LEGAL ISSUES RELATING TO NEWBORN SCREENING

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1 INTRODUCTION

The New Zealand Law Foundation-sponsored Human Genome Research Project is a multi-disciplinary and international project. It examines issues raised by developments and advances in human genome-based technologies and knowledge; the responsible use of such technologies and knowledge to improve the health and well-being of New Zealanders; and inter-relationships between genetic knowledge, medical sciences, clinical practices, the humanities, government regulation and policies and the law. In the second year of the project, part of the research work involves looking into new and emerging initiatives and applications in genetics for newborns. The current New Zealand newborn metabolic screening programme is chosen as the starting point for a number of reasons.

Firstly, the newborn screening programme provides a useful case study for examining the relationship between advances in knowledge and technology that stem from the sequencing of the human genome and the need or desirability for public policy to catch up.[1] As an intervention to improve population health, the programme has very wide coverage in terms of participation numbers and has been running for a long time.[2] Ongoing experiences with the programme provide a rich context for analysing emerging ethical, regulatory and legal questions raised by changing expectations, new frameworks and law reform, and also for investigating future policy options.

Secondly, the current newborn screening programme measures changes in the levels of blood compounds, for example enzymes, amino acids, hormones and metabolites, to detect the possibility of early onset genetic disorders. In effect, the programme involves an indirect method of genetic screening. Close study of some of the issues that emerge will be helpful in terms of thinking ahead about future genetic screening programmes.[3] Related to this are questions about the changing paradigms that will be driven by advances in genome-based knowledge and which will have implications for public health.

Thirdly, as part of an examination of relevant new technologies, our attention has been drawn to the two novel technologies of tandem mass spectrometry and DNA microarray, which are revealing a range of applications in areas that include newborn screening. While the former was introduced to the newborn screening programme in December 2006, and provides a basis for inferring the presence or absence of the genetic metabolic condition in question, emerging knowledge about the latter seems to point to exciting potential applications with the ability to test for DNA directly.

Fourthly, the dried blood spots collected on Guthrie cards that are currently stored indefinitely represent an inchoate 'DNA bank'.[4] The collection of samples was estimated in 2003 to be in the order of 1.9 million[5] and to date potentially comprises the DNA profile of almost every person in New Zealand born since blood spot

samples were taken in the late 1960s or early 1970s. The collection of samples[6] has been described by a parliamentarian[7] as a 'virtual databank'. With relevant legislative proposals having been introduced,[8] now seems to be a good time to revisit and clarify the justification for, and policies regarding, the retaining of the samples after the central aims of the programme have been fulfilled.

Fifthly, it came to the notice of the Project during its early stages that the Government of the United Kingdom had referred to the possibility in the long term of screening babies at birth as part of the standard postnatal checks, and of producing a comprehensive map of their key genetic markers or entire genome that would be stored on their electronic patient record.[9] The intended purpose of such genetic profiling at birth would be for tailoring prevention and treatment regimes to their needs throughout their lifetime, using increasingly available knowledge about how genes affect risk of disease and response to medicines. It is our view that we should be ready to participate in debates about the issues when they start to take shape. It is timely to begin looking into future technologies and emerging knowledge, and to think ahead about regulatory options relating to the existing newborn screening programme around which any such proposal is likely to be based.

Central to the examination of the legal issues relating to newborn screening are the findings made by two Commissioners. The first set of findings arose from the investigation of the Health and Disability Commissioner into a complaint relating to *informed consent* for neonatal blood tests.[10] The second was the result of the inquiry of the Privacy Commissioner into the *collection, retention, use and release* of newborn metabolic screening test samples.[11]

Events surrounding the involvement of both Commissioners had their genesis in *H v G*[12] in which an application was made to the Court for the release of a deceased baby's dried blood spot sample card, retained by the newborn screening programme, for the purpose of DNA analysis to prove paternity. The baby's mother opposed the application, claiming that it would involve using the sample *for a purpose other than the original intent for which it had been obtained.* Salmon J ordered the release of the sample and it was tested. On appeal[13] by the baby's mother, Morris J held that the analysis of the sample had been undertaken under an order of the Court and the results of the DNA identification were admissible as evidence.

The Court decision of *H v G* was given in 1999 and the recommendations from the Privacy Commissioner were provided in 2003. Since then, there has been a considerable number of overseas studies and developments in scientific knowledge; much research into parental perceptions; and new policy proposals and State initiatives. In New Zealand, although progress appears to have been slow, some movement on a number of initiatives is discernible.[14]

In our view, the findings of the High Court and the Commissioners taken together draw attention to issues about the newborn screening programme that can be separately conceptualised in terms of:

1. the collection, use and storage of dried blood spots *for fulfilling the aims* of the newborn screening programme; and
2. the storage and use of dried blood spots for purposes *beyond fulfilling the aims* of the newborn screening programme.

This report will discuss the involvement of parents in terms of 'participation' in the newborn screening programme and then examine issues relating to (1). Issues relating to (2) will be discussed in a separate, subsequent report.

2 PARTICIPATION IN THE NEWBORN SCREENING PROGRAMME

A broad view is taken in this report of the involvement of parents and the responsibilities of health-care providers with regard to newborn screening within the New Zealand legal and ethical framework. The expression 'participation' is used here to give recognition to an increase in shared decision-making between health professionals and parents in order to protect and promote the interest of newborns, and to emphasise the importance of enlisting the support of parents who are invited to take part in the programme so that blood samples from their newborns can be obtained for screening.

Standard approaches adopted when analysing or framing medico-legal issues typically begin with or inevitably converge on the issue of informed consent. The Privacy Commissioner and Health and Disability Commissioner clearly communicated the need for informed consent. This is reflected in the language used in their reports and in the formulation of their findings and recommendations. The approach taken in this report reinforces that of the commissioners by emphasising the importance of informed consent for voluntary participation in newborn screening.

Examination of the issues makes more explicit the context and the supporting elements in which informed consent is to be meaningfully sought by health-care providers and granted by parents or legal guardians in the circumstance of participation in a screening programme. The analysis is grounded in the rights-based Code of Health and Disability Services Consumers' Rights 1996 ('Code of Rights'), and takes into account both the public health paradigm that justifies population-based interventions for securing overall beneficial outcomes and the striking aspects of dealing with genetic risks in families.

Screening has to be distinguished from personal clinical services. Clinical services are provided to individuals who seek medical help because they suspect or have symptoms of a problem. In contrast, screening is offered to a defined population, such as women in a specific age group for breast or cervical screening, or, in the case of the newborn metabolic screening programme, all newborns. Parents[15] are approached to identify individuals who could be helped by further tests or treatment to reduce the risk of a disease or its complications.[16]

Underpinning the nature and scope of a population-based intervention such as the newborn screening programme is the framing of regulatory and policy mechanisms to give weight to maximising participation from members of the target population. However, while measures with legal force used in traditional public health contexts were prescriptive in form and, in effect, compelled participation, they are now written in enabling terms based on patient or consumer rights and actively promote informed choice and the obtaining of consent. This shift in public health is consonant with approaches in the context of genetic medicine, extending beyond prevention, treatment and care to incorporate future reproductive decisions relevant to the family.

3 THE CODE OF RIGHTS AND THE NEWBORN SCREENING PROGRAMME

Informed consent in the New Zealand health-care context is formally and explicitly recognised in statute[17] and provides the central focus for patient and consumer rights and advocacy. Informed consent is specifically defined to mean consent freely given to any health-care procedure by or on behalf of a health consumer and obtained according to the Code of Health and Disability Consumers' Rights ('the Code').[18] The Code systematically sets out the rights of consumers and obligations placed on providers of health and disability services. Three elements are vital to successful participation in the newborn screening programme: *communication* between the health provider or professional and the consumer; availability of *adequate information* for the consumer; and the opportunity for the consumer to exercise *informed choice and consent*. These are encapsulated in rights 5, 6 and 7 of the Code respectively. Rights 5 and 6 are important, in that they represent the two key steps prior to the exercising of choice and consent set out in right 7.

3.1 Right 5: Right to effective communication

Right 5(1) of the Code provides that every consumer has the right to effective communication in a form, language and manner that enables the consumer to understand the information provided.[19] Right 5(2) provides that every consumer has the right to an environment that enables both the consumer and provider to

communicate openly, honestly and effectively. Right 5, in essence, requires a mode of communication that enables understanding. The matters raised here are different from the question of what particular information is given, which is more appropriately discussed later under right 6.

3.1.1 Timing and distribution of leaflets to parents

The references to communication in right 5, particularly that which relates to the appropriate *manner* and *environment* for communication to take place to enable understanding, raise questions about the opportunities available for consumers to ask questions, digest responses to questions and information and familiarise themselves or be acquainted with the facts. Crucial to these would be issues of timing, including those times at which opportunities are provided to facilitate this process; the length of time allocated for each of those opportunities; and the length time separating the taking of the baby's blood sample from the parents' considered decision regarding participation.[20] Empirical studies or quality assurance reports regarding these issues are not presently available for newborn screening in New Zealand.

In the complaint investigated by the Health and Disability Commissioner, the right to effective communication was not an issue. This was because the two relevant health professionals had not, for separate reasons, sought informed consent.[21] Of relevance to the issue of effective communication is the Privacy Commissioner's informal survey of eleven maternity hospitals relating to screening information contained in a leaflet *Your Newborn Baby's Blood Test* that is produced by the National Testing Centre. The Privacy Commissioner said the presentation of the leaflet to the mother 'at least provides a natural opportunity for the lead maternity carer ('LMC') to explain those matters in which the leaflet is inadequate in its explanation'. However, the Commissioner found that 'even the leaflet distribution appears to be unreliable'.[22]

Timing is a key factor in the distribution of the leaflets in order to initiate meaningful discussion about the conducting of the screening test. It is difficult to regard the circumstances following childbirth as providing the appropriate conditions and environment for this to happen. Elkin and Jones, who have also made this point, noted the findings of a study that found that when women were asked for consent just before the test, they tended to feel 'psychologically committed' to the test and so were less receptive to disclosed information.[23] Researchers for the STAR-G Project in the United States learned from their participants that the ideal time to educate parents about newborn screening is during the third trimester of the prenatal period.[24] Also, recently published findings from research involving focus groups comprising parents, providers and experts reported that all participants thought that parents should receive information in concise, easy-to-read brochures preferably in the third trimester of pregnancy.[25]

The draft *Interim Standards for the Newborn Metabolic Screening* is not currently publicly available.[26] A supplementary report by the Victorian Privacy Commissioner to the final report by the Victorian Newborn Screening Review Committee for the Minister of Health in August 2006 recommended that 'information should be conveyed in a manner that allows time for reflection and consideration of the implications … [and] should be provided early during prenatal care and/or during the third trimester'.[27] Likewise, the United Kingdom Newborn Screening Centre provided guidance in 2005 to this effect in their *Policies and Standards for Newborn Blood Spot Screening in the UK* where reference to provision of a pre-screening leaflet is made three times under 'Process Standards', 'Consent and Communication' and 'Blood Sampling Guidelines'.[28] One of the earliest references to the suggested provision of information about screening in the third trimester was made by the United States Task Force on Newborn Screening in 1999.[29] New Zealand would benefit from a requirement that information about screening be given to parents by or in the third trimester and from explicit statement of that requirement in current policies and standards, and also in relevant legal documents or arrangements relating to the provision of maternity services.[30]

3.2 Right 6: Right to be fully informed

Right 6 of the Code provides consumers with the right to be fully informed. Right 6(1) states that every consumer has the right to the information that a reasonable consumer, in that consumer's circumstances, would expect to receive. More specifically, right 6(1) states that the consumer is entitled to:

(a) an explanation of his or her condition;

(b) an explanation of the options available, including an assessment of the expected risks, side effects, benefits, and cost of each option;

(c) advice on the estimated time within which the services will be provided;

(d) notification of any proposed participation in teaching or research, including whether the research requires and has received ethical approval;

(e) any other information required by legal, professional, ethical and other relevant standards;

(f) the results of tests; and

(g) the results of procedures.

3.2.1 Right 6 and the context of screening

The language in right 6(1) appears to be drafted with a primary focus on situations in which a consumer has presented with symptoms or complications that need diagnosis and treatment. Expectations and standards for diagnosis and treatment in

the context of clinical services may vary from those in the context of screening. The caution voiced by Sandra Coney about the applicability of the Code to the context of screening needs to be borne in mind.[31] The list in right 6(1) is, however, illustrative and not exhaustive, and would thus, where appropriate, allow for adaptation or modification to the circumstances demanded by a particular screening programme.

The significance of a 'screening pathway' must be appreciated. The Women's Health Action articulated the following in the context of breast and cervical screening in July 2000:

> *There has been a tendency … to inform women just about each step at a time, to seek consent only [for] the screening test. Having a smear or a mammogram is just one step on the screening pathway. For women who are recalled, there will be other decisions to be made. It is a critical part of informed consent for women to know about the entire screening pathway and the possible things that might occur before they take the first step. This is a complex form of informed consent because it requires potential participants to be able to imagine themselves in situations that might occur before they are faced with them. This is important to prepare women for the possibility of an abnormal result and for recall. In research on screening programmes women have consistently said that they want more information at the time of the initial screening test about the meaning of an abnormal result and what will happen after that. (Emphasis added.)*[32]

In 2003, the National Health Committee (NHC) provided formal guidance emphasising that screening is not just about the initial test but also embraces a sequence of events that comprises the screening pathway.[33] The 'screening pathway', which is described as the screening process from a participant's perspective, includes:[34]

1. an invitation to be screened;
2. being given information about the purpose of the screening, the likelihood and possibility of false positive/negative results, the uncertainties and risks attached to the screening process, any significant medical, social or financial implications of screening for the particular condition or predisposition, follow up plans, including availability of counselling and support services;[35]
3. being questioned or offered a test;
4. having the test;
5. receiving of test results;
6. assessment and diagnosis if the test is positive;
7. possible treatment; and
8. understanding that there are activities to monitor and evaluate all these stages.

It can be seen therefore that screening involves a number of discrete steps, each of which is associated with particular obligations on the part of the provider, actions on the part of the consumer, and attention and commitment by both parties to see through the whole of the screening process. Each screening programme also has its unique aspects with its own target population and particular goals that will need to be reflected in the specific details of the screening pathway.

A set of overseas recommendations may be helpful at this juncture. In 2000, the American Academy of Pediatrics (AAP) Task Force on Newborn Screening published a 'blueprint' for the future of newborn screening to address concerns about variability among the newborn screening programmes in the United States, and to develop a strategy to resolve ongoing and emerging challenges.[36] Several key factors for successful screening were identified.[37] Most notably, the AAP provided seven recommendations regarding what should be present in parents' educational material:[38]

1. the benefits of screening;
2. the potential risks of the screening test;
3. how parents will be informed of screening results;
4. the possibility of a false-positive test result;
5. the importance of responding to a positive test result;
6. how to respond to a positive test result; and
7. the screening program's policy for sample storage and use of stored samples.

The list set out by the NHC and, where appropriate, the AAP recommendations can inform the application of right 6(1) in the newborn screening context and should be reflected in relevant material and sources, such as the leaflet distributed to parents, the National Metabolic Screening Programme ('NMSP') website or the interim standards.

Rights 6(1)(a) and (b) are predicated on clinical diagnosis and treatment for which the patient or consumer initiates the contact or therapeutic relationship. The NHC's recommended screening process (1), which is expressed in terms of an invitation to be screened, should be adopted and made more explicit when approaching potential screening participants. Highlighting the invitation for screening would emphasise the element of choice, and the voluntary nature of the process, and remove any hint of compulsion in relation to participation.

The matters spelt out at length in the NHC's screening process (2) can be incorporated within the scope of right 6(1)(e). This would, among other things, clearly and unambiguously emphasise that the purpose of participation in screening is the primary interest of the child. The leaflet, as currently worded, seems acceptable and

clear, although it could be strengthened with explicit reference to the 'interest' of the newborn.

3.2.2 Notification of results

Right 6(1)(f) and (g) and the NHC's screening process (5) raise the issue of notification of test results. The newborn screening programme has been operating on the basis that 'no news is good news' and the practice is presently communicated upfront in the leaflet given to parents. Arguably, this would satisfy the AAP's third recommendation regarding the way in which parents will be notified of the screening result, and how this procedure is expressed in parents' educational material (in this case, they will not be informed if the test result is negative). The 'no news is good news' practice has not been unique to New Zealand. However, overseas trends are shifting. In the 2005 United Kingdom Policies and Standards document, one of the principles under 'Consent and Communication' states that:

> *Parents have a right to information about their baby's screening result including: the reason for any repeat samples; and 'normal' as well as 'abnormal' screening results.*[39]

The Victorian Newborn Screening Review Committee in their report to the Minister of Health (August 2006) recommended '[t]he development of a result notification system for the newborn screening program'.[40] Additionally, the Committee's draft model information sheets for presentation to parents when seeking their consent provide that:

> *All parents are notified by mail of their baby's newborn screening results (if normal), or by telephone (if re-testing required).*[41]

Without any New Zealand survey or empirical data available, it is difficult to form a view as to whether the 'no news is good news' practice should continue or be changed so that parents are notified of their newborn screening test results. The Victorian Newborn Screening Review Committee recommendation appears to have been based on research commissioned by the Health Issues Centre that called for '[r]esults of the screening to be sent to all parents' and voiced the principle that '[p]arents have a right to information about their baby's screening result'.[42] The survey by the Centre found that:

> *A majority of consumers stated they would like to have some confirmation of the results sent to them. Several commented that the current practice of 'if you don't hear anything, it's OK' was not adequate.*[43]

For New Zealand, 'no news is good news' seems to have been a practice that has worked well to date. The practice is less contentious when general awareness and

knowledge of newborn screening is low or non-existent. However, if newborn screening increasingly operates within a paradigm that more actively promotes the exercise of informed choice and consent, 'no news is good news' could be regarded as falling short of the expectations of parents to be informed throughout the screening process, including being notified about the result of the screening test. An argument against changing the practice of 'no news is good news' is that most results will be normal because the disorders for which screening presently takes place are rare. Nevertheless, the future may demand a change when complex issues arise with technologies that reveal carrier status or if screening is extended to include late-onset disorders. It is important for surveys and empirical studies to be undertaken in New Zealand to inform policy in this area. Additionally, public discussion needs to take place, such as debate by parliamentarians in the chamber after public submissions on the issue.

3.2.3 Information about quality assurance activities

Activities to monitor and evaluate newborn screening raise interesting issues relating to the right to be fully informed under right 6(1) with regard to the NHC screening pathway, and also relating to rights 7(9) and 7(10) which will be discussed at 3.3.4. For brevity, activities to monitor and evaluate, as with the activities stated at right 7(10)(c), will be referred to as 'quality assurance' (QA) activities.

Right 6(1) does not explicitly mention QA-related activities but includes reference to the right to receive 'any other information required by legal, professional, ethical, and other relevant standards'.[44] This allows for the incorporation of the NHC's screening pathway of which an element recommends 'understanding that there are activities to monitor and evaluate'[45] from a participant's perspective. All the other elements of the NHC screening pathway – consistent with the detail and intent of right 6(1) – promote understanding, communication, choice and ongoing involvement for the participant.

The view that is taken at this point – subject to refinement in the discussion about right 6(1) in the context of rights 7(9) and 7(10) – is that activities to monitor and evaluate the newborn screening programme need to be more explicitly stated in information provided to parents in line with the NHC's screening process (7) and right 6(1).[46] Participation in a screening programme encompasses more than just the screening test and includes other essential components or activities such as monitoring and evaluation.[47] Presently, information for parents is provided in the leaflet 'Your Newborn Baby's Blood Test' and the NMSP website. The suggestion here is that the leaflet should mention that QA-related activities will be undertaken. It would also be helpful if the leaflet pointed parents to the website on which more detailed information could be provided.

3.2.4 'Reasonable patient'

Rights 6(1) and 6(2) of the Code provide that the right to be fully informed means every consumer has the right to information that 'a reasonable consumer, in that consumer's circumstances' would expect to receive, or needs in order to make an informed choice or give informed consent. This raises two issues. Firstly, there is the issue of the amount of information that should be provided (i.e. 'how much' information should be provided); this has been discussed above at 3.2.

Secondly, there is the issue of timing (i.e. 'when' information should be provided). Right 6(2) specifies that the right to information should be implemented just before the consumer makes an informed choice or gives informed consent. By comparison, right 6(1), which is drafted in more general and open-ended terms, is not limited in that way. It has been noted that right 6(1) applies to post-operative communications.[48] The applicability of the Code to the screening context needs to be kept in mind; therefore right 6(1) should be read as applying the standard of disclosure of information at *every* step of the screening pathway from the start to the end and not just in relation to the screening test, i.e. the taking of heel-prick blood samples.

The standard of disclosure of information under right 6 needs to meet the threshold required by 'a reasonable consumer, in that consumer's circumstances'. It has been observed that:

> *[w]hat information 'a reasonable consumer, in that consumer's circumstances' would need or expect calls for a judgment from the provider, and, in determining a complaint, the Commissioner. The open-ended nature of this over-arching right allows for standards of disclosure to develop over time.*[49]

It can also be added here that the language of the test is drafted broadly enough to allow standards of disclosure that accommodate the special features of the newborn screening context. These features include, for instance, standards of disclosure that take into account every element of the screening pathway as provided in the NHC guidance; developments in genetic knowledge and technologies; and the increase in the number of disorders being screened. All this can potentially make the obligation to meet the standard of disclosure seem quite onerous given that the programme has near 100 per cent coverage with the parents of over 55,000 newborns being involved.[50]

However, a couple of factors should be considered. First, the rarity of the disorders means that the number of newborns diagnosed and followed up will be small. Secondly, the profile of parents involved can be worked out in specific detail well in advance and within a limited number of discrete categories, such as first-time parents, second-time parents etc. (A note of caution should be sounded here against the making of assumptions that second-time parents will have good familiarity and recollection of newborn screening information previously provided when they

had their first newborn.) Additionally, these two factors, considered together, may help inform thinking about how the standard for information disclosure relating to newborn screening can be satisfied, and also about attitudes and expectations of parents regarding information on participation in newborn screening.

3.2.5 Aims of the newborn metabolic screening programme

The discussion has focused so far on the rights contained in the Code in the context of the NHC's screening pathway, from a participant's perspective. The pathway begins with the recommendation for an invitation to be screened, includes the test itself, and ends with the participant's understanding there are activities to monitor and evaluate at all stages. Each of these stages contributes to the fulfilling of the aims of the newborn metabolic screening programme, which are:[51]

1. to enable early detection of pre-symptomatic newborns;
2. to ensure appropriate early treatment of newborns;
3. to ensure newborns born with congenital metabolic disorders have their development potential impacted as little as possible from the disease;
4. to inform the community of all aspects of newborn screening including advantages and outcomes;
5. to facilitate early diagnosis, appropriate treatment and continuous monitoring of specific metabolic diseases;
6. to facilitate continuous quality improvement through the development of a quality assurance, reporting and strategic planning framework;
7. to provide educational resources to programme participants.

In the context of dried blood spots collected for the screening programme, there have been specific instances in which they have been released for purposes *not* related to the fulfilling of the aims of the programme. For example, in *H v G* the Court ordered release of dried blood spots relating to a deceased child for the purpose of proving paternity in a civil suit. Samples have also been released for use in the criminal context to match biological material found at a crime scene in order to identify victims.

As for the use of dried blood spots for the purposes of health research, the NTC Director has said that no research has occurred on stored de-identified dried blood spots in New Zealand.[52] However, even if the use of dried blood spots in health research may not be regarded as fulfilling the aims of the programme, some research proposals that involve the use of dried blood spots may inform or indirectly relate to the aims of the screening programme. Looking to the future, the potential benefits from the use of dried blood spots for research that yields generalisable knowledge will be significant in advancing many areas of health, from knowledge of clinical

genetics to population-based initiatives in public health. It is the view of the authors of this report that health research involving the use of de-identified dried blood spots collected for newborn screening should be encouraged as a matter of principle. In practice, research proposals must be carefully scrutinised on their merits and prioritised, given that the remaining dried blood spot samples are finite.

The past instances in which dried blood spots have been released for purposes *not* related to fulfilling the aims of the programme, and the potential benefits of using them for the purposes of health research that may inform or indirectly relate to improving the programme, bring home two points. First, it is important to distinguish between purposes that fulfil the aims of the programme and those that go beyond the aims of the programme. Secondly, there has to be clear policy for the retention and use of the dried blood spot samples that conveys that distinction. The right to be fully informed, pursuant to right 6, means that information about retention and use of dried blood spots, relating to purposes that fulfil the aims of the programme and purposes that go beyond the aims of the programme, must be given to participants so that they can exercise informed choice and consent. The issue of timing, and the amount of information to be given, is crucial.

3.3 Right 7: Right to make informed choice and give informed consent

3.3.1 Right 7 and the context of screening

The Code of Rights, as provided in right 7(1), brings legal force to the ethical requirement for informed choice and informed consent where a health-care procedure is not required by law.[53] Right 7(1) provides that:

> *Services may be provided to a consumer only if that consumer makes an informed choice and gives informed consent, except where any enactment, or the common law, or any other provision of this Code provides otherwise.*

Even though it is qualified by a standard drafting phrase that allows for exceptions, right 7(1) is written in strong, unambiguous language that communicates clear intent for general application. The definition of 'services' includes health-care procedures and 'consumer' includes a person entitled to give consent on behalf of a consumer.[54] 'Services' is a term wide enough to encompass the newborn metabolic screening programme, and the taking of heel-prick blood samples for newborn screening comes within the word 'procedure'. In the case of newborn screening, the newborn is the consumer on whose behalf parents or legal guardians can lawfully give consent for participation. As has been noted, the right of the consumer to make informed choice and give informed consent applies in relation to the whole screening pathway if the aims of the screening programme are to be fulfilled.

The Health and Disability Commissioner, in his opinion on Case 99HDC09011, said health-care professionals 'should not undertake the Guthrie Testing procedure without ensuring that informed consent has been obtained'.[55] The Commissioner also called for a policy to ensure that informed consent is obtained from parents or legal guardians.[56] Presently, an explicit policy to that effect does not appear to be publicly available although it seems conceivable that the policy will be contained in or will underpin the documents that are signalled as being in the planning stages, e.g. 'Quality and policy standards' and 'Storage, retention and use of blood spot cards'.[57] In place of the former are interim standards that exist but are not yet posted on the NMSP website.[58]

The Ministry of Health, in the document *Improving quality: A framework for screening programmes in New Zealand*, pointed out a number of matters that are relevant here. Principle 6 emphasises that:

> *Obtaining informed consent from eligible individuals is paramount. This includes the right to make an informed choice not to participate in screening, based on sound information.*[59]

To encourage principle 6 to be put into action, so as to deliver on a 'screening programme quality agenda',[60] the Ministry of Health has given guidance in the form of a checklist of expectations of provider responsibilities for implementation at various levels (i.e. at the level of the individual, team and organisation) which provides that:[61]

- the individual (health professional) is responsible for 'obtaining informed consent from people participating in the programme after discussing both the benefits and limitations of screening';
- the team (of providers) is responsible for 'ensuring that individuals within the team are supported to obtain informed consent'; and
- the organisation is responsible for 'ensuring that individuals have the time to obtain informed consent and provide necessary explanations to people participating in the programme'.

We suggest that principle 6 and the guidance provided be clearly incorporated into the interim standards if this has not already been done.

With regard to the right to exercise informed choice and consent in the context of the Code, the specific provisions of rights 7(9) and 7(10) are of particular significance and require careful consideration. The recent amendment to right 7(10) makes explicit and emphatic reference to issues regarding monitoring and evaluation for the first time in the Code. Closer examination of the issues relating to practice, implementation and

operation reveals that the interpretation of right 7(10) demands careful thought in relation to right 6(1), which promotes the right to full information, and right 7(9), which deals with return or disposal of bodily material. The discussion that follows contains some comment about retention or storage of dried blood spots, for fulfilling the aims of the newborn screening programme, and QA for newborn screening and an examination of the inter-relationship between rights 6(1), 7(9) and 7(10).

3.3.2 Retention of dried blood spots and physical or 'scientific' limitations on storage

The current practice of the National Testing Centre is to store the blood spot cards indefinitely after the tests for the seven disorders have been completed. The National Testing Centre has been storing blood spot cards ('Guthrie cards') for thirty-four years.[62] It is estimated that it currently holds more than two million samples, from both living and deceased New Zealanders.[63]

The utility of these blood samples is heavily dependent on the state of the technology. Twenty years ago, information that could be garnered from these samples was not of such great consequence. The utility of the samples was confined to the purpose of the newborn screening programme. With the advent of DNA technologies, including DNA profiling,[64] and the completion of the Human Genome Project, the degree and scope of information that can be derived from retained dried blood spots can potentially be very significant and have far-reaching implications.

In the report commissioned by the Health Resources and Services Administration, U.S. Department of Health and Human Services for public comment, entitled *Newborn Screening: Toward a Uniform Screening Panel and System*, it was stated, in the section discussing 'Establishing Principles for the Development of Newborn Screening Guidelines', that:[65]

> ***Newborn screening specimens are valuable health resources. Every program should have policies in place to ensure confidential storage and appropriate use of specimens.***
>
> *Specimens obtained for newborn screening have tremendous long-term value. They can be used for purposes of program quality management, to help inform deliberations about program expansion, and for research on testing technology and treatment and for epidemiologic studies. This is not to imply that every State should store all specimens forever but, rather, that there should be sufficient member States with diverse populations and long term storage of residual specimens to provide this critical resource. Regardless, it is important to ensure the confidentiality of those persons whose specimens are stored. The use of specimens for non-therapeutic purposes must not alter the willingness of the public to participate in newborn screening programs and related activities.*

A complete DNA profile of an individual can be obtained from the blood spots on the cards.[66] DNA profiles can be used to match DNA samples, from blood, tissue and other specimens, to individuals. The blood spot cards can also be used to identify genetic defects that lead to early and late-onset diseases in individuals. The cards may also provide information on a person's susceptibility to other diseases and disorders. These advances in technology mean that the blood spot cards can be used for purposes beyond which the samples were originally taken; that is, screening for metabolic disorders. The Australian Law Reform Commission found that:

> *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.*[67]

New Zealand's own Privacy Commissioner has commented that privacy concerns in relation to stored blood spots are heightened by the developing range of information which can be obtained by the analysis of old blood samples and the decreasing cost of carrying out such analysis.[68]

Worldwide, blood spot cards are now used for a variety of purposes, many of which go beyond the ambit of newborn screening programmes:

- investigation of cases missed by the screening programme (confirmatory diagnosis of false negatives);[69]
- screening programme development, method development and establishing normal ranges for new and existing tests (evaluation of new and existing tests);
- requests from health professionals for the purpose of clinical investigation;[70]
- coronial and forensic purposes;[71]
- research purposes, including:
  - epidemiologic surveys of infectious diseases[72]
  - population-based studies of environmental and pharmacologic exposures[73]
  - etiologic studies of birth defects and developmental disabilities[74]
  - population-based studies of haplotype and allele frequencies for genetic disorders and potentially significant gene polymorphisms.[75]

In New Zealand, the purposes of storage appear at first glance to be limited. The leaflet issued by the Newborn Metabolic Screening Programme states that:

When the testing of your baby's blood is completed, the sample card is stored so that if a baby has one of the conditions tested for, but does not have a positive test result we can find out why the mistake occurred again. Some of the blood might be used to set up new screening tests; if a leftover scrap of your baby's blood is used for this all

the information about your baby will be disconnected from the blood so any results cannot be traced back to you and your baby.

This conveys that there is a dual purpose in the storing of blood spot cards. First, they are stored to allow investigation of instances in which a baby, who has been screened and tested negative, develops one of the conditions for which the baby has already been screened. Some of the conditions tested for do have the potential to present as late-onset disorders, e.g. in adulthood. Other disorders may have milder forms that are not detected by initial newborn screening. The leaflet provides little indication as to how long samples are needed for this purpose. The second stated purpose is for the setting up of new screening tests. In connection with this purpose, the leaflet does not provide any indication as to the degree of identifiability of the stored sample cards, e.g. whether use of the cards for this purpose will be in de-identified, identifiable or anonymised form.

The use of blood spot samples stored on filter paper is subject to two physical or 'scientific' limitations. The first relates to the actual physical size or amount of the blood spot samples. The second relates to the stability of the analytes present in the blood spots.[76] The stability of the analytes, and the amount available for testing, limits any proposed future use of the blood spot samples.

The first limitation relates to the amount of blood initially taken and eventually stored. When samples are taken, the blood is spotted onto four rings marked on filter paper.

Holes are punched from the four dried blood spots for the purposes of newborn metabolic screening. The amount used during the testing process can range from one blood spot sample (that is, one ring) to all of the blood spot samples.[77] In some instances, more blood may be required from the newborn to complete testing, such as when initial samples are inadequate, or it is necessary to follow up investigation of false negatives.

Metabolites initially tested during the newborn screening process have a finite life span.[78] At some point these metabolites become unstable or are degraded in such a way as to no longer produce meaningful test results (or, in some instances, any results at all). Storage conditions are known to affect the stability of metabolites. The Council of Regional Networks for Genetic Services concluded that maximum stability could be achieved for most analytes when samples are stored at low temperature and in controlled low humidity.[79] The Council also noted that its search of scientific literature on the stability of analytes in dried blood spots was of minimal value; there was a lack of published data that would allow meaningful conclusions about long-term storage to be drawn.[80] The Australian Law Reform Commission has commented that proper long-term storage of newborn screening cards for research purposes would be complex and expensive.[81] Given these observations, it is important to appreciate

that the stability of metabolites in dried blood spots is not infinite; at varying times metabolites will lose their ability to produce meaningful results. Storage conditions can enhance the length of time for which metabolites will be stable, but they cannot do so indefinitely.

One substance in dried blood spots is known to persist longer than any other metabolite – deoxyribose nucleic acid (DNA).[82] In 2005, Chaisomchit et al. concluded that genomic DNA is stable in dried blood, stored on filter paper at ambient tropical conditions, for at least eleven years. However, it was found that DNA quality for amplification of larger DNA fragments decreased when the specimens were stored for longer than ten years.[83] The authors of the study noted that no other published data exist regarding the effects of long-term storage of dried blood specimens on the stability of genomic DNA.[84] The New Zealand case involving criminal investigations into the suspected murder of Olivia Hope and identification of hair and blood samples suggests that DNA is viable for analysis for up to twenty-one years.[85] In that case, DNA analysis was carried out on dried blood spots, that had been collected for newborn screening, with the purpose of comparing and identifying DNA extracted from hair and blood samples found at the alleged crime scene.[86]

The physical or 'scientific' limitations must be borne in mind when considering any policy development in relation to the retention and long-term storage of newborn blood spot cards. Current and relevant scientific literature on the stability of metabolites, DNA extraction and testing technology and optimal storage conditions needs to be taken into account in any policy development in this area.

3.3.3 Right 7(10) and quality assurance activities

Right 7(10) of the Code states that 'no body part or bodily substance removed or obtained in the course of a health-care procedure may be stored, preserved or used otherwise than … with the informed consent of the consumer'.[87] One of the exceptions to this is provided 'for the purposes of a professionally recognised quality assurance programme, an external audit of services, or an external evaluation of services' where these activities are 'undertaken to assure or improve the quality of services'.[88]

In *Screening to improve the health of New Zealand: Criteria to assess screening programmes*, the National Health Committee defines 'quality' as embracing the following concepts:[89]

- **Safety** – the extent to which harm from a service is kept to a minimum;
- **Consumer focus** – the extent to which a service meets the needs of consumers, incorporates community values and allows opportunities for participation and input into decision-making;

- **Access** – the extent to which people are able to receive a service on the basis of need and irrespective of factors such as ethnicity, age, location, impairment or gender;
- **Effectiveness** – the extent to which a service achieves an expected and measurable benefit;
- **Efficiency** – the extent to which a service obtains the greatest possible benefit from available funding, i.e. value for money.

The National Health Committee has also noted that:[90]

> *Poor screening programme quality, or a decline in screening programme quality can tip the balance between benefits and harms the wrong way. Once the invitation to be screened is issued, there is an ethical obligation to ensure that the programme can deliver the potential benefits through appropriate quality management. It often requires significant investment to achieve and maintain the level of quality necessary to ensure the expected benefits occur.*

The Health and Disability Commissioner has commented that:[91]

> *consent to the provision of a health service implies consent to proper audit and evaluation of the service. The public assumes that medical laboratories have QA procedures in place to check that specimens have been read correctly.*

The three activities afforded exemption under right 7(10)(c) – i.e. 'quality assurance', 'external audit' and 'external evaluation' – are aimed at ensuring screening programme quality. Some definitions may be helpful in order to shed light on these expressions, which may seem like terms of art, specific to a certain profession or group of experts. We searched for relevant comments or guidance on these expressions in the screening context.

'Quality assurance' has been defined by the NHC as the 'detection of problems through external or internal inspection, and their correction through systematic activity'.[92] By implication, the Privacy Commissioner seemed to suggest that, for the newborn metabolic screening programme, a formal audit process would involve going back to a representative sample of the cards and retesting the samples.[93] An 'external evaluation' has been described by the National Screening Unit (NSU) as 'monitoring and assessing the service delivery and outcomes of a screening programme, which may include assessing overall programme effectiveness, cost-effectiveness and acceptability'. The NSU also noted that an 'evaluation will determine whether the programme is actually delivering on its objectives'.[94]

In 2003, the Privacy Commissioner observed that:[95]

> *The National Testing Centre considers that its handling procedures, rather than any questions of 'reading' or evaluating samples or test results, are what matters in terms of quality assurance. The Centre undergoes regular accreditation audits. These do not involve subsequent re-testing of samples or their retention for that purpose.*[96]

The Health and Disability Commissioner's complaint investigation in Case 99HDC09011 was provided with the following information:

> *Where there is a mistake made in the testing of the sample and a case of one of the screened conditions is missed … the original sample would be sent to another newborn screening laboratory for checking on different equipment. This is a crosscheck on the sample and also useful from quality assurance perspective.*

The investigation of cases missed by a screening programme has been noted as the primary purpose of retention of screening samples in the HGSA's *Policy statement on the retention, storage and use of sample cards from newborn screening programs.*[97] The newborn screening programme undergoes peer review every two to three years and internal audits are carried out annually.[98]

Our search for information in this area reveals that New Zealand participates in an organised QA programme; one in which more than fifty other countries participate. The Newborn Screening Quality Assurance Programme (NSQAP) is run by the Centres for Disease Control and Prevention and the Association of Public Health Laboratories. The programme has two core components designed to ensure the quality of newborn screening programmes: quality control and proficiency testing.

The quality control component involves distribution of dried blood spot materials at six-monthly intervals by the NSQAP. Participating laboratories are expected to return quantitative results from five different analytical runs, having utilised the dried blood spot materials supplied. The NSQAP compiles and distributes the reported results. Laboratories can compare their results with those obtained by other laboratories in order to gauge the accuracy of their quantitative results.

Proficiency testing involves the distribution of quarterly panels of dried blood spots that participants are expected to analyse once. Participants are expected to return analytical results and qualitative (clinical) assessments. These results and assessments are performance graded by the NSQAP. As with the quality control programme, the NSQAP compiles and distributes proficiency testing reports. These reports show the distribution of analytical values and qualitative assessments reported by participants.

Involvement in this QA programme would come within the definition of a 'professionally recognised quality assurance programme' under right 7(10)(c)(i). Hence, storage for such purposes *without* the informed consent of the consumer may not result in a breach of the Code of Rights, as might have been the case before the 2004 amendments to right 7(10).

3.3.4 Inter-relationship of right 7(10), right 6(1), right 7(9) and the National Health Committee (NHC) screening guidance, and options for reconciling them

Prior to the amendment of right 7(10), the consumer's informed consent was necessary for the storage, preservation or use of any body parts or bodily substances removed or obtained during the course of a health-care procedure. Recent changes to right 7(10) removed the need to obtain informed consent for QA activities, i.e. 'activities that are … undertaken to assure or improve the quality of services', such as professionally recognised QA programmes, external audits of services or external evaluations of services.[99]

A plain reading of right 7(10) by itself may give cause for concern that QA activities can (and, as they are essential steps in the screening pathway, will) inevitably take place and involve the use of a consumer's bodily material regardless of that consumer's knowledge or continuing support for or involvement with the health service at issue. The Health and Disability Commissioner has responded to concerns over the implications of the new right 7(10) by saying that it applies in limited circumstances and safeguards exist in the Code and Ethics Committee review.[100] The Health and Disability Commissioner has also pointed out that right 7(9) continues to be available for consumers who require the destruction or disposal of their bodily material.[101]

A few observations can be made about the relationship between the amended right 7(10) on the one hand, and right 7(9), right 6 and the NHC guidance regarding the screening pathway on the other. By implication, the exception created by the newly amended right 7(10) seems to limit or restrict the full extent of the operation of the right to be fully informed as provided by right 6. Secondly, even though the recent changes to right 7(10) may seem supportive of one of the elements of the NHC's screening pathway, by enhancing the participant's understanding of the fact that there are activities to monitor and evaluate at every stage of the screening process, right 7(10) does not seem to be in keeping with the spirit of the NHC's screening pathway as a whole in terms of encouraging ongoing engagement with screening participants. Thirdly, QA activities can be conducted under right 7(10) without informed consent only so long as right 7(9) is not exercised by consumers to require the return or destruction of their bodily material.

The question that arises here is:[102] How can the consumer's right to require the return or disposal of bodily material[103] and, more generally, the right to be fully informed,[104] and the NHC screening guidance for informed choice and consent and understanding about QA activities,[105] sit consistently alongside the exception in right 7(10) that allows QA activities to be undertaken on bodily material without informed consent?

RIGHT OR GUIDANCE	RELATIONSHIP
Right 6 provides the right for consumers to be fully informed	Not consistent with right 7(10)(c) but consistent with NHC guidance
The NHC screening pathway includes elements that promote:	
• informed choice and informed consent (elements of pathway, taken as a whole)	Not consistent with right 7(10)(c) but consistent with right 6
• understanding of the fact that there are activities to monitor and evaluate at all stages of the pathway (last element of pathway)	Not consistent with right 6 but consistent with right 7(10)
Right 7(10) precludes the obligation to obtain informed consent to store, preserve, or use bodily material for QA purposes	Not consistent with right 6
Right 7(9) provides the right for consumers to require the return or disposal of their bodily material	Not consistent with right 7(10)(c) but consistent with NHC guidance

Table 1: *Inter-relationship of rights 6(1), 7(9), 7(10) and NHC screening guidance*

After examining the underlying tensions arising from right 7(10), right 7(9), right 6 and the NHC guidance, and reflecting on how they can be reconciled in practice with minimal conflict to achieve the best possible result, we propose the following two options.

Option A: Status quo with active communication of more information about right 7(9) and right 7(10)

Option A involves opting for the status quo with retention of the dried blood spot samples indefinitely and with the programme being subject to the general operation of the law. However, to give greater effect to the Code and enhance awareness of

the rights therein, so that they can be meaningfully exercised, two things have to be actively done. First, parents need to be told about the use of samples without informed consent for the purposes of QA-related activities as consistent with right 7(10)(c). Secondly, at the same time, parents also need to be told that they have the right to ask for return or disposal of their newborn's dried blood spot sample, consistent with right 7(9).

Given the underlying tension between right 7(10)(c) and right 7(9) in practical operation, we suggest that communication of both these rights be accompanied by reassurances about safeguards relating to use of the samples for QA-related activities, and strong encouragement regarding participation in the programme for the benefit of the newborn. A specific example of this is to tell parents that samples will, for practical purposes, need to be retained for a particular period of time for QA-related activities to be carried out and that the exercise of right 7(9) before or during that period will prevent this from happening. A specific, designated time period will go some way towards providing parents with a timeframe during which to defer the exercise of right 7(9). It would be helpful for this information to be included in the leaflet distributed to parents and posted on the NMSP website, and also for the Interim Standards to include guidance for LMCs to convey the information to parents.

There appear to be good arguments in favour of the status quo, strengthened by the suggestion of conveying reassurances and encouragement to parents and, incidentally, the wider public. Until now, there seems to be no overt, publicly documented concern or objection regarding the way in which things are working. It would be reasonable to continue with the status quo until such time as current arrangements should be reviewed, such as when significant numbers of samples were needing to be returned or destroyed so that QA-related activities could not be undertaken in a statistically meaningful way. This option is effectively underpinned by the policy to persuade parents themselves to waive their exercise of right 7(9) for a specified period.

Option B: Legal authority to prescribe a minimum retention period to guarantee all samples are available for QA-related activities

Option B involves prescribing a minimum period during which the programme can retain dried blood spot samples so that all the samples are available for QA-related activities, hence enabling the screening pathway to be completed. However, as it is open for parents to exercise the right for disposal or return of the samples at any time, legal authority (i.e. in the form of statutory authority) will be needed to override the ability to exercise right 7(9) for a specified period in order to guarantee availability of all samples for QA-related activities. Another way of characterising this is that legal authority would be needed for temporary, time-limited suspension of right 7(9) while QA-related activities on all samples could be carried out unhindered.

With this option, at least two things should be done in order to strengthen current arrangements relating to the voluntary system of newborn screening in New Zealand. First, parents need to be told explicitly about the statutory authority that – for a temporary, time-limited period – overrides or suspends their right to ask for disposal or return of the samples. Secondly, to preserve the participation rate in the programme of, to date, almost 100 per cent of newborns, the message should be communicated to parents that QA is a key component of the screening programme.

Several matters can be noted regarding this option (Option B) that require additional consideration. The first is the question of the implication on the future participation rate for the screening programme. Presently, no relevant studies are available from which any observations can be made; any comments that can be made would be speculative. Hence, research or surveys must be undertaken to ascertain parental attitudes in relation to this option so as to inform policy-makers and decision-makers before its adoption.

Secondly, there is the question of exactly when samples can be returned or destroyed. It is important to emphasise that, as this option involves temporarily overriding or suspending the exercise of right 7(9), the specified minimum period for retention of samples must be explicitly stated in years, and set out clearly and emphatically in information given to parents. Thirdly, to earn support for this option and at the same time avoid or minimise the likelihood of drop-out rates in participation for newborn screening due to misunderstanding, poor awareness or knowledge or groundless fears, clear and unambiguous information that explains and justifies what is being done – including information about the specified minimum sample retention period as previously discussed– should be provided to parents in the leaflet, for example, and on the programme website, and also noted in the Interim Standards.[106]

These two options, which are not necessarily the only solutions, illustrate the range of possibilities – from continuing with the status quo to prescribing specific requirements with the authority of the law. It is vital, and to be seen overtly, to take policy or legislative steps to reconcile the tensions underpinning the way in which right 7(10), right 7(9), right 6 and the NHC guidance relate to each other. It should be noted that the discussion has so far focused only on the scope of activities related to fulfilling the aims of the newborn screening programme. Issues regarding the extent or limits of storage and use of dried blood spots for purposes beyond the aims of the programme will be examined later, after brief reference to documentation regarding informed choice and consent and refusal of services and withdrawal of consent under right 7(1) and right 7(7) respectively.

3.3.5 Documentation in the context of right 7

The Code, framed in the language of claim rights, is written from the consumer's perspective. Documentation of actions taken, and the administrative systems relating to the obtaining of informed consent, the various stages of the screening pathway and the screening programme generally, is a matter that rests with the providers or health-care professionals involved. The Code makes no explicit reference to or requirement regarding documentation. Nevertheless, the Ministry of Health, in *Improving quality: A framework for screening programmes in New Zealand*, has stated that '[i]nformed consent should be documented'.[107]

In his comments on Case 97HDC8205,[108] the Health and Disability Commissioner repeated his predecessor's recommendation for 'an integrated documentation system where records were comprehensive, appropriate, and available to all staff' and said he 'cannot emphasise enough the importance of integrated documentation systems'.[109]

More generally, the Commissioner noted past investigations, which had found patient notes had been fragmented and held in a number of places or in the same location on a number of forms, and pointed out that this means

> *that those who are involved in providing care to the consumer may lack access to crucial information at important times, or are unable to refer to the necessary information because they cannot find it amongst a myriad of forms. This results in confusion for all involved in the consumer's care, and sometimes in crucial information being overlooked by staff, with significant consequences [for] the consumer.*[110]

The Commissioner, in Case 97HDC8205, found that the fragmented documentation system and poorly co-ordinated service provision failed to ensure quality and continuity of care and that the providers and professionals involved were in breach of rights 4(5) and 4(2).

In Case 99HDC09011, where the complaint related to informed consent for blood samples obtained in relation to newborn screening, the Health and Disability Commissioner recommended that LMCs document the fact of disclosure of information, the giving or refusal of consent from parents to the taking of newborn blood samples and the outcome regarding storage or disposal of those samples.[111]

The importance of documentation was recently stressed by the Victorian Newborn Screening Review Committee, which stated that:

There should be documentation in the mother's/baby's hospital record stating that there has been discussion about the newborn screening test. The hospital record should also show a record of completion of testing. … Any parents refusing testing

are requested to sign a written statement saying that they understand the potential risks to the healthy development of their baby.

Documentation for retention, release and use of blood spot sample cards is recommended in the *Policy statement on the retention, storage and use of sample cards from newborn screening programs* developed by a joint subcommittee of the Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians:[112]

> *3.2 Retention of sample cards*: The screening program should document how long it will retain their cards, including the purposes for retention.
>
> …
>
> *2.2 Release of sample cards*: All releases of residual samples should be documented. Such records should include the purpose of release, what material was released, by whom it was used, and the authority for use.
>
> …
>
> 4.3 *Uses of sample cards*: Specific uses include, but are not limited to, the following. Each program policy should state what permission and documentation are required in each situation:
>
> 4.3.1 Investigation of cases missed by the screening program …
>
> 4.3.2 Screening program development, method development, and establishing normal ranges for new or existing tests …
>
> 4.3.3 Individual requests (i.e. requests for cards to be returned to the family) …
>
> 4.3.4 Requests from Health Professionals …
>
> 4.3.5 Research Studies …
>
> 4.3.6 Coronial and forensic use … .

In New Zealand, the right of every consumer to refuse services and withdraw consent to services is provided by right 7(7), which is the mirror opposite of right 7(1) stating that services may be provided only if the consumer exercises informed choice and consent. Documentation that records the refusal or withdrawal of consent should also record instances in which consent is given. There is guidance in New Zealand for implementing provider responsibilities at various levels in relation to obtaining informed consent for screening programmes in general (as discussed earlier). Specific policies or procedures setting out the requirements for documentation of informed consent, or refusal or withdrawal of services, in the newborn screening context should be made explicit and publicly available.[113] This would help increase parental and public awareness, understanding and confidence. The United Kingdom

Newborn Screening Programme Centre provides precedence for material containing such policies and procedures to be posted on the internet.[114]

4 FINDINGS

The analysis in this part of the report is grounded in the rights-based Code of Rights, and takes into account the public health paradigm and the striking aspects of dealing with genetic risks in families. In this context, it is important to distinguish public health screening from personal clinical services. Traditionally, public health law has been prescriptive in form and has compelled participation. This is in contrast with the increasingly adopted consumer-based approach that promotes informed choice and consent and which is in keeping with the complexities and sensitivities surrounding genetic medicine.

Information about newborn screening should be given to parents by or during the third trimester, and again before the samples are taken. The applicability of the Code in the context of screening needs to be borne in mind. The significance of the 'screening pathway' needs to be appreciated: 'It is a critical part of informed consent … to know about the entire screening pathway and the possible things that might occur before [taking] the first step'. Clear, unambiguous information needs to be provided, emphasising that the purpose of 'participation' is in the interests of the newborn.

If newborn screening increasingly operates in a paradigm that actively promotes informed choice and consent, parents or guardians will need to be informed throughout the screening process; this includes being notified about results. The policy of 'no news is good news' may have to be reconsidered in the light of complex issues raised by technologies that reveal carrier status, or if screening is extended to include late-onset disorders. Surveys and empirical studies should be undertaken in New Zealand to inform policy in this and other areas. Public discussion, e.g. led by parliamentarians, needs to take place.

Activities to monitor and evaluate the programme need to be more explicitly stated in information given to parents. Related to this, is the importance of distinguishing between initiatives taken for the purposes of fulfilling the aims of the programme and those that go beyond aims of the programme. Policies regarding retention and use of samples that clearly make and communicate that distinction need to be provided, especially to parents.

The utility of the retained blood samples is heavily dependent on the state of the technology. With DNA technologies and profiling, the degree and scope of information that can be derived from dried blood spots will potentially be very significant and have far-reaching implications. There is tremendous long-term value in retention, for example, for the purposes of quality management, programme expansion, research

on testing and treatment and epidemiologic studies. Current and relevant scientific literature on the stability of metabolites, DNA extraction and testing technology and optimal storage conditions needs to be taken into account with regard to any policy development in this area.

Two options are provided in order to reconcile the inter-relationships between the various provisions of the Code of Rights on consent, storage and quality assurance and the NHC screening guidance: to maintain the status quo by more actively communicating information about right 7(9) and right 7(10); or to prescribe, with legal authority, a minimum retention period to guarantee all samples are available for QA-related activities.

The public availability of policies and procedures setting out, for example, the taking and documenting of informed consent would be helpful, in order to increase parental and public awareness, understanding and confidence.

ENDNOTES

1 This would be echoed by the Victorian Privacy Commissioner who recently commented that '[t]he public interest in maintaining confidence in newborn screening will not be served by artificially insulating the newborn screening program from the large public policy issues that arise from mankind's rapidly growing knowledge of genetics. We will serve the public interest best if we acknowledge – and make clear to the public – that the newborn screening program is unavoidably connected to advances in genetics, and that the implications of this can be managed properly under the law by confident use of the familiar techniques of a democratic society'. See P. Chadwick (Victorian Privacy Commissioner), Supplementary Report of the Victorian Privacy Commissioner, Appendix 3, in Victorian Newborn Screening Review Committee, Department of Human Services, Final Report for the Minister for Health, August 2006.

2 The programme, which has been in place since the 1960s, is reported to have near 100 per cent coverage with over 55,000 newborns screened per year. Available at: http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/2a5a1e01dddf6315cc2570c0000bf76c?OpenDocument viewed 18 December 2006.

3 The seven disorders currently screened for are biotinidase deficiency, congenital adrenal hyperplasia, cystic fibrosis, galactosemia, hypothyroidism, maple syrup urine disease (MSUD) and phenylketonuria (PKU). While, for example, PKU is the result of mutations in the structural gene for phenylalanine, the cause of thyroid dysgenesis (the most common cause of congenital hypothyroidism) is largely unknown; however, a small minority of cases have been attributed to mutations in the genes that control thyroid gland development and in genes that are responsible for iodine trapping. Since 1 December 2006, the number of disorders screened for has been expanded. The newly added disorders, broadly grouped into disorders of amino acid breakdown and fatty acid oxidation (PKU and MSUD, which have already been on the panel of tests, are classified in the former group), help in the detection of early-onset disorders that have an underlying genetic basis.

4 In the 4 August 2000 report on Case 99HDC09011, the Health and Disability Commissioner noted the existence of 'a "bank" of Guthrie Test samples at the NTC from the majority of babies born in New Zealand in the last 28 years'. See http://www.hdc.org.nz/files/hdc/opinions/99hdc09011.pdf viewed 18 December 2006. It was pointed out, in the United States context, that the 'collections of Guthrie cards stored by state newborn-screening laboratories can … be viewed as inchoate "DNA

banks"'. See McEwen J.E. and Reilly P.R. Stored Guthrie cards as DNA banks. Am J Human Genet July 55(1): 196–200, 1994.

5 Privacy Commissioner, Guthrie tests. A report by the Privacy Commissioner following his inquiry into the collection, retention, use and release of newborn metabolic screening test samples, pursuant to section 13(1)(m) of the Privacy Act 1993, September 2003, para 4.8. Available at: http://www.privacy.org.nz/filestore/docfiles/70989185.pdf viewed 18 December 2006.

6 Pending policy decisions governing the use and storage of the blood spot cards, the Memorandum of Understanding relating to the disclosure of newborn blood spot samples and related information by the Ministry of Health to the New Zealand Police 2006 ('MOU') sets out protocols between the Ministry of Health and the Police in relation to the release of the blood spot cards held by the ADHB for purposes other than the primary reason of fulfilling the aims of the screening programme. Available at: http://www.moh.govt.nz/moh.nsf/0/58182C57A54ECF4DCC257138001500CE/$File/memorandum-of-understanding.pdf viewed 18 December 2006.

7 Kedgley, S. (M.P.) DNA databank danger, 21 November 2001. Available at: http://www.greens.org.nz/searchdocs/PR4902.html viewed 18 December 2006.

8 The Explanatory Note to the Human Tissue Bill (introduced to Parliament on 7 November 2006) notes several areas of concern relating to non-therapeutic uses of tissue, and specifically makes reference to the retention of Guthrie cards that have accumulated from 'a type of genetic screening where blood samples are taken from newborn children for metabolic testing'. See the Explanatory Note, 22.

9 Department of Health, United Kingdom. Our inheritance, our future – Realizing the potential of genetics in the NHS, June 2003, paras 3.36 to 3.38.

10 The Health and Disability Commissioner received the complaint on 18 August 1999, commenced investigation on that date and issued an opinion on 4 August 2000. See Report on opinion – Case 99HDC09011. Available at: http://www.hdc.org.nz/files/hdc/opinions/99hdc09011.pdf viewed 18 December 2006.

11 Privacy Commissioner, Guthrie tests. A report by the Privacy Commissioner following his inquiry into the collection, retention, use and release of newborn metabolic screening test samples, pursuant to section 13(1)(m) of the Privacy Act 1993, September 2003. Available at: http://www.privacy.org.nz/filestore/docfiles/70989185.pdf viewed 18 December 2006.

12 High Court, Auckland, M1868/98, May 1999.

13 (2000) 18 FRNZ 572.

14 The NMSP webpage entitled 'Project planning' lists five items of work intended to be undertaken: policy and interim standards; storage, retention and use of blood spot cards; Memorandum of Understanding with Police regarding the use of residual blood spot samples; expansion of screening to cover more metabolic conditions; and educational material for families and health professionals. Available at: http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/cabf221ef8da9061cc2570c0000c5097?OpenDocument viewed 18 December 2006, where it is noted that the contents were last updated on 11 November 2005.

15 Clause 4 of the Code provides that 'consumer' includes a person entitled to give consent on behalf of that consumer.

16 The National Advisory Committee on Health and Disability ('National Health Committee') adopts the following definition of screening, which is based closely on that of the United Kingdom National Screening Committee:

Screening is a health service in which members of a defined population, who do not necessarily perceive they are at risk of, or are already affected by, a disease or its complications, are asked a question or offered a test to identify those individuals who are more likely to be helped than harmed by further tests or treatments to reduce the risk of disease or its complications.

(NHC, Screening to improve health in New Zealand: Criteria to assess screening programmes, April 2003, 5. Available at: http://www.nhc.govt.nz/publications/PDFs/ScreeningCriteria.pdf viewed 18 December 2006.)

17 See the Health and Disability Commissioner (HDC) Act 1994 and the Code of Health and Disability Services Consumers' Rights that is promulgated under the HDC Act.
18 See the definition of 'informed consent' in s 2 of the HDC Act.
19 The extent of this right has been the subject of comment by the Health and Disability Commissioner who says, '[d]octors are required to facilitate understanding, but cannot be expected to *guarantee* patient understanding, and the law makes no such requirement' (emphasis added). See Paterson R. Informed consent in New Zealand: Medical myths. NZ Med J 116: 1183, 2003. Available at: www.nzma.org.nz/journal/116-1183/628.
20 As will be discussed later, consumer expectations and ethical standards for screening may differ from those associated with clinical diagnosis and treatment.
21 The two health professionals involved were an independent LMC and a staff member at Auckland Healthcare. The former considered her legal obligation, spelt out contractually under the 'section 51 notice', was 'to ensure that newborn screening was provided, not to obtain informed consent for the procedure' (HDC report at 12). With the latter, the HDC found an assumption was made by Auckland Healthcare that informed consent had been given because Auckland Healthcare believed that the LMC had contractual responsibility to obtain informed consent (at 13).
22 Privacy Commissioner's Report, para 8.6, in which, at the end, the Commissioner noted that practice may have changed since the survey was undertaken.
23 Holtzman et al. Effect of informed consent on mothers' knowledge of newborn screening. Pediatrics 72(6): 807–12, 1983. Cited in Elkin and Jones. Guthrie cards: Legal and ethical issues. New Zealand Bioethics Journal, October 2000, 22 at 23.
24 See http://www.newbornscreening.info/felsi.html. The Screening, Technology And Research in Genetics (STAR-G) Project is a multi-State collaborative effort, led by the Hawai'i Department of Health, to obtain research data, identify strategies and develop materials for addressing the financial, ethical, legal and social issues (FELSI) surrounding the use of MS for neonatal metabolic screening of culturally and ethnically diverse populations. The participating states include Alaska, California, Idaho, Oregon and Washington.
25 For example, Davis T.C. et al. Recommendations for effective newborn screening communication: Results of focus groups with parents, providers, and experts. Pediatrics Vol. 117, No. 5, May 2006, S326–40, and Faulkner L.A. The newborn screening educational gap: What prenatal care providers do compared with what is expected. American Journal of Obstetrics and Gynecology (2006) 194, 131–7.
26 The Newborn Metabolic Screening Programme (NMSP) website notes that interim standards have been drafted in a collaborative effort between the National Screening Unit (NSU) and the Director of the NMSP and that, in the first year of the NSU's responsibility for the NMSP, and through its relationship with LabPlus, the standards will continue to be edited and consultation will be continued in order to form a comprehensive and robust document to provide strategic direction and monitoring for the programme. See: http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/5d147a9d39a89eefcc2570c0000c3fa0?OpenDocument viewed 18 December 2006.
27 First recommendation in the supplementary report of the Victorian Privacy Commissioner, Final Report for the Minister of Health, August 2006 by the Victorian Newborn Screening Review Committee. Available at: http://www.privacy.vic.gov.au/dir100/priweb.nsf/download/9553F3C0F6D2A231CA257218000109B3/$FILE/NBS%20Report%20Final.pdf viewed 18 December 2006.
28 Parts 1, 2 and 3 at pages 14, 20 and 22 respectively. See United Kingdom Newborn Screening Programme Centre. Newborn blood spot screening in the UK, Policies and Standards, April 2005. Available at http://www.ich.ucl.ac.uk/newborn/download/policies_standards.pdf viewed 18 December 2006. At footnote r, page 20, it is noted that, '[c]ommunication during pregnancy is recommended because it is recognised that after birth can be a difficult time for parents to make an informed choice'.

29 The United States Task Force on Newborn Screening recommended that prospective parents receive information about newborn screening during the prenatal period, preferably during a routine third-trimester prenatal care visit. See, Task Force on Newborn Screening. Serving the family from birth to the medical home: A report from the Newborn Screening Task Force convened in Washington DC, 10–11 May 1999. Pediatrics 106(2 pt 2): 383–427, 2000.

30 For example, the Primary Maternity Services Notice which is issued pursuant to s 88 of the New Zealand Public Health and Disability Act 2000. Consultation on the Primary Maternity Services Notice is underway. Available at: http://www.moh.govt.nz/moh.nsf/pagesmh/4097/$File/proposed-section-88-maternity-231106.pdf viewed 18 December 2006. This was preceded by the Review of Maternity Facility Access Agreement Consultation. Available at: http://www.moh.govt.nz/moh.nsf/pagesmh/4097/$File/Access+Agreement+Consultation.doc viewed 18 December 2006.

31 In the context directed primarily at cervical and breast screening, Sandra Coney has commented 'the Code is not entirely adequate for screening. Screening differs from the usual relationship between providers and users of services in that the consumers are not sick people seeking treatment. They are well people who are invited by others to take part'. Informed Consent in Screening Programmes, Healthy Women Workshop Education and Training for Health Promotion, National Cervical Screening Programme and BreastScreen Aotearoa, Health Funding Authority, Napier, 23 June 2003.

32 Coney S. (ed.) Women's health update, Vol. 4, No. 2, July 2000. Available at: http://www.womens-health.org.nz/publications/WHU/whuvol4.htm viewed 18 December 2006.

33 National Health Committee (NHC) (National Advisory Committee on Health and Disability). Screening to improve the health of New Zealand: Criteria to assess screening programmes, April 2003, 5. Available at: http://www.nhc.govt.nz/publications/PDFs/ScreeningCriteria.pdf.

34 NHC (National Advisory Committee on Health and Disability). Screening to improve the health of New Zealand: Criteria to assess screening programmes, April 2003, Glossary, 30. Available at: http://www.nhc.govt.nz/publications/PDFs/ScreeningCriteria.pdf. Note that the bullets, as they originally appear in the NHC report, are numbered here for ease of reference.

35 Re (2): the list suggested by the United Kingdom General Medical Council for informed consent in relation to screening is adopted here. See, Seeking patient's consent: The ethical considerations, 1998, para 34. Available at http://www.gmc-uk.org/guidance/current/library/consent.asp viewed 18 December 2006.

36 Task Force on Newborn Screening. Serving the family from birth to the medical home: A report from the Newborn Screening Task Force convened in Washington DC, 10–11 May 1999. Pediatrics 106(2pt2): 383–427, 2000.

37 The factors identified by the AAP for successful screening included expansion of public health infrastructure, establishing advisory committees, implementing programme surveillance and research and providing parents with comprehensive educational material.

38 See also Fant K.E. et al. Completeness and complexity of information available to parents from newborn screening programs. Pediatrics 115: 1268–72, 2005; and Hiller E.H., Landenburger G. and Natowicz M.R. Public participation in medical policy-making and the status of consumer autonomy: The example of newborn-screening programs in the United States. Am J Public Health 87: 1280–8, 1997.

39 United Kingdom Policies and Standards, at 20. Also, one of the procedures under 'Blood Sampling Guidelines' sets out the requirement to '[i]nform parents how and when they will receive results'.

40 Victorian Newborn Screening Review Committee, Department of Human Services, Final Report for the Minister for Health, August 2006, 3.

41 Victorian Newborn Screening Review Committee, Department of Human Services, Final Report for the Minister for Health, August 2006, 35 and 36.

42 Victorian Health Issues Centre. Informed parental consent for newborn screening in Victoria. Final Report, December 2005, at 26 and 34 respectively.

43 Victorian Health Issues Centre. Informed parental consent for newborn screening in Victoria. Final Report, December 2005, 24.

44 Right 6(1)(e).
45 See the last step in the NHC's definition of 'screening pathway' which, for ease of reference in this paper, is numbered as screening process (8).
46 The distinction between rights 6 and 7 is that while the latter imposes a duty on providers and professionals to obtain informed consent from the consumer, the former puts the obligation on providers and professionals to give full information. In general, the satisfactory discharge of duties under right 7 by providers and professionals may be wholly or substantially dependent on prior discharge of obligations under right 6. However, even if right 7 creates exceptions regarding the duties to consumers of providers and professionals, i.e. in the case of rights 7(10)(b) or 7(10)(c), we are of the opinion that right 6 can still be exercised by consumers to require providers and professionals to discharge their obligation to provide full information. We arrive at this opinion on the grounds that even though rights 6 and 7 are closely related to and dependent on each other, right 6 – with the exception of right 6(2) – stands separately from right 7. The exercise of rights 6(1), 6(3) and 6(4) is not necessarily precluded by the exceptions in rights 7(10)(b) and 7(10)(c). We note that none of the rights in the Code is absolute, nor are the rights hierarchically ranked. Hence, where an interpretation of the rights, taken together, can afford maximum potential for the consumer to exercise the rights without conflicting or contradictory outcome, that reading is to be preferred so as to give full effect to the intent of the Code in protecting and promoting consumer rights.
47 The Report of the National Screening Unit sets out criteria for assessing screening programmes and includes reference to a criterion that '[t]he health care system will be capable of supporting all necessary elements of the screening pathway, including diagnosis, follow-up and programme evaluation'. National Screening Unit. Improving quality: A framework for screening programmes in New Zealand, October 2005, Appendix 2, at 32. Available at: http://www.tree.net.nz/dscgi/ds.py/Get/File-6346/NSU_quality_framework4.pdf viewed 18 December 2006.
48 Manning J. Informed consent to medical treatment: The common law and New Zealand's Code of Rights. (2004) 12 Medical Law Review 181, footnote 75, notes Case 01HDC00755.
49 Manning J. Informed consent to medical treatment: The common law and New Zealand's Code of Rights. (2004) 12 Medical Law Review 181, at 215.
50 http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/2a5a1e01dddf6315cc2570c0000bf76c?OpenDocument viewed 18 December 2006.
51 http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/62fc7b26351a0df7cc2570c0000c103a?OpenDocument viewed 18 December 2006.
52 Oral query and response, 24 October 2006.
53 This has also been expressed by the Privacy Commissioner in his report 'Guthrie tests', at para 6.5.
54 Clause 4 of the Code. Note the specific reference to the inclusion of person.
55 Report on opinion – Case 99HDC09011, 13.
56 Report on opinion – Case 99HDC09011, 15.
57 http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/cabf221ef8da9061cc2570c0000c5097?OpenDocument viewed 18 December 2006.
58 http://www.moh.govt.nz/moh.nsf/238fd5fb4fd051844c256669006aed57/5d147a9d39a89eefcc2570c0000c3fa0?OpenDocument viewed 18 December 2006.
59 See Ministry of Health, *Improving quality: A framework for screening programmes in New Zealand*, 2005, 16.
60 See Ministry of Health, *Improving quality: A framework for screening programmes in New Zealand*, 2005, 17.
61 See Ministry of Health, *Improving quality: A framework for screening programmes in New Zealand*, 2005, 28.
62 According to the Health Commissioner, in 2000 the National Testing Centre had been storing the cards for twenty-eight years.

63 In 2003, the Privacy Commissioner reported that the total collection of samples was in the order of 1.9 million. Since September 2003, there have been 175,070 registered births. See: http://www.stats.govt.nz/products-and-services/info-releases/births-and-deaths.htm.
64 DNA profiling is also called DNA fingerprinting. These expressions refer to the techniques used to distinguish between individuals of the same species using samples of their DNA only. DNA profiling was implemented in New Zealand in the late 1980s. See www.esr.cri.nz.
65 At 31. See: ftp://ftp.hrsa.gov/mchb/genetics/screeningdraftforcomment.pdf accessed 18 December 2006.
66 Subject to DNA amplification requirements.
67 Chapter 19, Human Tissue Collections (at 499), in Australian Law Reform Commission Report 96, *Essentially yours: The protection of human genetic information in Australia*, 14 March 2003. Available at: http://www.austlii.edu.au/au/other/alrc/publications/reports/96/.
68 Privacy Commissioner (B.H. Slane), Guthrie tests, 25 September 2003. Available at: http://www.privacy.org.nz/filestore/docfiles/70989185.pdf.
69 Investigations may also be made into false positive results.
70 This may occur in cases where a newborn has died and the health professional wishes to investigate the cause of death or to gain genetic information. Health professionals may also request cards from siblings within a family to investigate the possibility of the presence of genetic disorder within a family. Such investigations can help to inform parents regarding their reproductive options.
71 The blood spot cards of missing persons may be accessed by the Police and Coroners to facilitate the identification of bodies or bodily substances. The blood spot cards of Ben Smart and Olivia Hope were accessed in order to ascertain their DNA profiles for comparison with blood samples found during the Police investigation.
72 For examples of this, see Barbi M., Binda S., Primache V. et al. Use of guthrie cards for the early diagnosis of neonatal herpes simplex virus disease (1998) 17 Pediatric Infectious Disease Journal 251; Gwinn, M., Pappaioanau, M., George J.R. et al. Prevalence of HIV infection in childbearing women in the United States: Surveillance using newborn blood samples (1991) 265 Journal of the American Medical Association 1704.
73 For examples of this, see Burse, V.W., DeGuzman, M.R., Korver M.P. et al. Preliminary investigation of the use of dried blood spots for the assessment of in utero exposure to environmental pollutants (1997) 61 Biochemical and Molecular Medicine 236; Centers for Disease Control and Prevention. Population-based-prevalence of perinatal exposure to cocaine (1996) 45 Morbidity and Mortality Weekly Report 887.
74 For examples of this, see Nelson K.B., Grether J.K., Croen L.A. et al. Neuropeptides and neurotrophins in neonatal blood of children with autism or mental retardation (2001) 49 Annals of Neurology 597; Shaw G.M., Zhu H., Lammer E.J. et al. Genetic variation of infant reduced folate carrier (A80G) and risk of orofacial and conotruncal heart defects (2003) 158 American Journal of Epidemiology 747.
75 For examples of this, see Larsen T.B., Lassen J.F., Brandslund I. et al. The Arg506Gln mutation (FV Leiden) among a cohort of 4188 unselected Danish newborns (1998) 89 Thrombosis Research 211; Crawford D.C., Caggana M., Harris K.B. et al. Characterization of beta-globin haplotypes using blood spots from a population-based cohort of newborns with homozygous HbS (2002) 4 Genetics in Medicine 328; Wilcken B., Bamforth F., Li Z. et al. Geographical and ethnic variation of the 677C>T allele of 5,10 methyl-enetetrahydrofolate reductase (MTHFR): Findings from over 7000 newborns from 16 areas worldwide (2003) 40 Journal of Medical Genetics 619.
76 The term 'analytes' refers to substances undergoing analysis.
77 Personal communication with the Director of the National Testing Centre, 24 October 2006.
78 These metabolites are phenylalanine, leucine, galactose metabolites, biotinidase, 17-hydroxyprogesterone, TSH and immunoreactive trypsin.

79 Therrell B.L., Hannon W.H., Pass K.A. et al. Guidelines for the retention, storage, and use of residual dried blood spot samples after newborn screening analysis: Statement of the Council of Regional Networks for Genetic Services (1996) 57 Biochemical and Molecular Medicine 116, at 117.
80 Ibid, at 117.
81 Chapter 19, Human Tissue Collections (at 499), in Australian Law Reform Commission Report 96, *Essentially yours: The protection of human genetic information in Australia*, 14 March 2003. Available at: http://www.austlii.edu.au/au/other/alrc/publications/reports/96/.
82 See comments by the Council of Regional Networks for Genetic Services: Therrell B.L., Hannon W.H., Pass K.A. et al. Guidelines for the retention, storage, and use of residual dried blood spot samples after newborn screening analysis: Statement of the Council of Regional Networks for Genetic Services (1996) 57 Biochemical and Molecular Medicine 116.
83 Chaisomchit S., Wichajarn R., Janejai N. and Chareonsiriwatana W. Stability of genomic DNA in dried blood spots stored on filter paper (2005) 36 Southeast Asian Journal of Tropical Medicine and Public Health 270.
84 Ibid. The authors note a study done by Cassol et al., which showed that the maximum length of stability of HIV proviral DNA in dried blood spots kept at ambient temperature and desiccated at -20°C was about three-and-a-half months. See Cassol S., Salas T., Gill M.J. et al. Stability of dried blood spot specimens for detection of human immunodeficiency virus DNA by polymerase chain reaction (1992) 30 Journal of Clinical Microbiology 3039. In addition Đglobin was detected by Rubin et al. in dried blood stored at ambient temperature for about a year. See Rubin E.M., Andrews K.A. and Kan Y.W. Newborn screening by DNA analysis of dried blood specimens (1989) 34 Human Genetics 134.
85 Ben Smart was twenty-one when he went missing. Olivia Hope was seventeen.
86 For references see: Elkin K. and Jones D.G. Guthrie cards: Legal and ethical issues (2000) October New Zealand Bioethics Journal 22; Written Question No. 15911 (Sue Kedgley to the Hon. Annette King), *Hansard*, 12 November 2001; Thomas C. Guthrie test samples: Is the problem solved? (2004) June New Zealand Bioethics Journal 25.
87 Right 7(10)(a), Code of Health and Disability Services Consumers' Rights.
88 Right 7(10)(c)(i), (ii) and (iii), Code of Health and Disability Services Consumers' Rights.
89 National Health Committee (National Advisory Committee on Health and Disability), *Screening to improve the health of New Zealand: Criteria to assess screening programmes*, April 2003, 22. Available at: http://www.nhc.govt.nz/publications/PDFs/ScreeningCriteria.pdf.
90 National Health Committee (National Advisory Committee on Health and Disability), *Screening to improve the health of New Zealand: Criteria to assess screening programmes*, April 2003, 9. Available at: http://www.nhc.govt.nz/publications/PDFs/ScreeningCriteria.pdf.
91 Paterson R. (Health and Disability Commissioner) Body parts – Changing the Code of Patients' Rights *NZ Doctor*, 30 June 2004.
92 National Health Committee (National Advisory Committee on Health and Disability) *Screening to improve the health of New Zealand: Criteria to assess screening programmes*, April 2003, 30.
93 See Privacy Commissioner (B.H. Slane) Guthrie tests, 25 September 2003, 5. Available at http://www.privacy.org.nz/filestore/docfiles/70989185.pdf.
94 National Screening Unit, *Improving quality: A framework for screening programmes in New Zealand*, October 2005, 10. Available at: http://www.tree.net.nz/dscgi/ds.py/Get/File-6346/NSU_quality_framework4.pdf viewed 18 December 2006.
95 See Privacy Commissioner (B.H. Slane) Guthrie tests, 25 September 2003, para 4.3. This report by the Privacy Commissioner followed his inquiry into the collection, retention, use and release of newborn metabolic screening test samples, pursuant to s 13(1)(m) of the Privacy Act 1993. Available at: http://www.privacy.org.nz/filestore/docfiles/70989185.pdf viewed 18 December 2006.
96 See Privacy Commissioner (B.H. Slane) Guthrie tests, 25 September 2003, para 4.3. This report by the Privacy Commissioner followed his inquiry into the collection, retention, use and release of

newborn metabolic screening test samples, pursuant to s 13(1)(m) of the Privacy Act 1993. Available at: http://www.privacy.org.nz/filestore/docfiles/70989185.pdf viewed 18 December 2006.

97 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *HGSA policy statement on the retention, storage and use of sample cards from newborn screening programmes*, August 2000, at para 4.3.1. Available at: http://hgsa.com.au/images/UserFiles/Attachments/PolicyStatementontheRetentionofsampleCardsfromNewbornScreeningPrograms.pdf viewed 18 December 2006.

98 Personal communication with the Programme Manager of the Newborn Metabolic Screening Programme, 24 October 2006.

99 See right 7(10)(c)(i), (ii) and (iii) respectively.

100 Paterson R. (Health and Disability Commissioner) Body parts – Changing the Code of Patients' Rights *NZ Doctor*, 30 June 2004.

101 'Bodily material' is used here as a shortened form of 'body parts or bodily substances' from the Code.

102 The question that is formulated here brings together a range of questions, for example:
• How can the exception in right 7(10), which allows QA activities to be undertaken on bodily samples without informed consent, co-exist with the consumer's right to be fully informed?
• How can the consumer's right to be fully informed and the right to require the return or disposal of bodily samples be honoured whilst ensuring that QA activities will not be hampered by the consumer's request for return or disposal of bodily samples?

103 Right 7(9).

104 Right 6.

105 NHC screening guidance (8).

106 We note here that Western Australia explicitly provides for the retention of samples for a period of two years: see http://www.genomics.health.wa.gov.au/publications/docs/NewbornScreen.pdf viewed 22 January 2007.

107 Ministry of Health, *Improving quality: A framework for screening programmes in New Zealand*, 2005, 16.

108 See http://www.hdc.org.nz/complaints/opinions?97HDC8205 viewed 18 December 2006.

109 *New horizons – Consumers rights and private hospitals*, presentation to Annual New Zealand Private Hospitals Association Conference, 10 October 2000. Available at: http://www.hdc.org.nz/publications/presentations?New%20Horizons%20-%20Consumers%20Rights%20and%20Private%20Hospitals.

110 *New horizons – Consumers rights and private hospitals*, presentation to Annual New Zealand Private Hospitals Association Conference, 10 October 2000. Available at: http://www.hdc.org.nz/publications/presentations?New%20Horizons%20-%20Consumers%20Rights%20and%20Private%20Hospitals.

111 Report on Opinion – Case 99HDC09011, 13.

112 Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians, *HGSA policy statement on the retention, storage and use of sample cards from newborn screening programmes*, August 2000, para 4.3.1. Available at: http://hgsa.com.au/images/UserFiles/Attachments/PolicyStatementontheRetentionofsampleCardsfromNewbornScreeningPrograms.pdf viewed 18 December 2006.

113 Informal communication with the NMSP in July 2007 indicated that the issue of documentation and the recording of a refusal by parents to participate in newborn screening, and the policy surrounding this, will be put out for public consultation in the very near future.

114 See http://www.ich.ucl.ac.uk/newborn/ viewed 18 December 2006.