genetic status of family members who have not chosen to be screened, raising concerns about privacy and the 'right not to know'. Screening may result in stigmatisation of certain genetic disorders. Those who refuse to be screened may also suffer social stigmatisation.⁵⁴ The information generated may have implications in contexts other than health, for example in employment and insurance.

24.53 Many of the ethical, privacy, and discrimination-related issues raised by population genetic screening programs are similar to those raised by genetic testing more generally. However, screening programs also raise distinct issues. In particular, the large scale or 'production line element' of population screening may place time constraints on obtaining consent and providing counselling.⁵⁵ Test reliability and cost also have distinct implications in the context of population screening programs. A range of privacy, ethical and other issues raised by population genetic screening programs is discussed below, with reference to views and comments expressed in submissions and consultations.

51 NSW Health Department, *Guidelines for Newborn Screening* (2001).

52 Department of Human Services Victoria, *Guidelines for Newborn Screening* (2001), 3.

53 National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002 App 2.

54 European Society of Human Genetics, *Population Genetic Screening Programmes: Recommendations* (2000), ESHG, Birmingham, II.

55 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

Privacy

24.54 Population screening for genetic conditions generates genetic information about individuals. People may wish to keep their genetic information private, and may be concerned that screening programs will lead to others—such as family members, employers or insurance companies—being informed that they have a genetic condition.

24.55 As in other contexts, the privacy issues raised by population genetic screening programs include those that derive from the familial or collective nature of genetic information, and from the need to respect the right not to know (see Chapter 7). If an individual participates in a screening program and learns that he or she has a genetic condition, this may indicate that family members (genetic relatives) also have the condition or are carriers. This raises issues in relation to disclosure of information by health professionals to genetic relatives and genetic relatives' rights of access to this information (see Chapter 21).

24.56 Some submissions raised concerns about the privacy of the information collected and stored by population screening programs.⁵⁶ Others argued that privacy concerns could present a barrier to population screening of disorders for which there are effective and available treatments.⁵⁷ The Office of the Federal Privacy Commissioner (OFPC) emphasised the need to protect the privacy of participants in screening programs.⁵⁸

Since population screening will give rise to large aggregations of genetic and other personal information, particular attention should be given to proposals for the use of that information for research or other secondary purposes to ensure that the privacy interests of participants are respected.⁵⁹

24.57 The OFPC asserted that the *Privacy Act* provides a 'robust and flexible framework for genetic privacy' for the purposes of population genetic screening.⁶⁰ The HGSA also submitted that the current regulatory system appears to provide adequate protection for genetic information collected in population screening programs.⁶¹

24.58 The Inquiry has concluded that, given the recommendations in this Report for reform of the *Privacy Act* and harmonisation of federal, state and territory information and health privacy legislation as it relates to human genetic information,⁶² no additional reform of privacy law is required to address issues raised by population genetic screening programs specifically.

56 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; K Monro and others, *Submission G219*, 3 December 2002.

57 For example, C Maurer, *Submission G113*, 18 March 2002.

58 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

59 *Ibid.*

60 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

61 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002. See also Department of Human Services Victoria Genetics Advisory Committee, *Submission G089*, 24 January 2002.

62 See Ch 7, 8, 21, 22.

Right not to know

24.59 The familial nature of genetic information means that participation in a screening program may reveal information about an individual's relatives, who may not wish to know their genetic status.

24.60 Protection of the right not to know is important where individuals are screened for conditions that have no cure or effective treatment.⁶³ While some people may wish to be screened for all kinds of conditions, others may wish to know their results only where the knowledge will enable them to take action to prevent or treat illness.

24.61 Screening newborn infants and children raises particular issues. If children are screened at birth, parents may be better able to understand their child's condition and make more informed decisions about later reproduction. However, the knowledge may place significant burdens on parents and child, especially where the condition is known to develop late in life. Individuals who are screened at birth or in early childhood may be given genetic information that they would prefer not to know. Children may have particular problems dealing with the psychosocial effects of knowing that they have a genetic condition that will not cause symptoms until later in life. For this reason, children are rarely tested for late-onset disorders.

24.62 Related concerns arise from the fact that an applicant for insurance is obliged to disclose every matter that is known to be relevant, or which reasonably ought to be known to be relevant, to the insurer. Where an individual is aware that he or she underwent a genetic test as a child, that individual is obliged to disclose the results for insurance purposes, if relevant to the risk, with possible adverse insurance consequences.⁶⁴ These matters are discussed in Chapter 26.

Consent and counselling

24.63 Individuals should not participate in testing as part of a population genetic screening program without giving informed consent. Where the program involves a research component, consent to participation in research should be obtained (see Chapter 15).⁶⁵ As genetic testing can yield information that is distressing to the recipient, whether they are the parent of an affected child or are themselves the subject of the test, adequate counselling should be made available both before screening and after receipt of test results.

24.64 The Institute of Actuaries of Australia observed that procedures for obtaining consent and counselling in population genetic screening programs may 'have to be more standardised than is best practice for testing of an individual or family in a clinic'.⁶⁶ Nevertheless, the need for voluntary and informed participation in population

63 Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003.

64 See Ch 26, Recommendation 26–3.

65 The use of newborn screening cards and other human tissue collections in research is discussed in Ch 19.

66 Institute of Actuaries of Australia, *Submission G224*, 29 November 2002.

screening programs remains critical, despite the benefits such programs may produce.⁶⁷ There may be a tension between the ethical consideration of individual autonomy and the utilitarian justification of the social good. Participants should be made fully aware of the implications of their involvement, including that in some circumstances they may be required to reveal the results of a screening test to insurers. In general, the OFPC submitted that there was 'a need for greater education and counselling to enable those individuals involved in screening programs to understand the implications of their participation'.⁶⁸

24.65 Where testing is offered by members of the medical profession, particularly in a hospital or other health service environment, individuals may feel obliged to follow what they perceive as a requirement or strong recommendation to take the test. In these circumstances, questions may be raised about whether consent is properly obtained. Consent for newborn screening is an example of this problem because consent is often obtained in hospital during labour or shortly afterwards. While parents are provided with information leaflets about newborn screening by hospital authorities, there is no formal check that they have read and understood them.⁶⁹ Doubts have been expressed about whether this constitutes proper consent.⁷⁰

24.66 Professor Loane Skene has suggested that the process of obtaining consent to newborn testing in Australia is more akin to 'informed refusal' than 'informed consent' because parents are informed about the test and may refuse it, but are otherwise taken to consent.⁷¹ A registered nurse has been quoted as referring to the consent process as involving 'uninformed nondissent' because many women do not understand what they are consenting to when they agree to their newborn being given a heel-prick test in the highly stressful situation of delivering a baby. She stated that 'most of them don't get it ... [t]hey don't even remember it'.⁷²

Reliability and cost

24.67 In the United Kingdom, the NSC has noted that 'it is because screening is rarely precise that much of the potential for harm may come'.⁷³ Genetic tests performed as part of a screening program should produce reliable and accurate results. Screening may cause unnecessary trauma and inconvenience if individuals are supplied

67 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

68 Ibid.

69 Children's Hospital at Westmead, *Consultation*, Sydney, 19 November 2002.

70 L. Skene, 'Access to and Ownership of Blood Samples for Genetic Tests: Guthrie Spots' (1997) 5(2) *Journal of Law and Medicine* 137, 138.

71 Ibid, 138.

72 Quoted in J Lyttle, 'Is Informed Consent Possible in the Rapidly Evolving World of DNA Sampling?' (1997) 156 *Canadian Medical Association Journal* 257, 258.

73 National Screening Committee, *First Report of the National Screening Committee* (1998), Health Departments of the United Kingdom [3.6.1].

with incorrect information about their genetic status. Lack of reliability will also render any population health information generated by the program scientifically unsound.⁷⁴

24.68 Screening tests can produce two main categories of unreliable result:

- False positive. The results indicate that the person has a genetic disorder when they do not. Individuals may undergo unnecessary treatment and suffer the anxiety of thinking they are ill.
- False negative. The results indicate that the person does not have a genetic disorder when they do. Individuals may ignore the symptoms of the disorder as they develop, thinking themselves to be healthy, and appropriate treatment may be delayed, with potentially serious consequences. Individuals may also continue with unhealthy behaviour where they have been found clear of genetic risk factors that can be offset by lifestyle changes.

24.69 In its submission, the HGSA highlighted the need for reliable screening tests. The HGSA stated that new tests for use in the public health arena ‘should be assessed for utility, sensitivity, specificity and likelihood of facilitating the desired outcome’.⁷⁵ Assessment should continue throughout the early stages of the implementation of a screening program to ensure these desired outcomes are being met. The HGSA/RACP policy statement provides that screening program performance should be assessed regularly, including assessment of test sensitivity, specificity, positive predictive value, timeliness of reporting, and outcome of diagnosed patients.⁷⁶

24.70 Screening for haemochromatosis is an example of a program that has been criticised for offering a test with insufficient predictive power. There is still scientific and medical debate as to whether there is sufficient correlation between having the genetic mutation tested for, and developing the consequences of haemochromatosis.⁷⁷

24.71 While population screening programs can produce long-term benefit in reducing disease, they may over-burden health care systems in the short-term. This may occur where individuals are identified as suffering from a condition that the health care system is not fully equipped to cope with. For example, the cost of treatment may not have been budgeted for, or counselling services to deal with the needs of recently diagnosed sufferers may not be sufficient.

74 For example, the reliability of current tests used to detect haemochromatosis has been queried: E Beutler and others, ‘Penetrance of 845G-A (C282Y) HFE Hereditary Haemochromatosis Mutation in the USA’ (2002) 359 *Lancet* 211.

75 Human Genetics Society of Australasia, *Submission G050*, 14 January 2002.

76 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *Policy Statement on Newborn Screening*, 1 June 1999 [3.6].

77 M Worwood, ‘Early Detection of Genetic Hemochromatosis: Should All Young Adults Be Offered the Genetic Test?’ (2000) 4 *Genetic Testing* 219, 226; E Beutler and others, ‘Penetrance of 845G-A (C282Y) HFE Hereditary Haemochromatosis Mutation in the USA’ (2002) 359 *Lancet* 211. Compare A Poullis and others, ‘Correspondence: Clinical Haemochromatosis in HFE Mutation Carriers’ (2002) 360 *Lancet* 411; K Allen, ‘Letter: Screening for Hereditary Haemochromatosis Should be Implemented Now’ (2000) 320 *British Medical Journal* 183.

24.72 Medicare funds only a small number of genetic tests (see Chapter 10) and this may act as a barrier to implementing some screening programs and as a disincentive to preventive screening in general practice.⁷⁸

Submissions and consultations

24.73 In DP 66 the Inquiry proposed the development of nationally consistent policies and practices in relation to the implementation and conduct of population genetic screening programs, covering such matters as informed consent, counselling and testing standards.⁷⁹

24.74 This proposal met with general approval in submissions. Many submissions highlighted the need for tests used in population genetic screening programs to meet agreed standards for reliability, sensitivity and utility.⁸⁰ Members of the National Public Health Partnership Public Health Genetics Working Group suggested that a harmonised approach would facilitate the collection of more comprehensive information about programs, enabling them to be evaluated more effectively.⁸¹

24.75 The Australian Society of Medical Research noted that population screening and genetic registers 'have been important components of the effective delivery of health care in this country' and stated that the Society 'would be concerned if new actions were taken that would make this genetic information regulated in a differential way to other sensitive medical information'.⁸²

24.76 DP 66 also asked whether the Human Genetics Commission of Australia (HGCA) should play a role in regulating population genetic screening programs. Opinion was divided on this issue. Some suggested that the HGCA might set standards for evaluating programs and give advice on the development of policy.⁸³ Others

78 National Health and Medical Research Council, *Promoting the Health of Australians: A Review of Infrastructure Support for Health Advancement* (1996), NHMRC, Canberra, 44.

79 Australian Law Reform Commission and Australian Health Ethics Committee, *Protection of Human Genetic Information*, DP 66 (2002), ALRC, Sydney, Proposal 21–1.

80 M Delatycki, *Submission G240*, 20 December 2002; Cancer Council Victoria Cancer Genetics Advisory Committee, *Submission G195*, 27 November 2002; Law Society of New South Wales, *Submission G285*, 18 December 2002; D Robins, *Submission G181*, 29 October 2002; N Stott Despoja, *Submission G198*, 27 November 2002; K Monro and others, *Submission G219*, 3 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Department of Health Western Australia, *Submission G271*, 23 December 2002; Genetic Support Council WA, *Submission G243*, 19 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002; Association of Genetic Support of Australasia, *Submission G284*, 25 December 2002; Office of the Federal Privacy Commissioner, *Submission G294*, 6 January 2003; Human Genetics Society of Australasia, *Submission G267*, 20 December 2002; Department of Human Services South Australia, *Submission G288*, 23 December 2002; Law Institute of Victoria, *Submission G275*, 19 December 2002; NSW Health Department, *Submission G303*, 13 January 2003.

81 National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002.

82 Australian Society for Medical Research, *Submission G124*, 18 March 2002.

83 Queensland Government, *Submission G274*, 18 December 2002; Centre for Genetics Education, *Submission G232*, 18 December 2002; Institute of Actuaries of Australia, *Submission G224*, 29 November 2002; N Stott Despoja, *Submission G198*, 27 November 2002; Centre for Law and Genetics, *Submission G255*, 21 December 2002.

considered that the HGCA should act as a regulatory body and assess whether tests and programs meet population screening standards.⁸⁴ The HGSA suggested the creation of a new national screening authority in Australia, similar to the NSC in the United Kingdom.⁸⁵

Inquiry's views

24.77 The Inquiry has concluded that it is desirable to have national standards for the development and implementation of population genetic screening programs and newborn screening programs.

24.78 Public acceptance is integral to the success of population genetic screening programs. National standards would improve public awareness of, and confidence in, genetic screening programs. They would also promote a consistent approach to the provision of counselling and educational programs for participants.

24.79 Access to genetic screening programs is currently subject to financial and resource constraints. By creating a focus for the development of screening programs, national standards for genetic screening programs might encourage the introduction of programs on an Australia-wide basis, addressing inequities in access.⁸⁶

24.80 Cost-benefit considerations have a significant impact on the introduction of population genetic screening programs. In some cases, such as in relation to the HaemScreen program, there is considerable professional debate about costs and benefits. Creating a national benchmark for determining whether a screening program should be established would help to address divergences of opinion on the efficacy of screening. National standards for assessing the costs and benefits of screening programs could also encourage the most effective use of limited health care resources, a goal to which a range of ethical considerations are relevant.⁸⁷

24.81 National standards can also promote comprehensive, standardised reporting and data collection on the incidence and prevalence of disorders and the impact of screening.⁸⁸ This would facilitate public health planning and the evaluation and improvement of screening programs.

84 Law Institute of Victoria, *Submission G275*, 19 December 2002; M Delatycki, *Submission G240*, 20 December 2002.

85 Human Genetics Society of Australasia, *Submission G267*, 20 December 2002.

86 The National Public Health Partnership Genetics Working Group stated that a national approach 'would ensure the capacity for a considered policy analysis to decide if screening is appropriate, and if so, ensure equity of accessibility across jurisdictions': National Public Health Partnership, *An Overview of Public Health Surveillance of Genetic Disorders and Mapping of Current Genetic Screening Services in Australia* (2002), National Public Health Partnership, Canberra, 36.

87 See National Health and Medical Research Council, *Ethical Considerations Relating to Health Resource Allocation Decisions* (1993), NHMRC, Canberra.

88 The National Public Health Partnership Genetics Working Group reported that data from newborn screening programs are not recorded or analysed in any systematic way: National Public Health Partnership, *An Overview of Public Health Surveillance of Genetic Disorders and Mapping of Current Genetic Screening Services in Australia* (2002), National Public Health Partnership, Canberra, 31. In the United Kingdom, the Nuffield Institute for Health has identified the lack of standardised data as a barrier

24.82 The desirability of a national approach to screening programs has been recognised previously. In 1996, in recommending that the National Public Health Partnership clearly delineate areas for which there should be national responsibility for action, the NHMRC suggested that these areas include clinical guidelines with particular reference to population screening.⁸⁹ The Inquiry understands that the National Public Health Partnership Public Health Genetics Working Group has offered to draw up national newborn screening guidelines for the Australian Health Ministers' Advisory Council but has not yet been asked to do so.⁹⁰

24.83 In the Inquiry's opinion, AHMAC is the most appropriate body to initiate the development of national standards for population genetic screening programs. This task fits within its function of advising on national health issues. AHMAC has also laid the foundations for these standards through the National Public Health Partnership's review of screening programs in Australia, and could draw on the experience and knowledge gained through this work. The NHMRC and the HGCA should have input into the development of these standards, to provide advice on the ethical and research aspects of screening, and on technical matters. Other key professional bodies, such as the HGSA, should also have input where appropriate.

24.84 New standards should take account of Australian and international best practice and existing Australian and international statements and guidelines, including those developed in the United Kingdom by the NSC. Population genetic screening standards should cover such matters as informed consent, counselling, testing standards (including quality assurance) and cost-benefit considerations. At a minimum, national standards should require for all population screening programs that:

- consent to screening is voluntary;
- consent to screening is informed about the purpose of the screening, the meaning of test results, and the factors that individuals should consider in deciding whether or not to participate;
- appropriate counselling mechanisms are in place;
- any available treatments are offered without delay;
- screening tests meet minimum standards of reliability and penetrance;
- quality assurance and auditing mechanisms are in place; and

to improving population screening programs: Nuffield Institute for Health, *Quality Management for Screening: Report to the National Screening Committee* (2000), University of Leeds, Leeds, 50. See also D Taylor, 'Quality and professionalism in health care: a review of current initiatives in the NHS' (1996) 312 *British Medical Journal* 326, 626–629.

89 National Health and Medical Research Council, *Promoting the Health of Australians: A Review of Infrastructure Support for Health Advancement* (1996), NHMRC, Canberra, xxxii.

90 National Public Health Partnership Public Health Genetics Working Group, *Consultation*, Melbourne, 22 October 2002. See Ch 23.

- each screening program has a designated person who is responsible for ensuring that standards are met; and
- there is an acceptable balance between the costs of the program and its likely individual and community benefits.

24.85 Concerns have been expressed that obtaining more explicit consent to newborn screening tests may have the unwanted effect of decreasing participation in screening programs.⁹¹ However, there are ways to address this issue: appropriate standards for population screening programs can promote universal participation as well as informed decision making. For example, one response could be to introduce a ‘two-stage’ consent process. Informed consent could first be sought well before delivery, for example at the time the hospital booking is made or when antenatal classes are taken. A confirmation of this decision could be sought after the birth when the heel-prick procedure would usually be performed. At that time, the newborn’s parents could be given any additional information they require and provided with the opportunity to confirm or revise their earlier decision.

24.86 The OFPC suggested that a form of ‘third party gene broker’, similar to the ‘gene trustee’ system described in Chapter 18, could be a solution to privacy problems posed by some population genetic screening programs.⁹² This system could be appropriate for some screening programs, particularly those that are relatively discrete in scope, by ensuring the privacy and anonymity of participants. A requirement for proponents of population genetic screening programs to consider the use of a gene trustee system could be included in national standards for population genetic screening.

Recommendation 24–1. The Australian Health Ministers’ Advisory Council, in cooperation with the National Health and Medical Research Council, the Human Genetics Commission of Australia and key professional bodies, should develop national standards in relation to the development and implementation of:

- population genetic screening programs—covering such matters as informed consent, counselling, testing standards, quality assurance, cost-benefit considerations, and reporting and data collection; and
- newborn screening programs—promoting both universal participation and informed decision making by parents.

91 In general, such concerns expressed in submissions to the Inquiry related to consent to the storage of samples (see Ch 19) rather than to consent to testing itself. In any case, it may be that the more time and effort that is given to explaining the purpose of screening tests and the possible outcomes, the more likely that people will comply. In relation to prenatal screening, see T Marteau and others, ‘Factors Influencing the Uptake of Screening for Open Neural-tube Defects and Amniocentesis to Test for Down’s Syndrome’ (1989) 96 *British Journal of Obstetrics and Gynaecology* 739.

92 Office of the Federal Privacy Commissioner, *Submission G143*, 22 March 2002.

LEAVE BLANK FOR GUTHRIE CARD



NSW NEWBORN SCREENING PROGRAMME

Baby's
Last name

Mother's
Full Name

Baby's
Date of Birth **Sex M/F**
Birth
Weightg **Gestation**.....weeks

Date of Sample Test less than 48 hr []
Feeds: Breast/Formula/Soy based/TNP/Others

Hospital of Birth

Hospital/Sample Source
Paediatrician/Doctor
In Charge

Relevant Clinical Information

Initial **Repeat**
Test [] Test []

COMPLETE ALL DETAILS REQUESTED ABOVE.
COMPLETELY FILL EACH CIRCLE-
BLOOD MUST SOAK RIGHT THROUGH PAPER



NSW NEWBORN SCREENING PROGRAMME

For Phenylketonuria, Hypothyroidism, Galactosaemia, Cystic
Fibrosis etc
(see pamphlet "Test to Protect Your Baby")

Collect from ALL newborns after 72 hours (but not less than 48) or
on discharge, if sooner. If child is discharged under 48 hours,
arrangements must be made for specimen to be recollected after 72
hours.

1. Warm heel before blood collection.
2. Puncture clean **dry** heel with a sterile disposable lancet (Point <2.4mm). Wipe away first drop of blood.
3. Blood must soak right through paper filling all circles completely. Do NOT layer blood. Only fill spot from one side.
4. Allow spot to dry before mailing (4 hours).
5. Return completed card **without delay**

To: NSW Newborn Screening Programme

COMPLETELY FILL EACH CIRCLE -
BLOOD MUST SOAK RIGHT THROUGH PAPER

S&S 903TM # W-981

