The ethical, legal and social implications of umbilical cord blood banking: Learning important lessons from the protection of human genetic information

David Weisbrot

Internationally networked umbilical cord blood banks hold great promise for better clinical outcomes, but also raise a host of potential ethical and legal concerns. There is now significant accumulated experience in Australia and overseas with regard to the establishment of human genetic research databases and tissue collections, popularly known as “biobanks”. For example, clear lessons emerge from the controversies that surrounded, stalled or derailed the establishment of some early biobanks, such as Iceland’s deCODE, Autogen’s Tonga database, a proposed biobank in Newfoundland, Canada, and the proposed Taiwan biobank. More recent efforts in the United Kingdom, Japan, Quebec and Tasmania have been relatively more successful in generating public support, recognising the critical need for openness and transparency, and ample public education and debate, in order to build community acceptance and legitimacy. Strong attention must be paid to ensuring that other concerns – about privacy, discrimination, informed consent, governance, security, commercial fairness and financial probity – are addressed in structural terms and monitored thereafter, in order to maintain public confidence and avoid a backlash that inevitably would imperil such research. Once lost, credibility is very difficult to restore.

INTRODUCTION

In February 2001, the Australian Government initiated a major inquiry into the Protection of Human Genetic Information, led by the Australian Law Reform Commission (ALRC), in association with the Australian Health Ethics Committee (AHEC) of the National Health and Medical Research Council (NHMRC). The project culminated in the publication of the report Essentially Yours: The Protection of Human Genetic Information in Australia¹ in May 2003.

The terms of reference for the joint inquiry (the Inquiry) directed the ALRC and AHEC to consider, with respect to human genetic information – and the tissue samples from which such information may be derived – how best to protect privacy; to protect against unfair discrimination; and to ensure maintenance of high ethical standards. These three central concerns were then explored across a wide array of actual and potential contexts, including (among others) the oversight of scientific and medical research; public health planning and administration; the delivery of clinical genetic services; law enforcement uses for forensic purposes; insurance underwriting; employment; sport; immigration; parentage and kinship testing; the construction of indigenous identity; the use of

DNA in criminal and civil courts; and the management of genetic registers, tissue banks and human genetic research databases, often referred to as “biobanks”.

The breadth of the undertaking accounts for the “super-sized”, two-volume, 1,200 page final report, which made 144 recommendations for reform, aimed at 31 different “actors”, including federal, State and Territory governments; health and medical policy-makers (such as the NHMRC); human rights, anti-discrimination and privacy officials; regulatory authorities (such as the Therapeutic Goods Administration); insurers; employers and trade unions; medical practitioners; universities and professional education providers.

In order to monitor and coordinate all of this scattered activity, and having regard to the fast pace of change in the science and technology, the report’s central recommendation was the establishment by statute of an independent Human Genetics Commission of Australia. The proposed Commission was modelled on the similarly named body in the United Kingdom, and envisaged as a broad-based agency capable of providing cutting-edge advice to governments, the health sector, industry and the general community about the scientific and technological advances in human genetics (including those “over the horizon”) as well as about the ethical, legal and social implication of these advances. In May 2005, as part of the annual budget process, the Australian Government announced that it had accepted this key recommendation and allocated new funding (initially $7.6 million over four years) to establish an independent, expert Human Genetics Advisory Committee, as a new principal committee of the NHMRC. A “whole-of-government” response to all of the findings and recommendations in Essentially Yours in December 2005 accepted about 90% of the 144 recommendations.

The ALRC report has gained significant attention (and praise) internationally. For example, in his keynote address opening the XIXth International Congress on Genetics in Melbourne in July 2003, Dr Francis Collins, the then Director of the United States National Human Genome Research Institute and head of the Human Genome Project, described Essentially Yours as “a truly phenomenal job … placing Australia ahead of what the rest of the world is doing”. In the years following this landmark report, the ALRC has been involved in detailed discussions about the report’s findings, recommendations and directions with leading government and non-government policy-makers in the United States, Canada, the United Kingdom, New Zealand, China, South Korea, Japan, Taiwan and the Pacific Islands, as well as UNESCO, the World Health Organisation, the OECD, the Human Genome Organisation and many others.

While the ethical, legal and social implications of emerging genetic science and technology have received this high level of attention and policy development in Australia and overseas, the advent of umbilical cord blood banking has prompted far less reflection, regulation and policy activity. To some extent, this must be because of the differences in scale. Every child born in Australia for the last 50 years receives a neonatal genetic test (the “Guthrie” or “heel prick” test), and very large amounts of public funding are allocated to human genetic research and clinical practice. By way of contrast, only about 3,300 umbilical cords are banked in Australia each year: less than 1% of the potential total, because of limited public funding, practical constraints and a general lack of knowledge about the procedure.

Nevertheless, there are more than 50 umbilical cord blood banks operating across 26 countries, storing about a half-million units of cord blood, with a fully networked international system for

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4 Dr Collins was appointed head of the National Institutes of Health by United States President Barack Obama in 2009.


locating and transporting matching samples for non-autologous cord blood transplants. Most of this activity will raise the same range of ethical, legal and social concerns as human genetic research and biobanking, so it is worth considering the lessons that have emerged from the inquiries into the protection of human genetic information and their application (or otherwise) to cord blood banking.

**PUBLIC CONSULTATION**

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

From the outset, the Inquiry acknowledged the critical need for public engagement and widespread consultation, involving the general community as well as experts and interest groups. Arguably, a deep commitment to extensive community consultation as an essential part of research and policy development is the defining characteristic of a modern law reform commission, and it is certainly the case that such consultation is firmly part of the ALRC’s institutional DNA. Ultimately, that is the attribute which distinguishes it from other bodies (think tanks, research institutes, parliamentary committees) with a law reform aspect to their work. Opeskin has noted:

The desirability of engaging the public in the process of law reform may be explained in many ways. For convenience, these can be divided into three groups: benefits for those consulted; benefits for the process of law reform, and benefits in terms of enhanced effectiveness of the law once reformed.

From its earliest days, under the leadership of the foundation Chair, Justice Michael Kirby, the ALRC entrenched the active engagement of the community as part and parcel of its basic approach to law reform, under the rallying cry that “law reform is much too important to be left to the experts”. The ALRC acknowledged that consultation had become an attribute of organised law reform since the creation of the English Law Commission in 1965; however, the ALRC committed itself to taking the “extra step” of ensuring, as a matter of basic philosophy, that “law reform should be conducted in a transparent way with opportunities for widespread public consultation”.9

The “traditional” approach of law reform commissions and other inquiries to consultation generally has followed a predictable – and in earlier times, a relatively successful – pattern. This involved the production of consultation papers; holding hearings, at which interested parties could provide oral evidence; and the solicitation of written submissions from stakeholders. However, it is now generally recognised that law reform and policy development are increasingly crowded fields, with a constant stream of parliamentary inquiries, departmental and interdepartmental inquiries, task forces, royal commissions, green papers and the like – so that “submission fatigue” is a very real phenomenon. It is no longer sufficient for a law reform commission to publish an issues paper or discussion paper, perhaps schedule a few public hearings, and then sit back and wait for the raft of comprehensive, thoughtful and beautifully crafted submissions from stakeholders to flow in.

Consequently, in the *Essentially Yours* Inquiry, the ALRC produced an Issues Paper and a Discussion Paper, both consciously designed to promote public education and debate; conducted 15 public forums around Australia in all capital cities and the major regional centres; conducted about 250 “targeted” meetings with key stakeholders and community organisations in Australia and overseas; and received and considered over 350 written submissions, ranging from those prepared by

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7 O’Connor et al, n 6.
peak associations to those from individuals and families relating their own personal stories. Similarly, in the subsequent ALRC inquiry into Australian privacy law and practice conducted between 2006-2008, which spent considerable time and energy on health privacy, the ALRC also held about 250 meetings, roundtables and workshops, and received over 600 written submissions.

The ALRC’s experience in dealing with the Australian public confirms the local and overseas literature on social attitudes with regard to the rise of the “New Genetics” and scientific and technological advances and challenges: that is, there are strong, but conflicting, feelings in the community about biotechnology. On the one hand, there is considerable optimism about the potential for research to lead to the development of whole new fields of medicine, such as gene therapy, regenerative medicine, and pharmacogenomics, as well as to produce important medical breakthroughs in the diagnosis, treatment and prevention of some terrible debilitating diseases, such as diabetes, cancers, Alzheimer’s and Huntington’s disease. There is also strong support in the general community for the use of DNA technology by law enforcement authorities, now squarely a part of popular culture through the many television police dramas built around this subject.

At the same time, there is an underlying general anxiety in the community about the rapid pace of change: concerns about the loss of control; fears about the beginnings of “genetic determinism” or perhaps even more radical eugenics; and nagging doubts about the capacity of public authorities to regulate this area effectively in the public interest. The specific concerns that emerged, and the suggested responses, are considered below.

**THE RECENT EMERGENCE OF “BIOBANKS”**

Following the success of the Human Genome Project in fully sequencing the human genome, researchers are now mainly endeavouring to compile large collections of genetic samples and related genetic and other health information, to look for associations and to aid studies into the causes of disease, drug reactions and environmental interaction with genetic status.\(^{13}\)

Over the last decade, human genetic research databases, now commonly referred to as “biobanks”, have gained increasing importance as the multidisciplinary field of bioinformatics develops, with computer science, developmental biology and mathematics combining to produce tools that enable the storage and analysis – at very high speeds by supercomputers – of large quantities of biological, particularly genetic, information.\(^{14}\)

The successor to the Human Genome Project, the International Haplotype Mapping (HapMap) Project,\(^{15}\) also operated on the basis of an international consortium committed to making all of its results available on a public access database, subject to an instant “clickwrap” licence to protect against “parasitic” patenting of this material.\(^{16}\)

Australia does not currently have – nor are there imminent plans to construct – a population-wide or comprehensive, national biobank,\(^{17}\) such as has been established in Iceland (deCODE), Estonia (the Estonian Genome Project), the United Kingdom (UK BioBank), Quebec (CartaGene), Japan (Japan BioBank), and elsewhere.\(^{18}\)

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\(^{14}\) To provide just one example of the sort of advances that are being made in this area, in 2003 the Australian media reported that a PhD student at the University of New South Wales was successful in making correlations among 15,000 mouse genes. This took a supercomputer (shared by a number of universities) 32 hours; a standard desktop computer would have taken 5,700 years to complete the same calculations: Mackenzie K, “5700-year Gene Puzzle Cracked”, The Australian (25 November 2003) p 29.


\(^{17}\) “HGRD” (human genetic research database) is the term used by the OECD, although “biobank” is also widely understood. This article does not deal with the DNA identification databases operated by all of the federal, State and Territory police forces, and used for criminal investigation and victim/missing person identification, all of which are established under legislation: see the Crimes Act 1914 (Cth), Pt 1D; Crimes (Forensic Procedures) Act 2000 (NSW); Crimes (Forensic Procedures) Act 2000.
However, there are State-wide projects in development in Western Australia and Tasmania involving the collection of samples from volunteers on a large-scale basis, as well as extensive data linkage wherever possible (and lawful). In Tasmania, the Menzies Research Institute (formerly the Menzies Centre for Population Health Research) maintains a research database comprising extensive genealogical data (eg, births, deaths and marriages records), genetic samples, and health information supplied by donors, in order to search for genetic causes of disease. All biological samples are donated by volunteers specifically for the Centre’s research projects, and formally consented to on this basis.\textsuperscript{19}

The University of Western Australia Institute for Medical Research (WAIMR) announced in August 2006 that it was leading a team proposing to create a State-wide biobank,\textsuperscript{20} with links to the Public Population Project in Genomics (P3G), an “international consortium for the development and management of a multidisciplinary infrastructure for comparing and merging results from population genomic studies”.\textsuperscript{21} The WAIMR initiative expressly recognised the critical importance of safeguarding privacy, calling for “national privacy legislation and beefed up genetic data protection laws, in line with the recommendations of the Australian Law Reform Commission’s \textit{Essentially Yours} report”.\textsuperscript{22}

The Peter MacCallum Cancer Institute in Melbourne established a tissue bank in 1998 in order to facilitate a number of molecular genetic studies in cancer. Cancerous tissue is obtained from patients via surgical and pathology staff, who liaise with the tissue bank. Patients give broad consent to the storage and use of their tissue in research, and the tissue remains identified to allow continued collection of clinical information that will be used in conjunction with the tissue sample as part of ongoing research.\textsuperscript{23} The Queensland Institute for Medical Research (QIMR) has also developed a major database of genetic samples taken from twins, which is used for a range of medical research purposes.\textsuperscript{24}

Of course, apart from the budding biobanks described above, there are a large number of smaller or unsystematised human genetic research databases in existence in Australia, maintained by universities, research centres and biotechnology companies; public and private hospitals (eg tissue banks, blood banks, pathology samples, paraffin blocks); pathology laboratories; and familial cancer registers,\textsuperscript{25} as well as, for these purposes, public and private umbilical cord blood banks.

Although not yet a significant feature of Australian research culture, donor tissue banks are being constructed on a commercial basis in the United States. Such banks collect tissue from hospitals and

\footnotesize{(ACT); Criminal Investigation (Identifying People) Act 2002 (WA); Forensic Procedures Act 2000 (Tas); Crimes Act 1958 (Vic); Criminal Law (Forensic Procedures) Act 1998 (SA). This area was covered in considerable detail in ALRC, n 1, Pt J, Chs 39-45.


19 The Menzies Research Institute is affiliated with the University of Tasmania; see its homepage at http://www.menzies.utas.edu.au viewed 29 September 2011


21 See the P3G website http://www.p3g.org/secretariat/ viewed 29 September 2011: the founding members of the consortium include CartaGene, the Estonian Human Genome Project and an eight-country EU-based biobank but there now also other members from the United States (the National Institutes of Health and the Center for Disease Control), Canada, Spain, the United Kingdom, Germany, Sweden, and a Central European group.

22 Skatssoon, n 20.

23 Submission of the Peter MacCallum Cancer Institute to the ALRC’s Inquiry into the Protection of Human Genetic Information, Submission G071 (7 January 2002).


process them ready for research. The banks themselves do not conduct research, but sell processed tissue to researchers at other institutions. One example is Gene Logic, a United States tissue repository containing more than 10,000 tissue samples that are made available to researchers and pharmaceutical companies.26 By way of contrast, the Perth Bone and Tissue Bank, established in 1993, operates as an independent, not-for-profit charitable organisation which collects, screens, stores and distributes donated human bone and tissue grafts for transplant purposes.27

Umbilical cord blood banking in Australia has always featured both public and private entities, and while the entrepreneurial drive to establish private blood banks initially attracted controversy, it now appears that the public-private distinction is blurring in the more mature and cooperative, if still small, market.28

Perhaps of most potential importance, there exists a major inchoate national biobank in Australia, in the form of vast numbers of newborn screening blood spot cards (aka “Guthrie cards”), stored in children’s hospitals around the country (with varying degrees of care) – representing a complete, if unorganised, DNA collection of almost everyone born in Australia in the last 50 years. Guthrie cards are regarded as “health records”, and thus issues relating to storage, access and disclosure are generally governed by privacy legislation.29 The Australian National Pathology Advisory Committee Guidelines recommend retention of the cards for 25 years, but there is no active program of removing or destroying older cards, while the policy in one State (Western Australia) is to destroy the cards after two years.

In Essentially Yours, the ALRC called upon the various health ministers and other key stakeholders (such as the NHMRC) to develop nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, newborn screening cards and other human tissue collections, including collections of pathology samples and banked tissue.30

Apart from getting the technical detail right and ensuring a greater level of consistency, the ALRC noted that any new rules governing the collection, storage and use of human tissue collections should contain policies about whether and how to facilitate population genetics. The use of such materials, perhaps including Guthrie cards, for epidemiological and research purposes would have beneficial society-wide (and perhaps international) effects in terms of scientific advances and public health planning and administration. Of course, this may mean some trade-offs against privacy protection for individuals – and is that a price most Australians would be happy to pay?

Accordingly, the ALRC called for an informed “national discussion” about how to proceed, in which “it will be important to identify and balance the relevant ethical considerations, including those applying at individual and community levels”;31 in other words, shifting from the traditional dominance of individual rights and interests, as encapsulated by the doctrine of informed consent, towards a more community-focused approach with respect to population genetics. This matter was

29 However, it is less clear what the status of the blood sample is once it is removed from the card: the blood/DNA no longer constitutes a “health record”, and would no longer be governed by the Human Tissue Acts (or, apparently, any other current law).
31 ALRC, n 1 at [19.104].
taken up again by the ALRC in 2008, in its comprehensive report on Australian privacy law and practice, which considered health privacy and research matters, among many other things.

The existing regime in Australia provides that health information may be collected, used and disclosed where necessary for research or the compilation or analysis of statistics, relevant to public health or public safety where:

- the purpose cannot be served by the collection of information that does not identify the individual;
- it is impracticable for the organisation to seek the individual’s consent to the collection, use or disclosure;
- the information is collected, used and disclosed in accordance with rules approved by the Privacy Commissioner;
- in the case of disclosure, the organisation reasonably believes that the recipient of the health information will not disclose the health information, or personal information derived from the health information; and
- the organisation takes reasonable steps to permanently de-identify the information before it discloses it.

The ALRC received strong submissions from the NHMRC, the Cancer Council and others suggesting that the current test, especially the requirement that the public interest in the research must “substantially outweigh” the interest in privacy protection, was resulting in overly cautious decision-making by Human Research Ethics Committees (HRECs), and thus the frustration of potentially important research. The ALRC ultimately agreed, and recommended dropping the modifier “substantially”, so that the test would become simply one of determining whether the balance of public interests favours the research proceeding. Unfortunately, while the Australian Government’s initial response to For Your Information in October 2009 was overwhelmingly favourable, accepting about 90% of the first tranche of 197 recommendations that it considered, the government effectively did not accept this one – but provided no commentary about why it came to a different conclusion. Thus, we have yet to have in Australia the necessary intelligent national discussion about how we wish to balance individual privacy with the public interest in facilitating promising, and otherwise ethically sound, medical and scientific research.

For the purpose of ensuring ethically sound and popularly accepted cord blood banking operations, it is also worth exploring, and seeking to derive salutary lessons from, some failed biobanking initiatives.

Perhaps the first and most notorious venture in this area was the similar project which has already been undertaken in Iceland. A private company called deCode Genetics was awarded an exclusive licence by the Icelandic Government to create and operate a database of genetic and other health information, known as the Iceland Health Sector Database. The deCode project brought together three

33 National Privacy Principles (NPP) 2 and 10, operating in combination.
34 National Health and Medical Research Council, Submission PR 114 (15 January 2007); ALRC, n 32 (see at [65.74]).
35 ALRC, n 32 (see Recommendation 65-4).
36 Oddly, the government chose formally to “accept with amendment” Recommendation 65-4, the amendment being to reinstate the phrase “substantially outweighs”, which directly reverses the intention of the recommendation. The response states that the “test should be that the Human Research Ethics Committee is satisfied that the public interest in the research activity substantially outweighs the public interest in maintaining the level of privacy. The requirement of substantiality ensures that there is a clear balance in favour of the research activity progressing. Such clarity is appropriate in circumstances where individuals’ personal information will be handled without their consent”: see Department of Prime Minister and Cabinet, Enhancing National Privacy Protection: Australian Government First Stage Response to ALRC 108 (October 2009) p 140, http://www.dpmc.gov.au/privacy/alrc_docs/stage1_aus_govt_response.pdf viewed 29 September 2011.
types of information: coded health information taken from Iceland’s national health care system records; genealogical data; and genetic information from samples obtained and analysed with the consent of Icelandic donors.37

Once it became better known, the project met with widespread public concern, particularly because:

• this was a top-down project, with little or no community consultation in the design of the ethical or governance systems, and little community education about the privacy and other safeguards to be deployed;

• it involved an exclusive licence for access to medical and genetic information, and to genetic samples;

• a private commercial entity was being given control over personal and sensitive human genetic information;

• the private company would apparently be free to patent its subsequent inventions – genetic tests, pharmaceuticals and so on – based on the use of the national database;

• there was considerable concern that the prospective commercial benefits would not flow to the general population in Iceland in an equitable fashion, nor were people convinced that they would benefit from any greater access to the medical and scientific breakthroughs that might emerge; and, indeed,

• deCODE had itself signed an exclusive licence for commercialisation and development with a single pharmaceutical company.

The Estonian Genome Project, established around the same time, shared some of the features of its Icelandic counterpart, but also contained a number of important features designed to protect the public interest to a higher degree. Estonia utilised a public, non-profit, foundation as its basis, rather than a private company. It also proposed to develop a centralised electronic database of the genetic information of about three-quarters of the country’s population, including blood samples and health data information, for use in large-scale association studies. Control of the database was vested in the government-owned Estonian Genome Project Foundation and EGee, an exclusive commercial licensee that provides most of the finances for the project.38

It is also useful to examine a highly publicised but unsuccessful early attempt to establish an Australian-controlled biobank in the Pacific. In November 2000, the biotechnology company Autogen informed the Australian Stock Exchange that it had signed an agreement with the Kingdom of Tonga, “using the unique population resources in the Kingdom of Tonga” to establish a major health database to identify genes that cause common diseases. It was noted that the “unique family structure and isolation of this population together with the high prevalence of a variety of diseases represents a major resource for geneticists”. Media reports also indicated that Autogen was looking to expand this initiative to include other countries overseas, as well as the Australian State of Tasmania.39

Although Autogen’s statement on ethics emphasised the prior informed consent of individual Tongan volunteers, and the company promised to provide needed health care facilities, the project soon ran into an international storm of protest and did not proceed. Among other things, there were serious concerns raised within Tonga and outside about the secrecy of the negotiations with a poor and vulnerable island state that is the region’s only remaining full monarchy; the potential for

“bio-piracy”, with the islanders’ genetic resources exploited without fair value; and the difficulty of obtaining truly informed consent from individuals, given the powerful and hierarchical communal social structure.40

In 2005, Taiwan proposed establishing a national biobank on a pilot basis, alert to the concerns that had emerged over the Icelandic experience and determined to build in a sound ethical framework emphasising informed consent, privacy and non-discrimination. From a medical point of view, researchers were excited that, apart from the substantial Han Chinese population that came from the mainland, Taiwan’s significant indigenous population had genetic links with peoples in southern Asia and other parts of the Asia-Pacific region. This held out the promise that medical breakthroughs occasioned by a Taiwan biobank would have health benefits across a huge population – and thus to the commercial promise to justify the expenses involved. However, the Taiwanese authorities substantially under-estimated the concerns of the already socially disadvantaged indigenous people about the capture and use of their genetic information. Absent an extensive process of community consultation – and especially absent a major education and trust-building exercise – it was decided the following year to shelve the project until there was greater consensus in support of such an exercise.

In light of these experiences, UK Biobank was consciously established on the basis of placing a high premium on ethics and governance. UK Biobank is a publicly funded, charitable company limited by guarantee, with the aim of collecting samples and health information from 500,000 volunteers.41 The project aims to provide sufficiently comprehensive health information and a large enough sample size to enable more effective studies of the interactions between genotype and environment. Participants will supply updated medical and environmental exposure information every five years. UK Biobank will not conduct any research itself, but rather will develop and maintain the database, and afford access to researchers following application and approval from an oversight body. Great care and attention has gone into developing channels of community consultation, ensuring a high level of ethical oversight, explicitly detailing access and equity issues, and ensuring sound governance arrangements.42

Similarly, it is now the case that any database or tissue collection built around the collection, storage and use of human tissue, including umbilical cord blood, will have to be very attentive to these issues, or risk public censure (or worse).

THE REGULATORY FRAMEWORK

In recent years the international community has been turning its attention to the protection of human genetic information and “genetic rights”. The UNESCO Universal Declaration on the Human Genome and Human Rights 1997 recognises that:

- research on the human genome and the resulting applications open up vast prospects for progress in improving the health of individuals and of humankind as a whole, but … that such research should fully respect human dignity, freedom and human rights, as well as the prohibition of all forms of discrimination based on genetic characteristics.43

While the Declaration is not a binding legal instrument, it is evidence of growing international concern and an indication of the general approach of the international community in this area. Article 2 of the Declaration states:

Everyone has a right to respect for their dignity and for their rights regardless of their genetic characteristics. That dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity.

41 See UK Biobank, http://www.ukbiobank.ac.uk/ viewed 29 September 2011; Medical Research Council and the Wellcome Trust, Draft Protocol for BioBank UK: A Study of Genes, Environment and Health (1 February 2002) at [2.3.1].
Article 6 goes on to declare:

No one shall be subjected to discrimination based on genetic characteristics that is intended to infringe or has the effect of infringing human rights, fundamental freedoms and human dignity.

The Council of Europe’s Convention on Human Rights and Biomedicine, which is a legally binding instrument and has been signed and ratified by 15 countries to date, gives a clear indication of the approach adopted in Europe in relation to this issue. Article 11 states:

Any form of discrimination against a person on grounds of his or her genetic heritage is prohibited.44

It is against this background that the ALRC was asked to consider whether the protection offered by existing legislation in Australia is adequate. The current methods of regulation and conflict resolution in this field in Australia involve the typical patchwork of federal, State and Territory laws; official guidelines; personal and professional ethics; institutional restraints; peer review and pressure; oversight by public funding authorities and professional associations; supervision by public regulatory and complaints-handling authorities; occasional media scrutiny and exposés; private interests; and market pressures.

The complexity of Australia’s federal system dramatically adds to the difficulties in describing, much less reforming, law and practice in this field. For example, the Federal Government and every State and Territory have enacted statutes – sometimes more than one – which touch on privacy interests in biological materials and health information. Similarly, there are nine different legislative regimes governing forensic procedures involving the collection, analysis and databasing of DNA profiles and other genetic information for law enforcement purposes, as well as many protocols and agreements aimed at facilitating the sharing of such information.

Adequacy of ethical oversight procedures

The NHMRC is the authority that promulgates the principles and procedures applicable to medical research and ethical matters relating to health in Australia, in accordance with the National Health and Medical Research Council Act 1992 (Cth). In 1999, the NHMRC issued the National Statement on Ethical Conduct in Research Involving Humans (the National Statement)45 – the “Bible” on ethical research in Australia – which was developed by AHEC after substantial public consultation.

The National Statement has been endorsed by all of the major stakeholders, including all Australian university heads, the Australian Research Council (which, besides the NHMRC, is the major research grants body), the Australian Academy of the Humanities, Academy of Science, and Academy of the Social Sciences.

Briefly, the National Statement

• contains ethical principles relevant to all research involving humans;
• requires that particular matters are to be addressed when research involves vulnerable persons – children and young people, persons with an intellectual or mental impairment, persons highly dependent on medical care, those in dependent or unequal relationships, collectivities, and Aboriginal and Torres Strait Islander people;
• requires that specific matters be addressed in the consideration and approval of research involving radiation, assisted reproductive technology, clinical trials, epidemiology, human tissue samples, or genetics; and
• sets out the formation, membership and functions of HRECs.

Besides the National Statement, the ethical framework for conducting research in Australia includes such important references as:


• the Human Genetics Society of Australasia’s *Guidelines for Human DNA Banking* (the HGSA Guidelines);\(^{46}\)

• standards for the scientific validity of research, notably the *Statement and Guidelines on Research Practice* issued by the NHMRC and the Australian Vice-Chancellors’ Committee;\(^{47}\) and

• NHMRC guidelines applying to specific types of research, such as gene therapy.\(^{48}\)

The Inquiry noted that a significant loophole exists in theory, insofar as compliance with the National Statement is only compulsory for “public” research, with no equivalent requirement for purely “private” research. However, virtually all human genetic research in Australia is either publicly funded to some degree or is carried out at least partially in publicly funded institutions (universities, public hospitals etc), so there is little or no non-compliance in practice. Further, researchers seeking publication of their results, and companies seeking formal recognition of their clinical trials by regulatory authorities, are unlikely to jeopardise this by avoiding the requirements of the National Statement.

The National Statement provides that research proposals involving human participants must be reviewed and approved by an HREC, whether this involves medical and scientific research or non-invasive social science research. The ALRC, operating the Inquiry in conjunction with the Australian Health Ethics Committee, spent considerable time analysing the adequacy and effectiveness of HRECs, whose primary function is to protect the welfare and rights of participants in research – but also explicitly have a secondary purpose under the National Statement, which is to “facilitate research that is or will be of benefit to the researcher’s community or to humankind”.\(^{49}\)

Responding to earlier criticisms about the perceived lack of independence of the earlier mechanism (institutional ethics committees), at least half the membership of an HREC must now be external to the institution, and must include a lawyer; a member with knowledge and experience in the relevant research area; a member with knowledge and experience in care and counselling; a minister of religion or a person who plays an equivalent role in the community (such as an Aboriginal Elder).

There is now a reasonable measure of independence, and sufficient incentives (especially in relation to public funding) to ensure a high degree of compliance. However, the ALRC identified a number of significant deficiencies, including that many HRECs are overloaded and under-resourced; there is a need for induction and training programs for members; there is a need for more accountability and transparency of processes; more meaningful monitoring needs to be done; multi-centre research, increasingly common, is handled in a very inefficient manner; and there is insufficient independence from the institution in some cases.

As a consequence, *Essentially Yours* made many recommendations directed to the NHMRC and aimed at strengthening the mechanisms through which compliance with the National Statement is enforced, including taking steps to better support the work of HRECs, and induction programs for new members and opportunities for “upskilling” of continuing members. In order to promote transparency and consistency, and permit effective monitoring by AHEC, the Inquiry recommended that new guidelines be developed and obligations imposed on HRECs to report on waivers granted (in accordance with provisions in the National Statement) to researchers for the use of genetic samples and information in medical research without the consent of the individual(s) who provided the original sample. The Inquiry also called for the NHMRC to develop a quality improvement framework for HRECs, and to consider the introduction of a formal accreditation system for HRECs.

Although AHEC has overall responsibility for monitoring the effectiveness of the system, there is no overarching approval framework and in essence, this is a decentralised, collegial system, with the expected strengths and weaknesses. There are about 220 HRECs in Australia (far higher than in


\(^{49}\) Preamble to the National Statement.
Germany, say, with its larger population) and there is a flat structure: unlike Japan, eg, there is no central body that reviews approvals or hears appeals from refusals. On the positive side, the Australian system is low cost, largely voluntary, and flexible, tapping local expertise and knowledge of the institution – and low cost is a particular virtue in a system that generally does not allocate research funds to cover compliance with regulatory requirements.

**Adequacy of informed consent procedures**

The theory and practices surrounding “informed consent” to medical procedures have long been vexed questions in bioethics, law and medicine. All researchers understand – and HRECs now require as a condition of approval – that special care must be taken to ensure that those providing tissue to a biobank, as with participants in any research project, have provided informed consent.

As a matter of practice, this usually involves signing a consent form, following perusal of a document that sets out the nature of the research project, any medical risks involved, potential benefits and outcomes, and other material relevant for an individual making a full, free and informed decision to participate. The quality of the documentation has improved markedly in recent years – but it has also lengthened accordingly, raising a host of other questions about whether consent truly can be meaningful in the circumstances.

When human genetic research reveals information important to the future health of an identified or potentially identifiable participant or her or his offspring, the research protocol must provide for the same consent, counselling and confidentiality protection as would apply in a clinical setting.

In the particularly dynamic contexts of human genetic research and biobanking, it is now common practice for researchers to seek consent from donors for their tissue to be used in a particular experiment and then stored for possible later inclusion in other experiments, the details and potential implications of which are unspecified. This practice has been criticised in some quarters as inappropriate because, as a matter of principle, valid consent cannot be provided in the absence of full and complete disclosure of the specific uses to which the tissue is to be put.

However, there is also broad consensus within the medical research community about the need for procedures under which prospective research participants may provide consent for unspecified future research, provided that such research is carefully scrutinised and approved by an HREC – which may, in some circumstances, require researchers to go back and obtain fresh consent from the donors.

Suggestions for appropriate procedures often focus on the idea of “tick a box” consent options; that is, prospective research participants could select from a graduated set of consent options, so that in addition to participants being asked to consent to participation in the specified research study, they also could opt to have their tissue or information stored for “related” or “unrelated” future research, and designate whether future use would, or would not, require fresh consent. Similarly, it is sometimes suggested that the language of consent should be augmented by the concept of “donation” or “gifting”; that is, altruistic individuals may wish to “gift” samples for research purposes without condition, eg by “leaving their body to science” or donating blood to the Red Cross.

The ALRC recommended that the NHMRC develop the National Statement to provide express ethical guidance on biobanking and, given the particular nature and patterns of genetic research, to offer ethical and practical guidance on obtaining consent to unspecified future research.

The ALRC heard concerns, from members of the community and from experts, about the effectiveness of simple “de-identification” practices (such as the use of pseudonyms) as a tool for protecting privacy, especially in relation to human genetic research databases and biobanks, since these often contain a large amount of information (birth date, marital status, postal code, hospital admissions, etc) that can be cross-matched and linked back to an individual.\(^5\) The ALRC also recommended that the National Statement provide guidance on the use and relative efficacy of such strategies as anonymisation, de-identification and encryption to protect the privacy and confidentiality

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of volunteers; and “gene trustee” or “genetic ombudsman” mechanisms, designed to ensure there is an independent element in the process to increase privacy protection and oversee the integrity of consent agreements.

These aims are now substantially achieved in the 2007 edition of the NHMRC’s National Statement, which incorporates a number of recommendations made in Essentially Yours, including improved waiver of consent and unspecified consent provisions, clearer guidance on identifiable, re-identifiable and non-identifiable data, more stringent conditions for the establishment and running of biobanks and genetic registers, and ethical issues specific to genetic research.

However, more work is still needed and creative thought must be given to adapting the traditional approaches to “informed consent” to the new circumstances and exigencies of the genomics era. Wjst et al have argued persuasively that informed consent can no longer remain “a one-off affair”, with the nature of the genomic research now requiring the addition of a temporal dimension: as long as researchers have access to an individual’s DNA, they should be obliged to maintain an ongoing relationship and interactions. Rather than a single broad consent, they propose a “circular process of information exchange” involving public presentations, feedback from research subjects, and information being provided by researchers to subjects (if desired) which can be facilitated through the clever use of information and communication technology, such as email, mobile phone messaging, regularly updated Web pages and social networking websites.

It is evident that many of the same informed consent issues and controversies will be in play in the umbilical cord blood banking area. Among other things, can consent truly be said to be voluntary in the highly charged and exhausting period of late pregnancy through to childbirth? How much information should be provided about the value of umbilical cord blood banking to the individuals concerned, including the evidence base underpinning the proven clinical effectiveness of autologous cord blood transplants? How much detail should be provided about the rigour of the particular cord blood bank’s security and privacy protections? Is there a further set of assurances that a private cord blood bank should make beyond the standard information provided by a public cord blood bank? Who controls access to the stored material in the event that the parents die, or the marriage dissolves, or a biological relative desperately requires access for medical purposes? Can law enforcement officials gain access to and test material as part of a criminal investigation? Could a private investigator gain access, eg, in an attempt to establish (or refute) paternity claims? Can a cord blood bank send material overseas, where it might be stored at a lower cost? What happens to the stored material if a private cord blood bank goes into receivership or liquidation?

Adequacy of Governance Arrangements

In Essentially Yours, the ALRC recommended that there should be management requirements for biobanks in relation to:

- the nomination of a database keeper/custodian who will have clear responsibility for the day-to-day operation of the database;
- compliance with standards for the collection, use, storage, disclosure and transfer of genetic samples and genetic information held by the database;
- annual reporting to the institutional HREC and AHEC on database operations; and
- provision for audit of the database and its operations, on request by the institutional HREC and/or AHEC.

51 In particular, ALRC, n 1, Recommendations 15-1, 15-2, 15-4, 16-1, 17-1, 18-1, 18-2 and 18-3.
52 See especially ss 2.2, 2.3 and 3.2 of the National Statement, as revised, on the general requirements for consent and for qualifying or waiving conditions for consent, and on how these matters should be applied in the specific context of databanking.
54 Cf NHMRC, Guidelines for Genetic Registers and Associated Genetic Material (2000) at 2.1(e).
In addition, guidance could be provided on a range of other matters relating to the operation of human genetic research databases including:

- governance structures, including guidance on the appropriate relationships between the institution, database custodian and the institutional HREC;
- ethical approval processes where external researchers seek access to samples and information stored on the database; and
- guidance on the appropriate use of independent intermediaries to hold codes linking genetic samples or information with identifiers (see also below).

By way of analogy, the Inquiry also made a number of recommendations aimed at strengthening the independent oversight and accountability of DNA databases used for law enforcement purposes. For example, it recommended that the Commonwealth Ombudsman and the Federal Privacy Commissioner become members of the board that oversees the operation of the National Criminal Investigation DNA Database (NCIDD).

As part of the package of suggested changes to the National Statement, the ALRC recommended that the NHMRC establish and administer a public register of human genetic research databases, develop conditions of registration, and include provisions so that no genetic research under the National Statement can be conducted using information from a database unless it is duly registered. The Inquiry concluded that a system for the registration of human genetic research databases would be capable of providing greater transparency and accountability in the operation and use of such databases, without subjecting institutions to onerous compliance costs.

Registration would oblige institutions to identify and regularise the research collections that they currently maintain and ensure that the operation of these collections is subject to appropriate governance structures and proper institutional and HREC scrutiny. Registration would mean that, for the first time, comprehensive information would be available to AHEC, and possibly to the public, about the number and type of research databases, and the kinds of research being conducted using the samples and information they contain. Such transparency may become increasingly important in allaying public concerns about the privacy, ethical and other implications of the continuing development of research databases. Registration also would provide AHEC with the information necessary to enable it to properly advise the NHMRC, and provide guidance to HRECs, on ethical issues relating to the operation of human genetic research databases.

Again, all of these issues and concerns are echoed with respect to the governance of umbilical cord blood banks, where matters of independence, probity, openness and accountability, good management and the robust protection of clients’ personal and financial interests are paramount to the maintenance of public support and the continued viability of the industry as a whole.

**STATUTORY AND OTHER LEGAL PROTECTIONS**

Beyond ethical regulation, the collection, storage, use and disclosure of genetic samples and information held in biobanks are regulated by a mixture of legislation, guidelines and standards. These include (among other things):

- the legislative framework for the protection of information and health privacy based on the federal Privacy Act 1988 (Cth) and similar State and Territory legislation; and
- guidelines and regulations made in accordance with the Privacy Act, including those promulgated by the federal Privacy Commissioner.

55 The Inquiry considered, but ultimately did not favour, a more heavy-handed licensing system. The registration approach is meant to be “light touch”, with registrants obliged to lodge a form with the appropriate details and showing that all ethical oversight requirements have been met (eg, HREC approval), rather than being put through a more elaborate or contested process.

56 Such as the Privacy and Personal Information Protection Act 1998 (NSW); Health Records and Information Privacy Act 2002 (NSW); Health Records Act 2001 (Vic); Information Privacy Act 2000 (Vic); Health Records (Privacy and Access) Act 1997 (ACT).

the State and Territory Human Tissue Acts, which govern consent for the donation of human tissues for research, and subsequent use;

• the federal, State and Territory laws prohibiting unlawful discrimination that (arguably) may extend to conduct based on a person’s real or perceived genetic status, such as the federal Disability Discrimination Act 1992 (Cth); and

• the common law duty to exercise reasonable care, which is owed by researchers, research organisations and HRECs to participants in research.

Privacy protection

The Privacy Act 1988 (Cth) is intended to protect the personal information of individuals and to give them control over how that information is collected, used and disclosed. The legislation sets out certain safeguards that government, private sector organisations and individuals must observe, and also gives individuals rights to access and correct their own personal information.

Under s 6(1), “personal information” is defined as

information or an opinion (including information or an opinion forming part of a database), whether true or not, and whether recorded in a material form or not, about an individual whose identity is apparent, or can reasonably be ascertained, from the information or opinion.

For the purposes of private sector coverage (see below), the Privacy Act also creates a special category of “sensitive information” and gives this a higher level of protection. Sensitive information is defined in s 6 as

information or an opinion about an individual’s racial or ethnic origin; political opinion; political association membership; religious beliefs, affiliations or philosophical beliefs; professional or trade association membership; union membership; sexual preferences; criminal record; or is health information about an individual.

“Health information” is separately defined in s 6 as

(a) information or an opinion about

(i) the health or a disability (at any time) of an individual; or

(ii) an individual’s expressed wishes about the future provision of health services to him or her; or

(iii) a health service provided, or to be provided, to an individual, that is also personal information; or

(b) other personal information collected to provide, or in providing, a health service; or

(c) other personal information about an individual collected in connection with the donation, or intended donation, by the individual of his or her body parts, organs or body substances.

In keeping with the pattern of legislation in most of the Western world, the Privacy Act contains privacy safeguards set out in a number of Information Privacy Principles (IPPs) and National Privacy Principles (NPPs). The IPPs cover collection, storage and security, use, disclosure and access to “personal information”, which is found in a “record”. The “golden rule” operating in this area is that personal information may only be collected and stored with the consent of the individual concerned, and may only be used for the purpose for which it was collected. An alleged breach of the IPPs may give rise to an investigation by the federal Privacy Commissioner, who has powers under the Privacy Act to make determinations which may only be enforced by the Federal Court after a new hearing. The Privacy Commissioner can also initiate investigations without a complaint and has powers to seek injunctions. In addition, the Commissioner has the power to audit the handling of personal information by Commonwealth agencies.

Initially, the privacy protection afforded by the IPPs extended only (with limited exceptions) to the personal information-handling practices of a federal government “agency”, but the Act was extended to the private sector in December 2001 to include such entities as private hospitals, doctors and other health practitioners, and insurance companies.

Private sector organisations must comply with the NPPs, which set out how to collect, use and disclose personal information, maintain data quality, keep personal information secure, maintain openness, allow for access to and correction of personal information, use identifiers, allow anonymity, conduct trans-border data flows and collect sensitive information. Some of these principles are similar
to the IPPs; however, among other differences, the NPPs contain special provisions for “sensitive information”, a subset of which is “health information”.

Under the Privacy Act, organisations and industries can develop their own privacy codes (for approval by the Privacy Commissioner), which must provide privacy protection of at least equivalent standard to the NPPs; where they do not reach this level, the NPPs apply as the default position. In practice, however, few if any industries have bothered to develop their own codes and submit them to the Privacy Commissioner for scrutiny.

Small business operators – defined by s 6D as those with an annual turnover of less than $3 million – have extensive exemptions from the Privacy Act. However, all organisations or individuals that provide health services and hold any health information (except in an employee record) are subject to the private sector provisions, regardless of their size and income. Due to the broad definitions used in the Privacy Act, health service providers are not limited to hospitals, medical practitioners and others traditionally considered to be part of the health care system. Such organisations and individuals may include gyms and weight loss clinics. Alternative medicine practitioners, pharmacists, mental health professionals, optometrists, and social welfare and counselling service providers also would be considered to be health service providers, whether the service is provided face-to-face, over the phone, via mail order or the internet. However, small business organisations that provide no health services, but merely collect and store health information on behalf of others, probably are not caught by the Privacy Act.

Under Australia’s federal arrangements, most State, Territory and local government bodies, including public hospitals and other health service providers, are not covered by the Privacy Act. Similarly, private sector health service providers working under contract for a State, Territory or local government agency are not covered by the Privacy Act. In all such cases, the applicable practices and protections must be found in the relevant State or Territory privacy legislation, although in many cases these apply similar principles to those in the federal law.

**Privacy implications for consent to the collection and use of genetic information**

Most genetic information about identifiable individuals is obtained from the taking of family medical history or from medical genetic testing, whether diagnostic or predictive, carrier or prenatal. Therefore, such genetic information would likely fit within the definition of health information. Diagnostic testing most clearly counts as health information, since it is information about the health of the individual. Family history and predictive testing would generally also qualify, since it is “information or an opinion about the health or disability (at any time) of an individual” in terms of s 6 of the Privacy Act, even where it deals only with probabilities.

The Federal Privacy Commissioner’s Guidelines on Privacy in the Private Health Sector (2001) state that health information includes “genetic information, when this is collected or used in connection with delivering a health service, or genetic information when this is predictive of an individual’s health”. For the same reason, genetic information provided to insurers or employers also may constitute “health information”, even though it is not taken for clinical or therapeutic purposes.

The position becomes less clear with respect to other forms of genetic testing. There are circumstances in which genetic information may not be health information as defined in the Privacy Act. For example, carrier testing might fall outside the definition of health information, since it is not information about the health or a disability of “an individual”. That is, the health of the test subject is not at issue: the information is about the health of future children. In Victoria, the Health Records Act 2001 (Vic), s 3(1), addresses this by defining health information to include “personal information that is genetic information about an individual in a form which is or could be predictive of the health (at any time) of the individual or any of his or her descendants” (emphasis supplied). Other forms of genetic information that may not fall within the definition of health information include genetic information collected and used to establish parentage or for the purposes of forensic investigation.

NPP 1 provides that an organisation must not collect personal information unless the information is necessary for its functions and must collect personal information only by lawful and fair means and
not in an unreasonably intrusive way. Individuals must be informed about various matters such as their access rights, the purposes of collection and to whom the organisation usually discloses information of that kind. In general, an organisation must collect personal information about an individual only from that individual, rather than from any third party.

The Privacy Commissioner’s Guidelines on Privacy in the Private Health Sector state that there are three key elements involved in seeking consent to use health information in particular ways:

• consent must be provided voluntarily;
• the individual must be adequately informed; and
• the individual must have capacity to understand, provide and communicate her or his consent.

Consent is of particular importance in the collection of genetic information, as compared with most other forms of health information, given the special characteristics of genetic information and the ethical considerations involved in decision-making about genetic testing. For consent to be truly voluntary, there must be no undue pressure or coercion. On one view, an individual’s consent may not be voluntary and valid if the individual is denied some benefit or is disadvantaged in some way because they refused consent. These dimensions of consent may become relevant when considering the application of the NPPs to genetic testing by an employer, prospective employer or for insurance purposes.

NPP 10 contains provisions dealing specifically with collection of health information for the purposes of providing health services. Under NPP 10, a health provider generally must not collect sensitive information (including genetic and other health information) unless the individual has consented. However, NPP 10 then sets out a number of specific circumstances in which an organisation may collect sensitive information without consent, including where collection is required by law; in specified circumstances relating to the provision of health services; and in circumstances related to public interest, such as for research relevant to health and safety, providing that collection is carried out according to certain professional rules of confidentiality.\textsuperscript{58}

Some collection of information necessary for research or statistical purposes may be done without an individual’s consent – but only where obtaining consent is impracticable, de-identified information would not be suitable, and the collection is carried out in accordance with guidelines issued by the NHMRC and approved by the Privacy Commissioner under s 95A of the Privacy Act.

NPP 2 provides generally that an organisation must not use or disclose personal information about an individual for a purpose other than the primary purpose of collection (that is, for a secondary purpose). NPP 2 then sets out a range of circumstances in which an organisation may use or disclose personal information for a secondary purpose, including where the secondary purpose is related (or directly related in the case of health and other sensitive information) to the primary purpose and the person would reasonably expect such use or disclosure; where the individual has consented to the use or disclosure; and in circumstances related to public interest, such as for research relevant to health and safety and for law enforcement purposes.

The Privacy Commissioner’s Guidelines on Privacy in the Private Health Sector provide a range of examples of secondary purposes for which the use or disclosure of personal information would usually be permissible without consent, provided it is within the reasonable expectations of the individual concerned. These include sharing information with other health service providers within a multidisciplinary health care approach. Other directly related secondary purposes may include activities or processes necessary to the functioning of the health sector, including use or disclosure in connection with providing an individual with further information about treatment options; billing or debt-recovery; management, funding, service-monitoring, complaint-handling, planning, evaluation and accreditation activities; addressing liability indemnity arrangements (eg in reporting an adverse incident to an insurer); or disclosure to a clinical supervisor by a psychiatrist, psychologist or social worker.

\textsuperscript{58} The Privacy Commissioner has noted that these professional rules must be binding on the health service provider (that is, the breach will give rise to adverse consequences) and must be established by a competent health or medical body, such as medical boards recognised in federal, State or Territory law.
Is the vexed issue of “ownership” of bio-samples a privacy issue?

The Inquiry was concerned about genetic samples and information held in tissue collections maintained chiefly by hospitals or pathology laboratories, which were not collected primarily for use in research (e.g., archived collections of preserved human tissue, or collections of Guthrie cards), but nevertheless may be valuable research resources for studies into the genetic causes of disease. Genetic testing of stored tissue samples also has potential uses in other contexts, including for parentage or other kinship testing, police forensic investigations, and as evidence in court proceedings.

These secondary uses raise important issues of ethics, privacy and consent. The Inquiry also recommended that the Australian Health Ministers’ Advisory Council (AHMAC) develop nationally consistent rules governing any disclosure or further use (including for law enforcement purposes) of genetic samples and information held in human genetic research databases and other human tissue collections. These rules should be based on the principle that any such disclosure is permissible only with the consent of the person sampled (or a person authorised to consent on her or his behalf), a waiver of consent granted by an HREC, or pursuant to a court order.

The Inquiry considered whether privacy interests in genetic information might be protected more effectively by recognising increased property (or intellectual property) rights over genetic samples. Each Australian State and Territory has enacted legislation that regulates the donation of human tissues and organs for transplantation and research (the Human Tissue Acts).

The Inquiry concluded there should be no change to the current position whereby hospitals and pathology laboratories have a proprietary right to preserved samples, but full property rights in genetic samples are not recognised. Instead, the ALRC recommended that better articulated privacy laws and consent regimes be used to protect the legitimate interests of tissue donors.

One of the more controversial recommendations in Essentially Yours was that the federal Privacy Act and other privacy and health information laws should be extended to cover identifiable genetic samples. Although this would shift privacy laws from the traditional information/data protection paradigm, there is a clear parallel between data electronically encrypted through computer technology in hard drives, disks or other devices – which is currently afforded protection under privacy laws – and genetic information that is “encrypted” in tissue samples, but now readily sequenced and analysed through biotechnology.

Importantly, coverage through privacy legislation would meet many of the concerns and anxieties expressed to the Inquiry by members of the general public about the privacy and security of their genetic information held by human genetic research databases – and the same would apply in relation to samples held by umbilical cord blood banks.

For example, NPP 6 of the Privacy Act states that, subject to some exceptions, if an organisation holds personal information about an individual, it must provide that individual with access to the information on request. However, there is no similar right to obtain access to a genetic sample.

NPP 2 contains a core obligation that organisations can disclose personal information only for the primary purpose for which it was collected or for directly related secondary purposes. However, no similar obligation applies to the disclosure (that is, “transfer”) of genetic samples. There may well be professional, regulatory, contractual or other consequences for the organisation transferring the samples, but remarkably there is no general legal obligation not to transfer possession (or sell) a sample without the consent of the individual from whom the sample comes. Privacy principles would also regulate such issues as lawful access by biological relatives or by other third parties. Although

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59 Comprised of representatives of all federal, State, Territory and New Zealand health ministries.

60 This template legislation is based on the recommendations of the ALRC in Human Tissue Transplants, ALRC 7 (1977).

61 The Inquiry did suggest that it was timely to review the Human Tissue Acts, given the changes in medical science and technology, as well as in some social attitudes and understandings, over the past quarter-century.

62 See ALRC, n 1, Ch 8, Privacy of Genetic Samples.

63 ALRC, n 1, Recommendations 8-1 to 8-4.
umbilical cord blood bank contracts are likely to cover to such matters, it is not inevitable that this will be the case, or that the relevant clause will be drawn in the consumer’s favour, or will be drafted effectively.

Similarly, organisations would have to comply with legally enforceable standards for the physical security of holdings of genetic samples and would not be permitted to retain samples without a clearly defined purpose.64 Under ordinary privacy principles, genetic samples would not be able to be sent outside of Australia unless reasonable steps were taken to ensure that the privacy interests in the samples are adequately protected by the recipient in the overseas jurisdiction.65

As the ALRC commented in relation to biobanks, but it would apply to umbilical cord blood banks with equal force:

> There are benefits in promoting more openness about, and public understanding of, the ways in which samples are dealt with. Openness and accountability may reduce the need for other sector-specific regulation – including, for example, the licensing or registration of human genetic research databases.66

Unfortunately, however, the government chose not to accept these recommendations, and thus lost this opportunity, arguing somewhat tautologically that:

> The Privacy Act is the appropriate vehicle for ensuring the privacy protection of personal information derived from genetic samples but the Act should not cover the handling of genetic samples. The privacy principles are designed to regulate the collection, use and disclosure of personal information, not the source of that information. Accordingly, the Government does not consider that privacy legislation is the appropriate place for regulating genetic samples. The concerns raised about the use and handling of genetic samples could be addressed in the Human Tissues Acts.67

### Individual, familial or communal rights?

The Human Genetic Project confirmed the very strong familial dimension of “personal” genetic information: we share 99.9% of our genetic sequence with all other human beings, and an even higher percentage with members of our own families and communities. However, most of the laws, ethical principles and regulatory models in Western societies (especially in the English-speaking world) are built around a powerful cultural preference for individual rights and autonomy, and in the medical context, on the primacy of the individual doctor-patient relationship.

The Inquiry heard often from doctors and, especially, familial cancer registries that they “live in dread” of the day they would receive a telephone call from a person dying of cancer, who would say, for example:

> You treated my sister, whose test showed a predisposition to a familial cancer (such as BRCA1, or colon cancer, or FAP), but we are estranged and she never told me anything about it. However, you are a health professional and all you had to do was make one phone call – and I would have sought my own medical advice, and I probably would not now be in the terminal stages of cancer.

Concern was expressed about both the legal ramifications of this scenario as well as the ethical and moral dimensions.

The late Professor Dorothy Wertz of the University of Massachusetts conducted a number of fascinating cross-cultural empirical surveys, which revealed a marked divergence in approach.68 For example, in response to questions about whether it would be proper to reveal to genetic relatives the fact that a patient tested positive for Huntington’s disease or for a familial cancer mutation, health professionals in Northern Europe, Western Europe and most especially the English-speaking countries (including Australia, the United States, the United Kingdom, Canada and New Zealand) placed their focus squarely on the individual doctor-patient relationship and were reluctant to breach this

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64 *Privacy Act 1988* (Cth), National Privacy Principles, NPP 4.
66 ALRC, n 1 at [8.56].
67 Australian Government, n 3, Recommendations 8.1 to 8.4.
confidence, whatever the consequences for other family members. By way of contrast, health professionals in African, Asian, Latin American, Middle Eastern, Eastern European and Southern European societies were much more likely to value familial and communal interests over individual autonomy.

In a similar vein, there is an urgent need to promote public debate about how we wish to proceed with work on population genetics, especially in relation to the use of collected genetic material. The use of such material – in the case of Guthrie cards, as mentioned, virtually a complete national collection – for epidemiological purposes would have beneficial society-wide effects in terms of research, planning and public health administration. Of course, this would mean some trade-offs against privacy protection for individuals – but is that a price that an informed citizenry would be prepared to pay?

Confidentiality has been a traditional cornerstone of the doctor-patient relationship, and should not be departed from lightly. Nevertheless, the Inquiry felt sufficiently strongly about this to recommend that the Privacy Act be amended to recognise that there may be circumstances in which doctors should be permitted to disclose confidential genetic information to genetic relatives without the patient’s consent, namely where this is necessary to lessen or prevent a serious threat to an individual’s life, health or safety – even where the genetic risk is not “imminent”, as was required under the then existing language in the Act.69 In order to provide sufficient guidance to health professionals, the ALRC further recommended that the NHMRC should develop guidelines, in consultation with the federal Privacy Commissioner, for dealing with the disclosure of genetic information to the genetic relatives of their patients.70

The government accepted these recommendations, and amended the Privacy Act accordingly.71 The NHMRC engaged in a series of consultation exercises to develop and refine the guidelines which, under the National Health and Medical Research Council Act 1992 (Cth), have the force of regulations. In December 2009, in cooperation with the Privacy Commissioner, the NHMRC released The Use and Disclosure of Genetic Information to a Patient’s Genetic Relative under Section 95AA of the Privacy Act 1988 (Cth): Guidelines for Health Practitioners in the Private Sector.

**Non-discrimination**

The fear of emerging genetic discrimination, especially in insurance, employment and access to government services, was central to the establishment of the ALRC-AHEC Inquiry. In the context of human genetic research databases and biobanks, researchers, doctors and community groups raised concerns that unless adequate legal protections were developed, people would become reluctant to volunteer for research studies or even to undergo needed clinical genetic tests, for fear of suffering adverse incidental consequences. For example, people might fear that a positive genetic test for a disease marker (such as Huntington’s disease, or heritable breast or colorectal cancer) could be used to deny them risk-rated insurance coverage (or to elevate premiums beyond reach) or employment opportunities.

Anti-discrimination laws exist at the federal, State and Territory levels in Australia. At the federal level, the major pieces of legislation include the Sex Discrimination Act 1984 (Cth), the Racial Discrimination Act 1975 (Cth), the Disability Discrimination Act 1992 (Cth) and the Age Discrimination Act 2004 (Cth). In addition, the Workplace Relations Act 1996 (Cth) contains provisions that prohibit discrimination on a range of grounds in respect of the termination of employment. However, none of these Acts specifically address discrimination on the basis of genetic status.

Although it is conceivable that a complaint of discrimination on the basis of genetic status could be based on race (say, by a carrier of Tay-Sachs disease or sickle cell anaemia, which run predominately in people of Jewish or African ancestry, respectively) or gender (say, by a woman with

69 ALRC, n 1, Recommendation 21-1.
70 ALRC, n 1, Recommendation 21-2.
71 Privacy Act 1988 (Cth), NPP 2.1(c)(iii) was amended to achieve this end.
a genetic mutation for breast cancer or male with “fragile X” syndrome), most such actions are likely
to arise under the Disability Discrimination Act 1992 (Cth). Under the Act, disability discrimination is
prohibited in employment, education, access to premises used by the public, provision of goods,
services and facilities, accommodation, buying land, activities of clubs and associations, sport and the
administration of Commonwealth government laws and programs.

A product of its time, the Disability Discrimination Act was designed to apply to unlawful
discrimination based on a person’s physical disability, mental illness, intellectual disability or
HIV-AIDS positive status (“the presence in the body of organisms capable of causing disease or
illness”) – but there is no express reference to genetic status. The Inquiry recommended that, in order
to provide a consistent approach to addressing genetic discrimination, the Disability Discrimination
Act and related laws and regulations72 should be amended to make clear that they expressly apply to
discrimination based on genetic status.73 In July 2004, the Productivity Commission completed its
own review of the Disability Discrimination Act, and supported the ALRC’s recommendation to
amend the definition of “disability” in the Act to clarify that the legislation applies to discrimination
based on real or perceived genetic status.74

In its 2005 response to Essentially Yours, the government accepted the recommendations of the
ALRC (and as echoed by the Productivity Commission) on this issue, “consistent with the
Government’s policy to eliminate discrimination, as far as possible, in all areas of community life”.75
The recommendations were implemented with the passage of the Disability Discrimination and Other
Human Rights Legislation Amendment Act 2009 (Cth),76 which amended the Disability Discrimination
Act to:

• address discrimination on the basis of genetic status where a person may have the genetic markers
for a condition but is presently asymptomatic77 and

• prohibit an employer from requesting or requiring information, including genetic information,
from a job applicant or employee, except where the information is reasonably required for
purposes that do not involve unlawful discrimination.78

These new provisions are very similar in nature to the American protections brought in (with
surprisingly bipartisan support) under the Genetic Information Non-discrimination Act 2008 (US).

Beyond their role in providing a safety net for individuals participating in genetic research
projects, these protections should also provide substantial comfort for individuals who are considering
making a umbilical cord blood bank deposit, but are concerned about potential discrimination or
stigmatisation problems down the track, where eg a prospective employer might infer from an
individual’s personal or family involvement in cord blood banking that he or she may be concerned
about genetic conditions or predispositions. Any employment decision now made on this basis would
clearly amount to unlawful discrimination under the Disability Discrimination Act.

SECURITY

Even where the developers of a biobank are attentive to issues of privacy protection, there is residual
anxiety about the secure nature of such sites, given the sensitive information held. At a number of the
ALRC’s public consultation meetings, concerns were raised about the apparent ease with which
computer hackers had accessed supposedly high-security websites run by banks and governmental

72 Such as the Human Rights and Equal Opportunity Commission Act 1986 (Cth) and the Workplace Relations Act 1996 (Cth).
The ALRC also made a parallel recommendation that the States and Territories should consider harmonising their
anti-discrimination legislation, and other relevant laws, in a manner consistent with the recommendations in the report: ALRC,
n 1, Recommendation 9-5.
73 ALRC, n 1, Recommendation 9-3.
75 Australian Government, n 3, Recommendations 9-1 to 9-5.
76 The Act received Royal Assent on 8 July 2009, and entered into force on 5 August 2009.
77 Implementing Recommendation 9-3 of ALRC, n 1.
78 Implementing Recommendation 31-3 of ALRC, n 1.
authorities (including military and intelligence agencies), as well as errors made which exposed restricted material (such as credit card details) on websites or lost laptops, or left paperwork (such as income tax returns) in a public place. Given the sensitive information and material held by umbilical cord blood banks, great attention must also be placed on security, both “hard” and “soft”.

**ACCESS AND EQUITY CONCERNS**

Another commonly expressed concern during the ALRC’s community consultation exercise on the protection of human genetic information was about access and equity: the fear that yet another major modern technology with the potential to make life better might, in practice, tend to drive up the costs of health care and increase the divide between the “haves” and the “have nots”.

For example, a number of people surmised that while “smart drugs” based on modern pharmacogenomics are ostensibly a good thing, the resulting markets for individualised and customised drugs would be smaller and more fragmented – resulting in more effective drug therapies, but at much higher prices. Concerns were expressed by indigenous people, among others, that this would tend to shape the research programs for drug companies, prompting them to focus more on “white, middle class, lifestyle diseases” than on diseases associated with poverty or those primarily affecting the poor and disadvantaged.

These issues also featured in the ALRC’s subsequent inquiry into *Gene Patenting and Human Health* (2004-2005), which explored the balance between encouraging investment and innovation in biotechnology and ensuring that further research and the delivery of cost-effective clinical genetic services are not compromised. It arose again in the ALRC’s privacy inquiry (2006-2008), with respect to the perceived “digital divide” between city dwellers with relatively inexpensive broadband access, and those living in rural, regional and remote communities.

There is little doubt that any sustained public scrutiny of umbilical cord blood banking operations would likewise incorporate concerns about equity, access and benefit-sharing. However, the issue may play out quite differently for umbilical cord blood banks than for biobanks. In the latter case, the dominant theme is ensuring that research volunteers should be

- provided with sufficient information to make an informed decision about consent to participate;
- assured that the project will be governed in an open and accountable way, and with probity and integrity;
- protected to the extent possible from any “collateral damage”, in terms of loss of privacy or future discrimination or stigmatisation occasioned by such participation; and
- entitled to clear and honest statement about beneficial ownership of the project (including intellectual property), commercial opportunities and benefit-sharing, with no hidden traps or secret deals.

With biobanking or participation in other large-scale cohort studies, research participants are generally acting for altruistic reasons, whether these stem from family involvement or not. Participants can volunteer or not (or volunteer and subsequently withdraw), but they generally do not believe that there will be direct personal benefits by virtue of such participation. Porter et al suggest there is also a strong element of altruism in umbilical cord blood banking, with donation considered to be a contribution to a valuable community resource and an aspect of good citizenship and “an act of good mothering.” Indeed, public opinion seems to be that a failure to donate umbilical cord blood amounts to the “waste” of an important resource, and that public health authorities should ensure that donation is easier and more available to those who want it.

In other words, community sentiment contemplates a positive “right” to have umbilical cord blood banked for potential future use – a right that is often frustrated in practice because of the limited

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81 Porter et al, n 80.
funding for this purpose and the consequential limited availability of collectors “out of hours”. As noted above, less than 1% of the available cords are banked each year in Australia.\(^\text{82}\) If more evidence (or perhaps more publicity) emerges about the clinical benefits of umbilical cord blood banking, then there will be increased pressure for availability and, in turn, equity concerns about whether access to this resource should be determined primarily by ability to pay for private services.

**COMMERCIALISATION**

It was striking that at virtually every public consultation meeting conducted by the ALRC, the same concern was expressed in almost identical terms:

We can see the value of medical research into genetics, and we generally would be happy to participate by giving information, blood or tissue to facilitate this research. However, we are not comfortable with the heavy degree of commercialisation of this research, and we definitely do not want our altruism to lead to billion dollar profits for multinational pharmaceutical companies [almost invariably referred to as “American pharmaceutical companies”].

Perhaps it is no surprise, then, that when novelist John Le Carre needed a new entity to replace the fallen Soviet Union as the “Evil Empire” in *The Constant Gardener* (2001), he chose “Big Pharma” for this purpose.

Another concern that clearly emerged at the public forums is the atavistic or primal fear among members of the community about their genetic material being sent “overseas” (again, often expressed as being “sent to the US”). At almost every event, someone in the audience expressed concern about volunteering for an experiment at an Australian university research lab or teaching hospital, then finding that the research group had “spun off” into a private biotech company, which then merged with or was taken over by American interests – and “the next thing you know, your DNA is overseas!”

Interestingly, it was not the profit motive or commercialisation per se that worried most members of the community, but rather the lack of transparency: there was considerable anger expressed by research participants who subsequently found out that their doctors or the research scientists had undisclosed financial interests in the project. Any hint of a monopoly or exclusive commercial arrangements – such as those associated with Iceland’s deCODE Genetics and pharmaceutical company Hoffmann La Roche – is likely to arouse great antipathy.

Empirical evidence about public attitudes to research in the United Kingdom is instructive. A survey conducted by the United Kingdom Human Genetics Commission has found that levels of public trust in the responsible use of human genetic information vary markedly, depending on the nature of the individuals or bodies holding it. In particular, respondents trusted academic scientists more than health and pharmaceutical companies.\(^\text{83}\)

Another report on qualitative research connected with the development of the UK BioBank initiative concluded that the fact that sample collection would be a “publicly funded initiative and not set up as a profit-making exercise was reassuring and important in communicating its credibility”.\(^\text{84}\) The report indicated:

[T]here are likely to be questions from the general public and in the media about commercial access to, and use of, the samples and information. Assuming samples are donated freely by donors, there needs to be careful explanation of the financial implications of this.\(^\text{85}\)

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\(^{82}\) O’Connor et al, n 6.


\(^{85}\) See n 17.
Leading Australian academics have expressed the view that, from an ethical perspective, “the potential for commercial exploitation” of genetic samples and other biological materials is a very relevant consideration when individuals decide whether to consent to participate in research, given that participation is typically altruistic in nature.86

The ALRC accepted that there is a clear need for open and transparent disclosure, to prospective research participants, of the potential commercialisation of research outcomes and the commercial interests of the researchers involved. It was suggested that such disclosure may protect the interests of both prospective research participants and researchers themselves:

In order to avoid feelings of exploitation, and possibly even deception, it is of crucial importance that they be given the opportunity to consent to participation in the knowledge that there is a possibility of commercial gain being made from their donated biological material. To do otherwise risks damaging the perception of research and may thereby reduce the willingness of people in the community to participate.87

The National Statement contains a number of provisions relating to the disclosure of funding and financial interests. However, there is no general requirement to disclose this information, or other information about the actual or anticipated commercial arrangements connected with the research, to research participants. The National Statement provides that a researcher must disclose to the HREC reviewing the research proposal the amount and sources or potential sources of funding for the research and must declare any affiliation or financial interest. The HREC must consider the extent to which the researcher should disclose information about funding sources to research participants.88 The HREC may decide that no such disclosure is justified. (The disclosure requirements in relation to clinical trials are somewhat more rigorous.)89

The Australian Academy of Sciences submitted to the ALRC-AHEC Inquiry that “all applications to HRECs should be required to include details of any commercial support obtained or envisaged”.90 The Australian Red Cross Ethics Committee stated:

[T]here should be transparent disclosure to research participants of the potential commercialisation of research outcomes, as well as any conflicts of interest … [The] Committee requires disclosure of commercial arrangements for funding or product development. No researcher has ever raised any objection. In fact most researchers provide a full explanation of the commercial aspects of the research. We should also point out that our standard-model information form includes specific questions on commercialisation. There has been a general acceptance of the disclosure principle.91

The ALRC endorsed this approach and recommended that the NHMRC develop information and advice on the disclosure by researchers, to research participants, of information about actual or anticipated commercial arrangements connected with human genetic research proposals.92

The parallels with umbilical cord blood banking are obvious. There is general community support for the activity, and it is suffused with an altruistic glow. Even the once-vexed public-private controversy over cord blood banking has been largely abated through cooperation and good practice. There have been no media “horror stories” about fortunes made or lost by public or private umbilical cord blood banks, nor about inappropriate dealing or experimentation – and it is critical that open, accountable and sound management ensures that this continues to be the case.

**Cost-benefit issues**

Finally, people want to be presented with unbiased information about the relative benefits of the research project or biobank. Although there is generally strong support in the community for human

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87 Nicol, Otlowski and Chalmers, n 86 at 93.
88 National Health and Medical Research Council, n 45 at [2.21].
89 National Health and Medical Research Council, n 45 at [12.5]-[12.6].
90 ALRC, n 1 at [16.47].
91 ALRC, n 1 at [16.49].
92 ALRC, n 1, Recommendation 16-1.
genetic research, given the high cost and the potential collateral risks involved, it is by no means universally accepted that it is wiser to focus, say, on “the gene for obesity” than on promoting good nutrition, or that biobanking is “the most effective and cost-effective way to reduce the incidence of common diseases”. 93 As one commentator has noted, “the only thing certain about these population-wide, genotype-phenotype resources is that they are staggeringly expensive”. 94

In order to develop and maintain public support, those establishing biobanks and umbilical cord blood banks must provide an honest assessment of the relative value and cost-effectiveness of these activities, walking the fine line between promoting work in an area they passionately believe in, and over-selling the known benefits. 95

CONCLUSIONS
There are a number of key lessons for umbilical cord blood banks that emerge from the ALRC’s community consultation program and from the controversies that surrounded, stalled or derailed the establishment of some of the early biobanks. 96

Contrary to the situation in Europe,97 it was evident in the meetings, consultations and submissions that Australians have not lost faith in the possibility of effective regulation of biotechnology in the public interest. In part, this is as a result of good management to date; however, it is at least as much the result of good fortune, insofar as Australia (unlike Europe) has not suffered any public health crises or major scandals in this area that have sapped public confidence. Similar surveys in North America indicate that Americans are “confidently supportive” and Canadians are “cautiously supportive” of genetic research, are willing to rely on the advice of scientific and technical experts about risk, and “want governments to manage the risks and take a leadership role”. 98

Strong attention also must be paid to ensuring that other concerns – about privacy, discrimination, informed consent, governance, security, commercial fairness and financial probity – are addressed in structural terms and monitored thereafter, in order to maintain public confidence and avoid a backlash that inevitably would imperil such research.

First and foremost, there is a clear need for openness and transparency, and ample public education and debate informed by good science, in order to ensure community acceptance and legitimacy. The critical lesson to be learned from international experience is that public trust and credibility must be carefully maintained, for once they are lost they are very difficult to restore.

95 See n 28.
97 Only 45% of Europeans agreed with the statement that their governments regulate biotechnology well enough, compared with 29% who disagreed, and 26% who were not sure: Eurobarometer 52.1, The Europeans and Biotechnology, http://www.ec.europa.eu/research/quality-of-life/eurobarometer.html viewed 9 February 2012.