
Epidemiology, quality control and consumer access in the medical marketplace: The changing landscape of human genetic technology regulation in Australia

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Advances in genetics, genetic therapeutics and the application of genetic technologies to many aspects of human life have challenged the capacity of regulatory authorities and legislative processes the world over. In Australia, developments in the "new genetics" prompted the government to initiate a major inquiry into the protection of human genetic information, resulting in the production and publication of Report 96, titled "Essentially Yours: The Protection of Human Genetic Information in Australia" in 2003. This article examines the recommendations set out in this report and how they have provided Australia with a framework to deal with the advances in human genetic technologies, using the examples of direct-to-consumer personal genome testing and whole-genome sequencing.

INTRODUCTION

Since 2000, many countries and their regulatory bodies have struggled with the "new genetics". Advances in genetics, genetic therapeutics and the application of genetic technologies to many aspects of human life have challenged the capacity of regulatory authorities and legislative processes the world over. Different countries have taken very different approaches. In Australia, the government initiated a major inquiry into the protection of human genetic information in 2001, led by the Australian Law Reform Commission (ALRC), in association with the Australian Health Ethics Committee (AHEC) of the National Health and Medical Research Council (NHMRC). The project resulted in the production and publication of Report 96, titled *Essentially Yours: The Protection of Human Genetic Information in Australia*¹ in May 2003. This report was, and still is, the only one of its kind internationally, developed over two years, following extensive research and community consultation. It remains the most comprehensive consideration of the ethical, legal and social implications of the "new genetics" ever conducted.²

From the beginning, the inquiry acknowledged the need for public engagement and widespread consultation with the general community. To that end, it released an Issues Paper³ and a Discussion Paper⁴ to promote public education and debate, conducted 15 open forums around Australia and hosted over 200 meetings with interested parties around Australia and overseas and, in response, received over 300 written submissions.⁵

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¹ Australian Law Reform Commission (ALRC), *Essentially Yours: The Protection of Human Genetic Information in Australia* (Commonwealth of Australia, 2003).

² ALRC, n 1, p 33.

³ Australian Law Reform Commission, *Protection of Human Genetic Information, Issues Paper 26* (October 2001).

⁴ Australian Law Reform Commission, *Protection of Human Genetic Information, Discussion Paper 66* (August 2002).

⁵ ALRC, n 1, p 33.

The final report, *Essentially Yours*, made recommendations for reform, intended to deal with existing and emergent issues involving the “new genetics”.⁶ The recommendations were aimed at 31 different interest groups associated with the implementation, regulation and use of these technologies, including federal, State and Territory governments; health and medical policy-makers (such as the NHMRC); human rights, anti-discrimination and privacy officials; regulatory authorities (such as the Therapeutic Goods Administration); insurers; employers and trade unions; medical practitioners; universities; and professional education providers.⁷

The inquiry found extensive evidence of a central conflict between public support for breakthroughs promising better medical diagnoses and treatments and assistance with law enforcement (including the identification of missing or deceased persons), on the one hand, and anxieties about increased loss of privacy, the potential for genetic discrimination, and the capacity to regulate genetic science, on the other. The inquiry sought to find a middle ground that fosters innovation in genetic research and practice that serves humanitarian ends but provides sufficient reassurance to the community that such innovations would be subject to proper ethical scrutiny and legal (and other) controls. A cornerstone of this approach involved the report’s central recommendation: the establishment, by statute, of an independent Human Genetics Commission of Australia. In May 2005, as part of the annual budget process, the Australian Government announced that it had accepted this key recommendation and established an independent principal committee of the NHMRC, the Human Genetics Advisory Committee.⁸ This advisory committee has been in operation since 2006, with its third triennium term as one of the main NHMRC committees finishing in 2015.

In December 2005, a full government response to *Essentially Yours* was undertaken and approximately 90% of the recommendations were accepted.⁹ These steps by the government placed Australia at the forefront of regulation of developments in the field of genetics and human genetics and in a position to anticipate further developments in genetic sciences. The most significant development since the report has, arguably, been the rise of direct-to-consumer personal genome testing since 2007 and the rapid rise of whole-genome sequencing in research and (possibly) in the clinic.

THE EMERGENCE OF DIRECT-TO-CONSUMER PERSONAL GENOME TESTING

Direct-to-consumer personal genome testing (also referred to as direct-to-consumer genetic testing) became a global phenomenon in 2006-2007¹⁰ following the launch of the companies 23andMe, Navigenics Inc and deCODE Genetics. These companies were the first of their kind, offering directly to consumers a personal genome profile for predictive health assessments, and, through some companies, ethnic ancestry testing – all based on a genetic sample submitted to the company by a consumer. There are several definitions of “direct-to-consumer” in the current literature. In relation to genetic testing, however, there are two prevalent models of accessing this technology. In the first model, the availability of a test is advertised to the public, but the test must be ordered by, and the results delivered to, a health care provider.¹¹ In the second model, genetic tests are not only advertised directly to consumers, but the purchase of genetic testing services is also initiated at the consumer’s request, and the results are delivered directly to the consumer, without the involvement of the

⁶ ALRC, n 1, p 33.

⁷ Weisbrot D, “The Ethical, Legal and Social Implications of Umbilical Cord Blood Banking: Learning Important Lessons from the Protection of Human Genetic Information” (2012) 19 JLM 525.

⁸ See Department of Health and Aging, *Leading Australia’s Health into the Future* (10 May 2005), <http://www.health.gov.au/internet/budget/publishing.nsf/content/health-budget2005-hbudget-hfact6.htm> viewed 1 February 2012.

⁹ Australian Government, Australian Law Reform Commission, *Full Australian Government Response to ALRC Report 96*, <http://www.alrc.gov.au/inquiries/health-and-genetics/full-australian-government-response-alrc-report-96> viewed 31 January 2012.

¹⁰ Borry P, Cornel MC and Howard HC, “Where Are You Going. Where Have You Been: A Recent History of the Direct-to-Consumer Genetic Testing Market” (2010) 1(3) J Community Health 101.

¹¹ Holgarth S, Javitt G and Melzer D, “The Current Landscape for Direct-to-Consumer Genetic Testing: Legal, Ethical and Policy Issues” (2008) 9 Ann Rev Genomics Hum Genet 161.

consumer's health care provider.¹² In this latter model, the test may be ordered by a physician hired by the testing company, in which case there is no doctor-patient relationship with the consumer, but for other purposes, the test can be classified as having been "ordered" by a physician.

The form and content of the test results vary from company to company. Many companies will return results via an email notification, inviting the consumer to log onto the company's secure website using a username and password selected by the consumer. Once logged into the website, the consumer is able to access both their data and online educational resources to explore their data. The information a consumer receives from a company after they have submitted their DNA sample is, of course, generally not the raw data (although it is possible to access and/or purchase this as well). Instead, consumers receive a report that provides a probability assessment for a number of diseases based on the presence or absence of particular combinations of gene polymorphisms, their carrier status for certain genetic conditions, and/or information about their ancestry or ethnicity. The results provided by direct-to-consumer personal genome testing can therefore be critiqued in terms of their analytic validity and their clinical or predictive validity. Analytic validity refers to the accuracy with which a particular genetic characteristic can be identified in a given laboratory test.¹³ Clinical validity describes the accuracy with which a test predicts a particular clinical outcome.¹⁴

For example, in Huntington's disease (a hereditary neurodegenerative disorder), the genetic marker for this condition on chromosome 4 can be tested for with high accuracy.¹⁵ This genetic mutation, in turn, is highly predictive that the person affected will develop Huntington's disease. Unfortunately, the test does not accurately predict the age of onset of the disease, the clinical symptoms the patient may experience and the degree of severity of those clinical symptoms. Testing for Huntington's disease may therefore predict that an individual will develop this disease with a high degree of certainty, but it cannot predict the impact it will have on the individual. Using genetic tests to predict whether an individual will develop other more common diseases, like diabetes mellitus, Parkinson's disease, ischemic heart disease, cancer or heart disease is much more problematic again, as these diseases generally result from the interplay of many (often poorly understood) genetic and environmental factors.

Furthermore, even when genes can be accurately identified, all they can provide is probability estimates, often described in terms of odds or risk ratios that an individual with the genetic profile will develop the disease state of interest. Importantly, these probability assessments, or risk predictions, are drawn from epidemiological data from "reference" populations and not from consideration of the individual's own "history". What this means for the consumer is that even if they submit the same sample to two different companies, where these companies use different reference populations, they could (potentially) receive differing results for the same condition investigated, a situation that is not uncommon.¹⁶ There are several other reasons for discrepancies between different companies' reporting of health risks: the inclusion/exclusion criteria each company uses when deciding which genetic markers or literature to use in their analysis can differ, and the methodologies used in the analysis of the literature combined with how the risks are calculated can also differ, all leading to slightly different

¹² Holgarth et al, n 11.

¹³ Cecile A et al, "Evaluation of Predictive Genetic Tests for Common Diseases: Bridging Epidemiological, Clinical and Public Health Measures" in Khoury MJ et al (eds), *Human Genome Epidemiology* (2nd ed, Oxford University Press, Oxford, 2010); Goold I, et al, "Quality and Safety of Genetic Testing in Australia and New Zealand: A Review of the Current Regulatory Framework" (2006) 3(1) Aust New Zealand Health Policy 13; Haddow JE and Palomaki GE, "ACCE: A Model Process for Evaluating Data on Emerging Genetic Tests" in Khoury MJ et al (eds), *Human Genome Epidemiology: A Scientific Foundation For Using Genetic Information to Improve Health and Prevent Disease* (Oxford University Press, New York, 2004); Burke W et al, "Genetic Test Evaluation: Information Needs of Clinicians, Policymakers and the Public" (2002) 156(4) Am J Epidemiol 311.

¹⁴ Cecile et al, n 13; Goold et al, n 13; Haddow and Palomaki, n 13; and Burke et al, n 13.

¹⁵ Walker FO, "Huntington's Disease" (2007) 369(9557) *The Lancet* 218.

¹⁶ United States Government Accountability Office, *Testimony of Gregory Kutz Before the Subcommittee on Oversight and Investigations, Committee on Energy and Commerce, House of Representatives: Direct-to-Consumer Genetic Tests – Misleading Test Results are Further Complicated by Deceptive Marketing and Other Questionable Practices* (United States Government Accountability Office, 2010); Ng PC et al, "An Agenda for Personalized Medicine" (2009) 461(7265) *Nature* 724.

results. Regardless of the causes of the differences, the information a consumer receives is imbued with several levels of interpretation, well before the consumer reviews and attempts to understand this information in relation to her or his own life.

GROWING REGULATORY CONCERNS REGARDING DIRECT-TO-CONSUMER PERSONAL GENOME TESTING

There is limited empirical research into the uptake and interest in direct-to-consumer personal genome testing. One study, conducted in 2009 on social networkers, found that 64% would consider using such testing, primarily for the purpose of obtaining useful health information.¹⁷ There are no published data on the usage of direct-to-consumer personal genome testing in Australia, although research is currently underway. In contrast, there has been extensive academic critique of testing since 2007, particularly regarding the type of information the tests purport to offer consumers, how consumers endeavour to understand this information, how this information is used by consumers and whether or not this information is, or should be, subject to regulation. Much of this commentary has focused primarily on the value of the information a consumer receives, particularly in terms of:¹⁸

- analytical validity – its “ability to accurately and reliably measure the genotype of interest”;¹⁹
- clinical validity – “the ability of the test to detect or predict the associated disorder – components of clinical validity include the test’s sensitivity, specificity, and positive and negative predictive value”;²⁰ and
- clinical utility – the balance of its associated risks and benefits if it were to be introduced into clinical practice.²¹

These three concerns about the value of the genetic information a consumer receives are the key areas regulatory bodies have concentrated on, as this is the level where it is hoped, and expected, that regulation will occur. The philosophical, bioethical and social science literature raises additional questions regarding the validity of genetic information, but these areas of scholarship are most focused on how individuals, groups and society understand and attach meaning to the genetic information received through direct-to-consumer personal genome testing and the ways in which this information can enable or disable certain value systems. Others have examined how genetic information may determine the moral obligations that individuals and groups have, the moral claims that individuals and groups can then make on one another and how this information may challenge or undermine existing cultural structures of authority and order.

UNITED STATES RESPONSES TO DIRECT-TO-CONSUMER PERSONAL GENOME TESTING

Since their entry into the marketplace in the United States, companies offering direct-to-consumer personal genome testing have been operating on the assumption that sooner or later the government would take an interest in their practices. In April 2010, a report was issued by the Secretary’s Advisory Committee on Genetics, Health and Society (SACGHS) titled *Direct-to-Consumer Genetic Testing*. The SACGHS was formed in 2002 by the Secretary of Health and Human Services as a public forum for deliberation on the broad range of policy issues raised by the development and use of genetic tests and to provide advice on these issues.²² In its report, the committee outlined steps that

¹⁷ McGuire AL et al, “Social Networkers’ Attitudes Towards Direct-to-Consumer Personal Genome Testing” (2009) 9(6-7) Am J Bioeth 3.

¹⁸ Hunter DJ, Khoury MJ and Drazen JM, “Letting the Genome Out of the Bottle – Will We Get Our Wish?” (2008) 358(2) NEJM 105.

¹⁹ Cecile et al, n 13; Goold et al, n 13; Haddow and Palomaki, n 13; and Burke et al, n 13.

²⁰ Cecile et al, n 13; Goold et al, n 13; Haddow and Palomaki, n 13; and Burke et al, n 13.

²¹ Cecile et al, n 13; Goold et al, n 13; Haddow and Palomaki, n 13; and Burke et al, n 13.

²² Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS), *Direct-to-Consumer Genetic Testing* (Department of Health and Human Services, 2010), http://oba.od.nih.gov/oba/SACGHS/reports/SACGHS_DTC_report_2010.pdf viewed 1 February 2012.

federal and State levels of government could take in regard to direct-to-consumer genetic tests, specifically to address the gaps and inconsistencies in federal regulations and to accelerate the coordination of programs that facilitate comprehensive and consistent consumer and health provider genetics education.²³ The SACGHS report raised particular concerns regarding:²⁴

- gaps in federal oversight of direct-to-consumer genetic testing, particularly the absence of review of claims and promotional materials by the Food and Drug Administration (FDA) due to existing policies regarding pre-market review and limitations under current regulatory practices;
- lack of evidence of clinical validity and/or clinical utility for most direct-to-consumer genetic tests;
- gaps in privacy and research protections for consumers utilising direct-to-consumer genetic services because federal regulations may not apply to companies offering such testing and State-level protections may be inadequate; and
- insufficient knowledge about genetics among many consumers, the limited involvement of health care providers to assist consumers in selecting genetic tests and in making health decisions based on the test results, and/or inadequate training for health care providers whose patients request information regarding test selection or the interpretation of test results.

While these recommendations were not explicitly acted upon, they did inform further inquiries throughout 2010. In May 2010 a leading pharmacy company, Walgreens, and the direct-to-consumer personal genome testing company Pathways Genomics announced their intention to market direct-to-consumer personal genome testing kits to consumers in Walgreens stores across America.²⁵ In response, the FDA issued a letter to Walgreens and Pathways Genomics, informing them that the kit they were attempting to market fell under the description of a medical device and as such, was subject to pre-market review by the FDA. The subsequent launch of the product was cancelled.

Then, in June 2010, additional letters were sent to the remaining companies offering direct-to-consumer personal genome testing (Knome Inc, 23andMe Inc, deCODE Genetics, Navigenics and Illumina Inc²⁶), bringing to their attention the fact that they had not lodged a pre-market approval application nor had they submitted information on the analytical or clinical validity of their tests to the FDA for clearance or approval.²⁷

Finally, on 22 July 2010 the Subcommittee on Oversight and Investigations of the United States House of Representatives interrogated three of the four major direct-to-consumer personal genome testing companies currently doing business – 23andMe Inc, Pathways Genomics and Navigenics Inc. Much of this interrogation centred on the results of an investigation by the Government Accountability Office (GAO) into direct-to-consumer personal genome testing. The concerns of the regulators can be summarised by an excerpt from the expert testimony provided by Dr James Evans (an expert in medical genetics asked to testify at the hearings and lead scientist consulted by the GAO for its report):²⁸

[W]e should encourage individuals to be involved in, and be the primary directors of their own healthcare ... but, it is also critical that they be assured that the information they receive is of high quality, that they have recourse to disinterested advice about the meaning of that information, that their privacy be protected and that claims made by the purveyors of such testing comport with reality.

²³ SACGHS, n 22.

²⁴ SACGHS, n 22.

²⁵ Genomeweb, *Walgreens to Sell Pathway Genomics' Sample Kit* (11 May 2010), <http://www.genomeweb.com/dxpgx/walgreens-sell-pathway-genomics-sample-collection-kit> viewed 2 February 2012.

²⁶ Illumina Inc is not a direct-to-consumer personal genome testing company but a biotechnology firm that manufactures the Illumina® Infinium HumanHap550 array used by deCODE Genetics and 23andMe.

²⁷ Food and Drug Administration (FDA), *Medical Devices – Letters to Industry*, <http://www.fda.gov/MedicalDevices/ResourcesforYou/Industry/ucm111104.htm> viewed 6 February 2012.

²⁸ United States House of Representatives, Committee on Energy and Commerce, *Hearing on Direct-To-Consumer Genetic Testing and the Consequences to the Public Health*, “Testimony of James Evans” (22 July 2010), <http://democrats.energycommerce.house.gov/index.php?q=hearing/hearing-on-direct-to-consumer-genetic-testing-and-the-consequences-to-the-public-health> viewed 1 February 2012.

In other words, the primary question concerned what the information provided by direct-to-consumer personal genome testing actually “means”, both in terms of its statistical and clinical properties – its accuracy and validity – and in terms of how it is understood and interpreted by the consumer. This concern was highlighted by Dr Jeffery Shuren, then director of the FDA, in his testimony:²⁹

None of the genetic tests now offered directly to consumers has undergone premarket review by the FDA to ensure that the test results being provided to patients are accurate, reliable and clinically meaningful.

As Dr James Evans stated in his testimony: “[F]inding out that you’re at double or half the ‘average’ risk of a common disease is simply not medically meaningful.”³⁰ This type of concern was, of course, not new, and echoed criticisms that had repeatedly been made about direct-to-consumer personal genome testing.³¹ They reflected growing unease over the absence of regulation of such testing in America.

Additional discussions in the United States around how direct-to-consumer personal genome testing should be regulated have centred on the classification of the testing kits. Two days prior to the House of Representatives hearings in July 2010, there was a two-day open forum hosted by the FDA and Centre for Devices and Radiological Health on the topic of laboratory developed tests.³² This meeting, which included a session on direct-to-consumer genetic testing, focused on whether such tests should be classified as medical devices, in vitro diagnostic devices or as laboratory-developed tests. This is an important question, as each type of classification would entail a different approach to regulation and thus requires different standards of evidence and levels of review before the tests would conform to the appropriate regulations. In March 2011, the Molecular and Clinical Genetics Panel of the Department of Health and Human Services hosted a panel to develop recommendations regarding the scientific assessment of direct-to-consumer genetic tests.³³ As of January 2013, no further details regarding this initiative have been announced.

AUSTRALIAN RESPONSES TO DIRECT-TO-CONSUMER PERSONAL GENOME TESTING

In Australia, critical commentary has been more limited. In August 2008, the NHMRC hosted a national meeting to discuss direct-to-consumer genetic testing. Key findings from this meeting focused on the validity of genetic tests, the effect of test results on consumers, the ownership of DNA, and the balance to be found between the need to protect consumers and the need to avoid over-regulation and inadvertent negative impacts on research and clinical practice.³⁴ Importantly, the meeting made reference to the fact that the ALRC’s report, *Essentially Yours*, foreshadowed the development of direct-to-consumer personal genome testing and offered some suggestion regarding its regulation in

²⁹ United States House of Representatives, Committee on Energy and Commerce, n 28, “Testimony of Jeff Shuren” (22 July 2010).

³⁰ United States House of Representatives, Committee on Energy and Commerce, n 28, “Testimony of James Evans” (22 July 2010).

³¹ Cecile et al, n 13; Samuel GN, Jordens CFC and Kerridge I, “Direct to Consumer Personal Genome Testing: Ethical and Regulatory Issues that Arise from Wanting to ‘Know Yourself’” (2010) 40(3) *Inter Med J* 220; Kuehn B, “Risks and Benefits of Direct-to-Consumer Genetic Testing Remain Unclear” (2008) 300(13) *JAMA* 1503; Hunter et al, n 18; Hudson K et al, “American Society of Human Genetics Statement on Direct-to-Consumer Genetic Testing in the United States” (2007) 81(3) *Am J Hum Genet* 635; Haga SB, Khoury MJ and Burke W, “Genomic Profiling to Promote a Healthy Lifestyle: Not Ready for Primetime” (2011) 34(4) *Nat Rev Genet* 347; Burke et al, n 13.

³² FDA, *FDA/CDRH Public Meeting: Oversight of Laboratory Developed Tests (LDTs)* (17 September 2010), <http://www.fda.gov/MedicalDevices/NewsEvents/WorkshopsConferences/ucm212830.htm> viewed 5 February 2012.

³³ FDA, *Molecular and Clinical Genetics Meeting Announcement* (FDA, 2011), <http://www.fda.gov/AdvisoryCommittees/CommitteesMeetingMaterials/MedicalDevices/MedicalDevicesAdvisoryCommittee/MolecularandClinicalGeneticsPanel/ucm245447.htm> viewed 3 February 2013.

³⁴ NHMRC, *Direct-to-Consumer Genetic Testing* (NHMRC, 2008), <http://www.nhmrc.gov.au/media/events/2008/direct-consumer-genetic-testing> viewed 4 February 2012.

Australia. Of particular note was recommendation 11-5 which stated:³⁵

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.

The NHMRC meeting also noted the Federal Government's response to this recommendation:³⁶

A working group, the National Coordinating Committee of Therapeutic Goods In Vitro Diagnostic Devices Working Group, established under the Australian Health Ministers' Advisory Council (AHMAC) has examined these issues. The working group has recommended that all health related in vitro diagnostic devices intended for home use (that is, tests not carried out under the supervision of a health care provider) should be regulated in accordance with the risk class of the test, taking into account particularly the aspects relating to the use of the test by the lay person. In October 2003, the matter was referred to AHMAC for consideration and AHMAC endorsed the banning of all health related home use genetic tests.

In response, in June 2010, the Therapeutic Goods Administration announced changes to the specification as to what constitutes a self-test, amending s 4.2.B of the *Therapeutic Goods (Excluded Purposes) Specification 2010* (Cth), such that if the medical device is intended for "genetic testing to determine the presence of, or predict susceptibility to, diseases in humans", then the device is excluded from listing and registration on the Australian Register of Therapeutic Goods (ARTG) – which is required for therapeutic goods to be lawfully supplied in Australia.³⁷ This specification change was followed by the commencement of a new regulatory framework from 1 July 2010 which ensures that all IVDs (in vitro diagnostic) medical devices undergo a level of regulatory scrutiny commensurate with the risks associated with their use.³⁸ This regulatory framework is particularly relevant to direct-to-consumer personal genome testing, as the specifications for access to IVDs for self-testing (home use) provide:³⁹

IVDs intended for self-testing are tests that are used in the home or a similar environment and are not carried out under the supervision of a health care provider. Certain types of self-testing IVDs will be prohibited from supply. These include:

- IVDs used to test for pathogens or diagnose notifiable infectious diseases;
- Tests to determine genetic traits;
- IVDs used to test for serious disorders, for example cancer or myocardial infarction;

It is arguable, therefore, that the tests provided by companies offering direct-to-consumer personal genome testing do not qualify for registration on the ARTG and thus may be prohibited. This view was reinforced by the subsequent revision (November 2011), *Classification of IVD Medical Devices* by the TGA, which states under Classification Rule 1.4 – IVD medical devices for self-testing:⁴⁰

[S]elf-testing genetic tests and self-testing IVDs used to test for serious disorders (such as cancer or myocardial infarction) are excluded from being entered on the ARTG.

This specification is important, because it indicates clearly that genetic tests and self-testing IVDs for health disorders are not listed on the ARTG. It is also clear that genetic tests for parentage and kinship testing are exempt from TGA regulation in this same framework.⁴¹

³⁵ ALRC, n 1, p 58.

³⁶ Australian Government, Australian Law Reform Commission, n 9.

³⁷ Therapeutic Goods Administration, *Australian Register of Therapeutic Goods* (3 May 2012), <http://www.tga.gov.au/industry/artg.htm> viewed 31 January 2012.

³⁸ Therapeutic Goods Administration, *Overview of the Regulatory Framework for In-vitro Diagnostic Medical Devices* (6 July 2011), <http://www.tga.gov.au/industry/ivd-framework-overview.htm> viewed 29 January 2012.

³⁹ Therapeutic Goods Administration, n 38.

⁴⁰ Therapeutic Goods Administration, *Classification of IVD Medical Devices* (Version 1.1, Department of Health and Ageing, 2011).

⁴¹ Therapeutic Goods Administration, n 39.

Despite this, it remains the case that while attempts have been made by regulatory agencies in Australia to streamline policy and construct a system that effectively prohibits “at-home” genetic self-testing, the regulatory landscape remains much the same as when *Essentially Yours* was commissioned:⁴²

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 complaint-handling authorities; private interest; and market pressure.

The status of “at-home” genetic tests in Australia is therefore precarious, at best. While changes during 2010-2011 suggest that “at-home” genetic tests may be prohibited, their online availability and the difficulty of regulating the internet means that Australians can order and obtain such tests without restriction.

THE RISE OF WHOLE-GENOME SEQUENCING

Since the completion of the Human Genome project, the next holy grail in sequencing technology has been the pursuit of a fast and affordable whole-genome sequence – the “\$1000 genome”. Advances in next-generation sequencing and dramatic reductions in the cost of sequencing and the time taken to sequence an entire genome mean that whole-genome sequencing is now becoming a reality and medicine is now moving inexorably towards comprehensive genomic medicine.

A recent article published by the National Institutes of Health’s division of genetic research, the National Human Genome Research Institute,⁴³ outlined a strategic plan for the future of genomics and medicine which makes clear the central significance of whole-genome sequencing. Key to this plan is the promotion and support for genomic medicine, which involves:

- making genomics-based diagnostics routine;
- defining the genetic components of disease;
- comprehensively characterising cancer genomes;
- developing practical systems for clinical genomic informatics; and
- elaborating the role of human micro-biomes in health and disease.

These same issues are apparent in public and professional discourses surrounding whole-genome sequencing in Australia. In May 2011, at a Whole Genome Sequencing Workshop hosted by the NHMRC stakeholder groups noted the broad impact that whole-genome sequencing may have on medicine, public health and society more generally and raised questions regarding the following:⁴⁴

- how the clinical indications for whole-genome sequencing would be defined and how whole-genome sequencing would be applied in the clinic;
- what steps would need to be taken to ensure that the next generation of medical professionals had sufficient undergraduate and professional expertise to be able to employ genetic technologies, such as whole-genome sequencing, wisely and in the public interest;
- what legislative, regulatory and policy strategies would need to be put in place to ensure equity of access to whole-genome sequencing such that its benefits are available to, and experienced by, all communities;
- whether policy needs to be informed by a better understanding of the issues involved in whole-genome sequencing and if so, that there was an urgent need to conduct a “pilot study” to assess the clinical utility of whole-genome sequencing; to identify infrastructural issues that need to be addressed for health purposes and broader applications; to identify mechanisms for knowledge, development and translation for capacity building; to inform the development of

⁴² Australian Government, Australian Law Reform Commission, n 9.

⁴³ Green ED and Guyer MS, “Charting a Course for Genomic Medicine from Base Pairs to Bedside” (2011) 470(7333) *Nature* 204.

⁴⁴ National Health and Medical Research Council (NHMRC), *NHMRC Whole Genome Sequencing Workshop Summary* (Canberra, 2011).

laboratory (processing) nodes across Australia; and to inform the development of tools, models of care, and linkages between research and service activities;

- what systems and structures are required for the safe and effective storage of whole-genome sequencing data – and how whole-genome sequencing would impact upon the development of personally controlled electronic health records.

In both the United States and Australia, regulation will undoubtedly develop in response to progress in human genetics and biotechnology, while also promoting and inhibiting this progress. Australia is uniquely positioned to not only lead the world in its approach to the regulation of genetic technologies but also in its approach to public engagement and consultation around what stakeholders need and expect from regulatory and legislative authorities in response to new and emerging human genetic technologies.

REGULATION, PUBLIC DISCOURSE AND THE FUTURE OF GENOMIC MEDICINE IN AUSTRALIA

Over the next decade, genetics will increasingly pervade every aspect of life, from the “clinical” to the social world – describing an individual’s future health, relationships and identity. While these changes will undoubtedly create policy, regulatory and public concerns, Australia’s experience with *Essentially Yours* provides the basis for further policy development.⁴⁵ This report began a broad dialogue with the public, industry, regulatory and research communities regarding genetics and the potential of genetics to change basic understandings about human life and social order; and it explicitly acknowledged that political, social and legal reform is needed to accommodate continuing developments in genetics and biotechnology.

The *Essentially Yours* recommendations set in motion a regulatory and discursive framework that could, with further elaboration, respond to the rise of direct-to-consumer personal genome testing and whole-genome sequencing. This framework enables the identification of what is important or “valued” and the elaboration of mechanisms for dealing with both ethical conflict and unforeseen developments in genomic science. The framework outlined by *Essentially Yours* will undoubtedly be challenged by what whole-genome sequencing offers and delivers, as this technology both reinstates “old” problems and poses new ones. The sheer volume of information and the expansion of ways in which information can be delivered and understood will challenge the very foundations of clinical practice and public health. This alone will raise questions – again – about the status and privacy of genetic information and about whether genetic-specific legislation is necessary to control the “new genetics”.

CONCLUSION

The discourse begun by the *Essentially Yours* report provides a sound basis for regulatory exploration; however, the regulatory framework requires further debate and is ultimately only as good as its application. While mechanisms are in place to assist in identifying what regulations could be expanded or refined in order for the genetic tests provided by direct-to-consumer personal genome testing to be appropriately regulated, we (collectively) are yet to have a comprehensive public discourse about the analytic and clinical validity and the clinical utility of the genetic information that consumers receive from direct-to-consumer personal genome testing or whole-genome sequencing. Given the vast amounts of information that direct-to-consumer personal genome testing and whole-genome sequencing will generate and provide, these questions are both timely and pertinent to ensuring that such services are ethically sound and work in the public interest. Inevitably, advances in the fields of genetics and human genetics will outpace public understandings of human identity and disease and challenge regulatory structures put in place to accommodate them. Despite this, the experience of *Essentially Yours* places Australia in a unique position, one where ethics, science and law could be brought together to shape the future regulatory landscape of human genetic technologies in Australia.

⁴⁵ ALRC, n 1.