

AUSTRALIAN INQUIRY INTO THE PROTECTION OF HUMAN GENETIC INFORMATION

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INTRODUCTION

In February 2001, the Commonwealth Attorney-General and the Minister for Health and Aged Care established an inquiry into the protection of human genetic information, to be conducted jointly by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC).¹ The inquiry is due to report its findings and recommendations by 31 March 2003.

The Government's decision to opt for a joint inquiry reflects the wide array of legal and ethical concerns and contentions surrounding this new field, and mirrors the Human Genome Project's establishment of working groups focusing on the various ethical, legal and social implications (ELSI) of the scientific advances.

The ALRC has long experience in dealing with legal issues that involve important ethical and social dimensions - including work that led to the adoption of *Human Tissue Acts* in all Australian jurisdictions - and well-tested processes for engaging in effective community consultation.² AHEC is a principal committee of the National Health and Medical Research Council (NHMRC). It advises the NHMRC on ethical issues broadly relating to health and medical research, and is also specifically responsible for developing guidelines for the conduct of medical research involving humans.³

* This article reflects work being conducted by the Members and staff of the Australian Law Reform Commission and the Australian Health Ethics Committee in *Protection of Human Genetic Information*, Issues Paper 26 (2001), ALRC, Sydney (hereafter 'IP 26'). Professor David Weisbrot is President of the Australian Law Reform Commission (ALRC); Mr Spiteri and Ms Carney are Legal Officers at the ALRC assigned to this inquiry.

¹ Attorney-General and Minister for Health and Aged Care, Joint News Release, 'Genetic Privacy', 7 February 2001.

² The functions of the ALRC are set out in *Australian Law Reform Commission Act 1996* (Cth) s 21.

³ The functions of AHEC are set out in the *National Health and Medical Research Council Act 1992* (Cth) s 35.

BACKGROUND TO THE JOINT INQUIRY

Concerns about the use of genetic information are not new. The extent to which genetic information should remain private, and the ability to treat people differently on the basis of their genetic information, are both matters that have been debated in the United States, Canada and Europe. Thus, this inquiry is in the fortunate position of proceeding from a foundation of discussion and research being generated in Australia and overseas.⁴

In common with many OECD countries, Australia has a policy, expressed in its *Innovation Report*,⁵ placing great reliance for its economic future on genetic technology, particularly human genetic technology. Significant steps have been taken to implement this policy:

- The *National Statement on Ethical Conduct in Research Involving Humans* (hereafter the *National Statement*)⁶ has set down a comprehensive national ethical regulatory framework for the conduct of research in general and genetic research in particular.⁷
- *Biotechnology Australia* is a whole of government initiative⁸ to coordinate efforts to develop biotechnology for the benefit of the Australian community.⁹
- The Ralph Report on taxation reform has recommended reforms to income tax arrangements to ensure that the Australian taxation regime for biotechnology companies is consistent with other OECD nations, as a means of encouraging investment in Australian biotechnology.¹⁰
- A major review of health and medical research in Australia has been undertaken. The Wills Report¹¹ refers particularly to the need to take

⁴ For example, see House of Representatives Standing Committee on Industry Science and Technology, *Genetic Manipulation: The Threat or the Glory* (1992), Parliament of Australia, Canberra; Federal Privacy Commissioner, *The Privacy Implications of Genetic Testing* (1996), Office of the Federal Privacy Commissioner, Sydney; Senate Legal and Constitutional Legislation Committee, *Provisions of the Genetic Privacy and Non-discrimination Bill 1998* (1999), Parliament of Australia, Canberra. See IP 26 for more information.

⁵ See the Government's Innovation Report 2001-2002, *Backing Australia's Ability: Real Results, Real Jobs* < <http://backingaus.innovation.gov.au/docs/200102.pdf> >.

⁶ Prepared by the Australian Health Ethics Committee under the relevant provisions of the *National Health and Medical Research Council Act, 1992* (Cth), and endorsed by the Australian Vice Chancellors' Committee, the Australian Research Council and the Learned Academies in 1999.

⁷ See Principles 16.1-16.23 and the section on Human Tissue (Principle 15).

⁸ Involving the Commonwealth departments responsible for industry, science, resources, environment and heritage, agriculture, fisheries, forestry, health and ageing, education, and training.

⁹ Commonwealth of Australia, *Australian Biotechnology: Progress and Achievements* (2000) 2.

¹⁰ Review of Business Taxation, *A Tax System Redesigned: More Certain, Equitable and Durable: Report* (1999), Commonwealth of Australia, Canberra (Ralph Report).

¹¹ Health and Medical Research Strategic Review, *The Virtuous Cycle, Working Together for Health and Medical Research* (1999), Commonwealth of Australia, Canberra (Wills Report).

advantage of advances in biotechnology to improve the health of the Australian population, to build the economy and to create valuable jobs.¹² It recognises that this window of opportunity would close given the pace of change unless Australia acts promptly.

- The Federal Government's announcement on 29 January 2002 that, as part of a shift towards setting national priority areas for research, one-third (or approximately \$170 million) of the Australian Research Council's funding grants for 2003 would be reserved for four designated key areas of scientific research, one of which is genome/phenome research.¹³

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

A central tenet of *Biotechnology Australia* is to ensure that 'consistent with safeguarding human health and ensuring environment protection, that Australia capture the benefits of biotechnology for the Australian community, industry and environment'.¹⁴ The previous federal Minister for Health and Aged Care, the Hon Dr Michael Wooldridge MP, has emphasised 'the driving imperative of identifying and managing any risks associated with the technology before all other matters, only then can we be truly confident about reaping the broader benefits'.¹⁵ The establishment of the genetic technology industry must be accompanied by appropriate legal and ethical regulatory regimes to protect the community and the research participant.¹⁶

PRECIPITATING FACTORS

A number of inquiries and events over the past decade have raised public consciousness about the breakthroughs in genetic science and technology, as well as the emerging ethical, legal and social issues attaching to those developments.

¹² Ibid 1.

¹³ See Dr Brendan Nelson, Minister for Education, Science and Training, 'Backing Australia's Ability - Funding for Research Priorities Announced', *Media Release*, 29 January 2002, available at http://www.dest.gov.au/ministers/nelson/jan02/n06_290102.htm. See also Dorothy Illing, 'Nelson says research quality won't suffer', *The Australian*, 6 February 2002. The other designated areas are nano and bio-materials; complex/intelligent systems; and photon science and technology.

¹⁴ See *Australian Biotechnology: Progress and Achievements* (2000) available at <http://www.biotechnology.gov.au/Industry_Research/National_Strategy/prop-biotech_prog_achiev.pdf>.

¹⁵ In the Second Reading Speech for the Gene Technology Bill 2000: see Commonwealth of Australia, *Parliamentary Debates*, House of Representatives, 22 June 2000, 18104 Michael Wooldridge, Minister for Health and Aged Care).

¹⁶ See *Australian Biotechnology: A National Strategy* (2000) available at <http://www.biotechnology.gov.au/Industry_Research/National_Strategy/prop-biotech_nat_strategy.pdf>.

In Australia, privacy issues relating to genetic technology were identified in the 1992 report of the House of Representatives Standing Committee on Industry, Science and Technology entitled *Genetic Manipulation: The Threat or the Glory?*¹⁷ This led to the federal Privacy Commissioner's release of an information paper in 1996, entitled *The Privacy Implications of Genetic Testing*.¹⁸ The Privacy Commissioner recommended a coherent and consultative approach to developing policy on privacy questions raised by genetic testing.

In March 1998, Australian Democrats Senator Natasha Stott Despoja introduced the Genetic Privacy and Non-discrimination Bill into Federal Parliament. This Bill was based on the *Model Genetic Privacy Act* (which has influenced genetic privacy laws introduced in some American states),¹⁹ and the US *Genetic Confidentiality and Non-discrimination Act 1997*.²⁰

The primary objectives of the Stott Despoja Bill were to: establish an enforceable right to privacy of genetic information of an individual; define the circumstances in which genetic information and DNA samples may be collected, stored, analysed, and disclosed; prohibit discrimination based on genetic information;²¹ and establish mechanisms to enforce the rights and responsibilities established under the Bill.

The Bill was considered by the Senate Legal and Constitutional Affairs Committee, which received more than 50 submissions from the public. In its March 1999 report on the Bill, the Committee concluded that, as genetic technology is still in an early stage of development, it would be premature to legislate on genetic privacy and non-discrimination, and that further examination of the appropriate regulatory structures was needed.²² The Committee also considered that creating specific legislation for genetic privacy and discrimination would cut across a number of regulatory systems already in place, or in the process of being established; and suggested that it would be more appropriate to amend existing privacy and discrimination legislation where necessary, to ensure that issues raised by genetic technology are adequately covered under that legislation.²³

Separate studies were conducted in Australia in 1999 by genetic counsellor Dr Kristine Barlow-Stewart and postgraduate law student David Keays, based on anonymous responses received from survey forms distributed by clinical geneticists

¹⁷ House of Representatives Standing Committee on Industry Science and Technology, *Genetic Manipulation: The Threat or the Glory* (1992), Parliament of Australia, Canberra.

¹⁸ Federal Privacy Commissioner, *The Privacy Implications of Genetic Testing* (1996), Office of the Federal Privacy Commissioner, Sydney.

¹⁹ Developed by Professors George Annas, Leonard Glantz and Patricia Roche of the Boston University School of Public Health.

²⁰ Bill sponsored by US Senators Peter Domenici, Christopher Dodd and James Jeffords.

²¹ Senate Legal and Constitutional Legislation Committee, *Provisions of the Genetic Privacy and Non-discrimination Bill 1998* (1999) Parliament of Australia, Canberra, 1.

²² Ibid 37.

²³ Ibid 37-39.

and genetic support networks in Australia and New Zealand.²⁴ Genetic discrimination, defined in these studies as less favourable or adverse treatment because of a positive genetic test result, was reported with respect to genetic tests for a wide range of medical conditions.²⁵

Most of the allegations of genetic discrimination touched on insurance, with 45 cases reported in respect of life insurance, income protection insurance, trauma insurance, superannuation, or health insurance.²⁶ The actions complained of involved loading premiums, denial of requested increases to pre-existing insurance coverage, and blanket refusal to provide insurance.²⁷ There were also five reported cases of alleged genetic discrimination in employment or application for employment,²⁸ as well as two cases in which individuals reported that they had been unfairly denied access to health services because of their genetic status.²⁹

The studies suggested that in many of these cases the discriminatory decision or action was inappropriate - that is, the action was based on misinformation or a misunderstanding of genetic test information and the nature of genetic disorders. The studies received widespread media publicity upon publication in mid-2000, and were a significant factor in the initiation of the joint inquiry.

In August 1999, the Investment and Financial Services Association (IFSA), whose members account for 98% of the life insurance industry in Australia,³⁰ lodged applications with the Australian Competition and Consumer Commission (ACCC) in relation to its draft policy on genetic testing. The ACCC was asked to grant authorisation under national competition laws³¹ for IFSA's draft policy on genetic testing.

²⁴ Dr Barlow-Stewart received 703 anonymous responses, in which there were 43 cases of alleged discrimination. Keays conducted interviews with five other persons who had reported instances of alleged genetic discrimination: see Kristine Barlow-Stewart and David Keays, 'Genetic Discrimination in Australia' (2001) 8 *Journal of Law and Medicine* 250, 251–252. Barlow-Stewart initially presented work in progress to the 'Dolly McBeal Genetics and the Law' Seminar in Sydney on 14 August 2000. Keays presented his findings to the Australian Institute of Health, Law and Ethics Conference in July 2000.

²⁵ Ibid 252. The conditions included haemochromatosis, inherited breast cancer, inherited bowel cancer, familial melanoma, Alzheimer's disease, Huntington's disease, and hyperlipidemia.

²⁶ Ibid 253.

²⁷ Ibid 253.

²⁸ Ibid 254.

²⁹ Ibid.

³⁰ In terms of market share.

³¹ Under the *Trade Practices Act 1974* (Cth), organisations who engage, or propose to engage, in certain anti-competitive business arrangements or conduct that could breach the Act may apply to the ACCC for authorisation of such arrangements or conduct. The ACCC may grant authorisation where the public benefit of the subject arrangements or conduct outweighs the public detriment, including the anti-competitive detriment. If granted, an authorisation provides immunity from legal proceedings under the Act in respect of the arrangements or conduct.

The critical aspect of IFSA's draft policy (contained in clauses 2 and 4) was that member insurers could ask individuals to disclose existing genetic tests for the purpose of risk assessment, but that member insurers could not initiate any genetic test on applicants for insurance (directly, or indirectly - such as through the offer of lower than standard premium rates for individuals with negative test results).

The ACCC's Draft Determination, issued 14 June 2000, proposed *not* to grant authorisation for such arrangements, on the basis that:

the provisions of the IFSA draft policy were not likely to result in a public benefit, and that significant anti-competitive detriment would arise from a collective agreement to prevent the offer of lower than standard premiums based on genetic test results.³²

In other words, the ACCC wanted insurance companies to have increased opportunities to *discriminate in favour* of persons who could show a 'clean' genetic bill of health by offering them discounts on premiums and other favourable terms.

However, after further consideration, the ACCC decided in November 2000 to grant IFSA a two-year authorisation, noting 'the complex issues involved' and deciding to provide a 'breathing space' during which the issues surrounding genetic testing could be debated and government policy developed. A major factor in the ACCC's decision was the government's announcement of the joint inquiry:

The ACCC considers that there are complex issues involved in this matter. The ACCC therefore welcomes the proposed government inquiry into human genetic information privacy and discrimination issues. There is a need for some consideration of all the issues by government. An important part of the debate would also include the issue of whether or not industry self-regulation is appropriate in respect of the issues involved.³³

Finally, the Human Genome Project (HGP) and Celera Genomics jointly announced the first near-complete draft of the entire DNA sequence of the human genome in June 2000, and publication of material by Public Sequencing Consortium of the HGP followed in US and UK science journals in February 2001. The first draft of the human genome sequence is a major starting point in the research effort to apply this knowledge into mainstream medical practice. It will provide a significant platform for other applied sub-branches of human genetic research and technology, including such topics as gene identification, genetic variation, gene expression monitoring, microarray technology, bio-informatics, systems biology, protein structure and proteomics.

³² Australian Competition and Consumer Commission, *Draft Determination re Application for Authorisation Lodged by Investment and Financial Services Association (IFSA) in Relation to the Implementation of a Draft Policy on Genetic Testing* (2000).

³³ Australian Competition and Consumer Commission Media Release, 'ACCC Authorises Life Insurance Bar on Genetic Testing' 22 November 2000.

SOCIAL REACTIONS TO RAPID SCIENTIFIC CHANGE

This research effort will continue apace in both the public and private sectors. The principal research tool is human biological material, in the forms of human tissue and the human genetic information derived from examination of that tissue. It is now a rare day when the news media fails to contain some coverage of an exciting development or a worrying controversy (or often both) arising out of genetic research and technology.

The pace of scientific advancement in biotechnology and in other related fields creates a high level of social ambivalence about the potential benefits and detriments of change. On the one hand, there is very strong public support for breakthroughs promising better medical diagnosis and treatments, and for assisting with law enforcement, including identification of missing persons. On the other, there is some general anxiety about uncontrolled or 'mad science', the spectre of eugenics, threats of biological warfare, reports of xenotransplantation (transplants from one species to another), and fears about the increased potential for loss of privacy and genetic discrimination.

Recent public opinion surveys commissioned by the UK Human Genetics Commission³⁴ and the European Community³⁵ clearly demonstrate this tension, with generally strong support for the controlled use of genetic testing and information for diagnostic purposes, for scientific research leading to advances in treatment, cure and prevention, as well as for law enforcement applications. However, the surveys also register serious concern about such matters as reproductive cloning and the creation of 'designer babies'.³⁶ A large proportion of those surveyed felt they were poorly informed about developments in biotechnology - but most expressed a willingness to learn.

It is an important lesson for Australians that, because of recent crises in Europe over foot and mouth disease, mad cow disease, Creutzfeldt-Jakob Disease (CJD), genetically modified (GM) foods, human cloning, nuclear fallout from Chernobyl and perceived inadequate government and corporate responses to these events, Europeans manifest a high (and growing) degree of scepticism about the ability of public authorities to regulate biotechnology adequately in the public interest.³⁷

³⁴ Human Genetics Commission, 'Human Genetics Commission Publishes Results of Major Survey on Attitudes to Human Genetics', Press Notice 2001/0001 <http://www.hgc.gov.uk/business_press10.htm> at 2 March 2001.

³⁵ Eurobarometer 52.1, *The Europeans and Biotechnology*, <<http://europa.eu.int/comm/research/quality-of-life/eurobarometer.html>> at 1 October 2001.

³⁶ That is, where parents are able to 'dial up' preferences for gender, and for physical and other features.

³⁷ Compared with 29% who disagree, and 26% who are not sure: Eurobarometer 52.1, *The Europeans and Biotechnology* < <http://europa.eu.int/comm/research/quality-of-life/eurobarometer.html> > at 1 October 2001.

In 2000, the University of Western Australia's Survey Centre conducted a telephone survey of 1,000 people in that State,³⁸ 75% of whom reported that they were aware that genetic research was being conducted using human DNA. Eighty-four per cent stated that such research would benefit the community generally, and 70% thought it would benefit themselves or their families, especially by way of elimination of genetically linked disease (50%), cures for general diseases and better quality of life (26%) and fewer children being born with birth defects (7%). Forty per cent of respondents expressed concerns about associated risks and dangers with genetic research, especially in relation to 'inappropriate use of information' and the fear of eugenics.

Biotechnology Australia has also commissioned major quantitative and qualitative studies in this area, the latest conducted by the research firm Millward Brown, in April–May 2001.³⁹ This survey, which updated a similar study carried out in 1999, found that there was:

- Some level of concern expressed about gene technology by 80% of the community, but these concerns were rated much lower than environmental concerns, such as pollution or 'greenhouse gases'.
- An increased awareness of biotechnology issues in Australia (67%), and a general view that genetic engineering would improve our lives over the next 20 years (51%).
- An increased acceptance of some applications, such as modifying crops to make them more pest resistant (37%), testing embryos for predisposition to disease (25%) and using human genes in medicines and vaccines (29%).
- A decreased acceptance of using animal genes in plants (31%), or of modifying human genetic material with animal genes (44%), and an increase in the perceived risk of using human genes in animals to grow organs for transplantation (75%). Particular concern was expressed about cloning, with 58% stating they believed it would make things worse in the next 20 years.
- Significant concern that screening for genes that may cause incurable diseases could lead to discrimination (59%).

Perhaps most concerning is the high level of anxiety that is evident about the pace of biotechnological change and society's capacity to regulate it effectively (at least in part pushed along by serious concerns about human cloning):

³⁸ University of Western Australia Survey Research Centre, *Attitudes Towards Human Genome Epidemiology*, University of Western Australia <<http://www.gshh.uwa.edu.au/survey.html>> at 1 October 2001.

³⁹ Millward Brown Australia, *Biotechnology Public Awareness Survey Final Report*, Biotechnology Australia <<http://www.biotechnology.gov.au/mbsurveyresults.pdf>> at 1 July 2001.

Most respondents felt that biotechnology is changing at such a rapid pace that developments cannot possibly be anticipated and legislated against. In addition, it was generally felt that Australian society and government are powerless compared to the international financial and political power of the large multinational companies driving biotechnological innovations. A key component of concern was the perception that there are no or inadequate controls over the process, motivations and outcomes of the development and application [of] biotechnology and gene technology in Australia. This was particularly a concern for those applications which were seen to raise complex, and disturbing questions about human life.⁴⁰

However, the 2001 survey also found an *increased* level of trust in Australian government agencies as both a source of factual information and as regulators - in stark contrast to the experience in the UK and the rest of Europe, as related above.

THE SCOPE OF THE JOINT INQUIRY

The terms of reference for the joint inquiry specifically ask the ALRC and AHEC to inquire into and report on:

whether, and to what extent, a regulatory framework is required -

- to protect the privacy of human genetic samples and information; and
- to provide protection from inappropriate discriminatory use of human genetic samples and information; and
- to reflect the balance of ethical considerations relevant to the collection and uses of human genetic samples and information in Australia; and
- any related matter.⁴¹

This must be accomplished in a manner that has regard to the range of Australian ethical opinion on application of human genetic information, as well as the benefits and potential benefits of the scientific and medical applications of the new technology. The terms of reference also note the 'global dimensions of issues relating to research, regulation and the protection of interests'.

There are a number of potentially related areas that the ALRC and AHEC have determined fall outside the ambit of the present inquiry, including: genetically modified organisms (GMO foods); access to assisted reproductive technology; human cloning, stem cell research and clinical approaches to gene therapy; parental choices associated with prenatal testing; and intellectual property rights for gene patenting. This is certainly not to minimise the importance of these issues, some of which are already under consideration by other bodies, including the AHEC, Federal Parliamentary committees and the Standing Committee of Attorneys-General, which may deserve a separate inquiry.⁴²

⁴⁰ Ibid 29.

⁴¹ The full terms of reference are set out in IP 26 at 5-7.

⁴² For example, the ALRC and AHEC have written to the Attorney-General suggesting that the gene patenting issue is a matter of considerable importance, which should be the subject of a

Following the central issues stipulated in the terms of reference, the inquiry has developed its research and consultation program around four 'building block' areas. Thus, the Issues Paper provides, in some detail for public edification, material on:

- the role of ethics, specifically the sub-field of bioethics, in influencing medical practice and medical and scientific research on human subjects, and in protecting societal interests;⁴³
- the existing framework of Australian law protecting privacy - especially in relation to the privacy of health information, including genetic information, medical records and other health information. These laws include the recent landmark extension of federal privacy laws to the private sector;⁴⁴
- the existing framework of anti-discrimination laws and practices in Australia, with special reference to the possible application of disability discrimination law to the area of genetic information, as well as competing or complementary regimes governing such areas as occupational health and safety;⁴⁵ as well as
- a basic primer on the emerging genetic science and technology, especially in relation to the availability and use of genetic testing and the construction of genetic information.⁴⁶

The inquiry then considers the application of these building blocks across a broad range of contexts in which genetic information is used, or may be used in future. These include:

- *Medical and other human research.* The inquiry is examining ethical, privacy and related legal issues with respect to the use of genetic samples and information in the conduct of medical and other research involving humans. The inquiry is asking questions about how the current system, largely based on review of research proposals by Human Research Ethics Committees, operates in practice and whether the existing regulatory framework adequately protects genetic samples and information.⁴⁷
- *Human tissue banks, and genetic databases and registers.* The inquiry is looking at related issues involved in the collection, storage, use and disclosure of genetic samples and information held in human genetic databases, including in human tissue banks maintained by hospitals

separate review under fresh terms of reference. In his letter to the ALRC of 11 January 2002, the Attorney agreed that this matter is beyond the scope of the current joint inquiry, and indicated that the government would give consideration to the establishment of a separate inquiry.

⁴³ See IP 26, ch 3.

⁴⁴ See IP 26, ch 4; the new laws took effect on 21 December 2001.

⁴⁵ See IP 26, ch 5.

⁴⁶ See IP 26, ch 2.

⁴⁷ See IP 26, ch 6.

and public and private research organisations. This is an important area in which the interests of researchers - and ultimately the interests of society, since the research is aimed at achieving advances in medical diagnosis and treatment - must be balanced against the interests individuals may have in exercising control over their own genetic information.⁴⁸

- *Medical practitioners.* The inquiry is considering the role of medical practitioners in the protection of human genetic information. Medical practitioners provide advice on diagnostic and treatment options for genetic conditions, facilitate access to genetic testing, and provide advice and counselling on the implications and results of genetic tests. As health care providers, medical practitioners are important 'gatekeepers' of genetic information: they collect and store genetic information in health and medical records, and help to determine when and how genetic information is used or disclosed, and for what purposes. The inquiry is examining the existing regulatory framework that governs how medical practitioners handle genetic information, including the common law, legislation, guidelines and professional ethics.⁴⁹
- *Health administration.* The collection, use and disclosure of genetic information and samples raise important *systemic* health administration issues. These include health resource allocation issues raised by the increasing availability and potential use of genetic testing; population screening programs, such as newborn screening ('Guthrie cards'); and the development of electronic health record systems, including the proposed national health information network.⁵⁰
- *Employment.* The inquiry is considering the use of genetic information in employment, both in the workplace as well as in the process of applying for work. Generally, employers might seek access to an employee or job applicant's genetic information for the purpose of minimising risk in the workplace or to the public generally; to minimise their own business costs; or for occupational health and safety reasons. Indeed, there are strict common law and statutory obligations imposed on employers to maintain high standards of health and safety for all workers, as well as for customers and others. On the other hand, federal and state laws affirm the rights of individuals to be free from unfair direct or indirect discrimination in the workplace. Individuals may also be concerned about the privacy of their genetic information, and may desire assurance that employers will neither gain access to such information, nor pass it on to third parties, without their consent. There are difficult issues and balances to be struck in this area, particularly where genetic testing may reveal a susceptibility or predisposition to a medical condition, but the person concerned does

⁴⁸ See IP 26, ch 7.

⁴⁹ See IP 26, ch 8.

⁵⁰ See IP 26, ch 9.

not presently show any symptoms of that condition, and in fact may *never* develop the condition.⁵¹

- *Insurance.* Given the factors that led to its establishment (see above) - and the continuing strong interest shown in this area during public consultations - the inquiry is exploring in some depth the potential use of genetic testing and information for the purposes of underwriting personal insurance policies. Insurance companies, especially life insurers, have collected and used family medical histories for well over a century. In recent times, however, the development of the potential to use information derived from DNA analysis has placed a greater spotlight on the collection and use of personal information by the insurance industry in Australia and overseas. Among the specific issues being canvassed are: how scientific reliability and the actuarial relevance of genetic information should be addressed by the insurance industry and government; the impact that the use of genetic information in insurance may have upon health and medical research; whether genetic information is necessary for underwriting insurance policies; and equity of access issues related to the use of genetic information.⁵²
- *Access to other services and entitlements.* Given the *potential* for widespread use across a vast array of situations, the inquiry is considering a range of areas outside of employment and insurance in which genetic information *might* be used to determine eligibility for, or the provision of, goods, services or entitlements. For example, genetic information could be used to: determine eligibility for certain social security and training programs; used by hospitals to determine the allocation of scarce resources (such as organ transplants); used as part of immigration screening for proof of a family relationship, or the good health of an intending immigrant; used to prove ethnic or Aboriginal or Torres Strait Islander identity; used by school or nursing home authorities as a factor in determining admissions; or used by sporting bodies to determine whether a person is fit to participate.⁵³
- *Law enforcement.* DNA profiling is already a major tool for Australian law enforcement authorities. Contrary to the case with DNA testing in the clinical and research contexts, forensic testing is performed on non-coding or 'junk' DNA, with respect to a number of agreed core loci, to construct a unique DNA profile for identification purposes, such as for use in criminal investigations (to exclude or to help identify a suspect), in searches for missing persons, and in the identification of unknown deceased persons or body parts. Given its prevalence and widespread community acceptance, the inquiry is not re-thinking the basic policy issues involved in whether DNA profiling should be permitted. Rather, the inquiry is considering a number of issues of principle and practice

⁵¹ See IP 26, ch 10.

⁵² See IP 26, ch 11.

⁵³ See IP 26, ch 12.

in relation to the collection, storage and use of genetic information by law enforcement authorities, including the protection of ‘vulnerable persons’, and the development of the National Criminal Investigation DNA Database (NCIDD), operated by the federal agency, CrimTrac.⁵⁴

- *Evidentiary uses.* Finally, the inquiry is looking at the presentation and admissibility of DNA evidence in court, both in criminal matters as well as in civil proceedings (such as in relation to establishing paternity in a Family Court matter, or determining causation or the award of damages in a personal injury suit).⁵⁵

BALANCING OF INTERESTS

Careful consideration of the legal and policy issues thrown up by the use of genetic samples and information requires a wide range of sometimes conflicting interests to be balanced. The current methods of regulation and conflict resolution involve a patchwork of federal, state and territory laws; official guidelines; personal and professional ethics; institutional restraints; peer review and pressure; oversight by public funding authorities and professional associations; supervision by public regulatory and complaints-handling authorities; private interests; and market pressures.

The inquiry believes its primary brief is to scrutinise the existing regimes, and then tailor them - if necessary, and to the extent possible - to the particular needs and demands of genetic testing and information. Where appropriate, we will recommend new forms of regulation to meet any resulting gaps. Successfully fulfilling this brief not only involves providing adequate protections against the *unlawful* use of genetic information, but also putting into place measures and strategies aimed at ensuring that where such information may be used lawfully, it is used properly, fairly and intelligently.

There are valuable lessons to be learned from other challenges faced by the health care system, legal system and social services in recent times. For example, as a community, Australia has done better than most with respect to dealing with the outbreak of HIV-AIDS in such a way as to take seriously the medical issues and the risk of the spread of infection, while at the same time endeavouring not to stigmatise or discriminate against persons who are HIV positive, nor unduly breach their privacy.

As suggested earlier, the major challenge for this inquiry is to strike a sensible and effective balance which recognises the need to foster innovations in genetic research, technology and practice that serve humanitarian ends (for example, in the case of research) or social interests (for example, in the case of law enforcement uses), while at the same time providing sufficient reassurance to the community that such innovations are subject to proper ethical scrutiny and legal control. Unless the

⁵⁴ See IP 26, ch 13.

⁵⁵ See IP 26, ch 14.

Australian public feels safe and secure in this respect, there is a real risk of a serious loss of confidence in the regulatory apparatus - and then a resulting backlash against otherwise beneficial activities.

Although relatively easy to articulate, achieving that balance may be difficult in practice, since various interests will directly compete and clash across the spectrum of activity. For example, consider the following:

- *Genetic researchers* need ready access to a pool of genetic samples and research participants to assist in experimentation aimed at making important medical discoveries. However, the people whose samples, information or participation is required may have concerns about their ability to exercise informed consent or about the privacy of their genetic information. Potential volunteers may fear that participation in medical research experiments or screening programs will generate information that they subsequently may be required to disclose to insurers, employers or others.
- *Employers* must fulfil common law and statutory duties to provide a healthy and safe work environment for all employees. They also have an obvious interest in productivity, and in reducing insurance premiums and legal liability. However, employees or job applicants may fear discrimination if they are found to have a genetic disorder, or a genetic susceptibility to a disease triggered by specific environmental factors or substances that may be present in the workplace.
- *Doctors and hospital authorities* must often make very difficult decisions about diagnostic testing, therapeutic options and the allocation of scarce resources, and might be aided in this respect by access to genetic information. However, individual patients have legitimate concerns about privacy and the right to consent to genetic testing and to the use and disclosure of any genetic tests results.
- *Insurers* underwrite many forms of insurance policies by assessing the level of risk each person brings to the pool and may wish to use genetic test information for this purpose (in the same way that they currently use other health information, including family history). However, insurance applicants may be concerned that they will be denied insurance or offered a policy with loadings or exclusions on the basis of genetic information.

COMING TO TERMS WITH HUMAN GENETIC INFORMATION

Information about a person's genetic make-up can be derived from:

- diagnosing a genetic disease or disorder by clinical examination;
- studying a person's family medical history; or
- directly testing and analysing that person's DNA (or other biological substances, such as RNA or proteins).

Genetic information can relate to a condition that is:

- *clinically apparent* - such as when a genetic test is performed to confirm a diagnosis in someone who has signs or symptoms of a particular disorder. In these circumstances, genetic tests are not distinctly different in nature from other forms of clinical diagnostic testing (such as blood tests, MRI or CAT-scans), but may be more accurate and less invasive. For example, it would be preferable to use a genetic test to diagnose cystic fibrosis in an infant rather than by a (less accurate and more painful) sweat test; similarly, a genetic test for haemochromatosis is far less difficult for the patient than a liver biopsy.⁵⁶
- *latent* - such as when a genetic test is done on someone who is apparently free of a disorder at present, in order to determine the likelihood that he or she will, or may, develop the disorder in the future, or may be a carrier for the disease or disorder. Such tests obviously raise greater ethical and social concerns than the former category, and require considerably more attention given to privacy concerns and to providing pre- and post-test counselling and support services.

Genetic information is not only pertinent to an individual but may tell us something about close blood relatives - including those in succeeding and preceding generations. Thus, genetic information can be said to flow 'from before the cradle to after the grave'. For example, demonstrating that an individual is a carrier for cystic fibrosis means that one of that person's parents is also a carrier. In some cases, genetic information is pertinent to whole communities. Tay-Sachs disease is primarily (but not exclusively) found in persons of European Jewish descent; sickle cell anaemia primarily affects persons of black African descent; and haemochromatosis is very common in persons of northern European descent.

Information generated by DNA testing can be very precise - indicating that a particular mutation (allele) is, or is not, present. However, this precision will often prove unhelpful when it comes to predicting the future health of an individual. Genetic information tends to be about possibilities rather than certainties, because only a proportion of those people with a particular disease-related mutation or other variant will develop the disorder. For example, the so-called 'breast cancer gene' (mutations known as BRCA1 and BRCA2) is found in about one per cent of the female population and its presence is said to increase the risk of developing breast cancer by a factor of five. However, only 5-10% of breast and ovarian cancers result from the inheritance of alterations in BRCA1, BRCA2 and other familial gene alterations not yet known. Furthermore, estimates of the number of women

⁵⁶ See, for example, Human Genetics Society of Australasia, *Submission 6 to Senate Legal and Constitutional Legislation Committee Inquiry into the Provisions of the Genetic Privacy and Non-discrimination Bill 1998*, 11 May 1998.

with the gene alterations who actually develop breast cancer varies widely (from 36-85%), as do estimates for age of on-set.⁵⁷

For much of the past century, the prevailing orthodoxy was that ‘nurture’ (environment) is far more important than ‘nature’ (genes) in influencing human development, at least outside of the basic inherited physical traits. The pace and weight of genetic research in recent times, however, appears to have tipped common wisdom in the other direction - perhaps too far in the direction of genetic exceptionalism and determinism (see below).

In fact, the picture is far more complex. A person is not the sum of a column of traits and behaviours determined by individual genes; instead, it is better to think of a person as comprising of:

- the product of his or her genes;
- the intricate interaction of those genes; and
- the elaborate interaction between that genetic legacy and environmental factors.

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

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

IS GENETIC INFORMATION ‘SPECIAL’?

For many years, doctors, employers, insurers and others have requested information about family medical information - much of which amounts, in effect, to genetic

⁵⁷ James Evans, Cécile Skrzynia and Wylie Burke, ‘The complexities of predictive genetic testing’ (2001) 322 *British Medical Journal* 1052.

information - in order to provide advice or to make assessments about the future health of an individual.

Medical information already receives special treatment with regard to the new privacy laws and policies.⁵⁸ One of the key issues for the inquiry is whether genetic information is so fundamentally different from other forms of health information that it requires a separate or special regime to regulate its collection, use and disclosure.

Some of the characteristics that may differentiate genetic information from other forms of health information include:

- Every cell in a person's body, with the exception of sex cells, contains all of his or her genetic code. Therefore, the testing of any biological sample can reveal the full complement of a person's genetic information. There is also potential for stored genetic samples to be re-tested as new tests and techniques are developed, or our understanding of genetic conditions is advanced.
- As noted above, while each person's genetic information is unique,⁵⁹ it can also reveal information about - and therefore have implications for - that person's parents, children, siblings and other relatives (and as mentioned, perhaps even people outside the family, such as members of an ethnic group). Similarly, genetic information is capable of revealing 'family secrets', including information about paternity (or non-paternity), adoption, or the use of artificial reproductive technology.
- The predictive nature of some genetic information means that disclosure may lead to unlawful discrimination or other negative consequences for the individuals to whom it relates.
- The science relating to genetic information is new and developing, increasing the possibility that genetic information may be inaccurate or subject to misinterpretation. Individuals may not always be able to be advised about the long-term implications of this information.

Professors Annas, Glantz and Roche of the Boston University School of Public Health, the authors of the US Model Genetic Privacy Act (which strongly influenced the Stott Despoja Bill in Australia) have argued that genetic information is sufficiently unique and more powerful than other forms of health information so as to require special protection or other exceptional measures:

⁵⁸ See IP 26, at 132-145.

⁵⁹ Even 'identical twins' have minute difference in their genetic code.

To the extent that we accord special status to our genes and what they reveal, genetic information is uniquely powerful and uniquely personal, and thus merits unique privacy protection.⁶⁰

Annas, Glantz and Roche offer three justifications for this view. First, that a person's DNA 'can predict an individual's likely medical future for a variety of conditions'; indeed, they argue that one's DNA is a:

Coded probabilistic future diary because it describes an important part of a person's unique future and, as such, can affect and undermine an individual's view of his/her life's possibilities. Unlike ordinary diaries that are created by the writer, the information contained in one's DNA, which is stable and can be stored for long periods of time, is in code and is largely unknown to the person. Most of the code cannot now be broken, but parts are being deciphered almost daily.⁶¹

Second, that genetic information about an individual also 'divulges personal information about one's parents, siblings, and children'.⁶² Third, that there is a legitimate worry about the possibilities of genetic discrimination, since there is a history of genetics being used to stigmatise and victimise. Gostin also suggests that there are 'compelling justifications' for special privacy protection for genetic information, which are grounded in:

the sheer breadth of information discoverable; the potential to unlock secrets that are currently unknown about the person; the unique quality of the information enabling certain identification of the individual; the stability of DNA rendering distant future applications possible; and the generalizability of the data to families, genetically related communities, and ethnic and racial populations.⁶³

However, there are also some strong arguments that genetic information is not fundamentally different from other sorts of health and medical information, and therefore does not merit special treatment. On this view, genetic information is neither distinctive nor unique in its ability to predict an individual's future health, but indicates only a rough range of probabilities. Information about family history, lifestyle (smoker or non-smoker, skydiver or race car driver, miner or office worker) and non-genetic test results (eg for hepatitis, HIV, cholesterol) also provide important clues to current and future health.

Similarly, other non-genetic test results will contain very sensitive information, with the potential for causing distress, discrimination and stigma (such as a positive result for HIV-AIDS, tuberculosis, Hepatitis-B, or a sexually transmitted disease).

⁶⁰ George Annas, Leonard Glantz and Patricia Roche, 'Drafting the Genetic Privacy Act: Science, Policy and Practical Considerations' (1995) 23(4) *The Journal of Law, Medicine and Ethics* 360, 365.

⁶¹ *Ibid* 360.

⁶² *Ibid*.

⁶³ Lawrence Gostin, 'Genetic Privacy' (1995) 23(4) *The Journal of Law, Medicine and Ethics* 320, 326.

An individual's need for health care will depend on a complex mix of factors - both genetic and non-genetic - and most diseases and risks do not fall neatly into either category. Thus, it may be making an artificial distinction to regulate separately the handling of genetic information, as opposed to other forms of health and medical information, in all cases and contexts.

The Chair of the ELSI Task Force on Genetic Information and Insurance, Thomas Murray, has suggested that much of the drive behind genetic exceptionalism is based upon a generalised image of genetic information as 'a mysterious, powerful and inexorable force that will dominate and control our futures'.⁶⁴ Murray disputes the view that the predictive nature of genetic information compels special treatment, given that many other factors and forms of health information 'afford equally interesting predictions' and 'have implications for future health that are every bit as cogent and sensitive as genetic predispositions'.⁶⁵

Similarly, Murray also dismisses the argument based around the greater potential for discrimination, stating that:

Again, genetics is not alone. Institutions and individuals can and have used all sorts of information, both visible and occult, as the basis for discrimination. In underwriting for health insurance, for example, insurers use evidence of current disease or future disease risk - whether it is genetic or non-genetic doesn't matter - to decide who gets a policy, what the policy covers and how much it costs.

... Perhaps what really frightens and galls us about discrimination on the basis of genetic information is its reliance on information about us over which we have no control and may not even know ourselves. Here again it is the hidden and mysterious nature of genetic information, joined with its aura of power and ubiquity, lurking close beneath the surface of our discomfort.⁶⁶

In abandoning 'genetic exceptionalism', Murray writes that the Task Force ultimately concluded that:

there was no good moral justification for treating genetic information, genetic diseases, or genetic risk factors as categorically different from other medical information, diseases or risk factors. ... Our need for health care in most cases will be the product of a complex mix of factors, genetic and non-genetic, both within the scope of our responsibility and outside of that scope. The distinction between genetic and non-genetic factors is not the crucial one.⁶⁷

⁶⁴ Thomas Murray, 'Genetic Exceptionalism and "Future Diaries": Is Genetic Information Different From Other Medical Information?' in Mark Rothstein (ed), *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (1997) 64.

⁶⁵ Ibid.

⁶⁶ Ibid.

⁶⁷ Ibid 71.

Other questions arise from the fact that, until now, individuals and society have not had to deal with predictive information of such quantity and ostensible accuracy, and there is no considered community view about access to and use of predictive genetic information by family members and people or organisations outside the family. There may be greater pressures to gain access to and incorporate genetic information into decision making by others (such as employers, insurance companies or public authorities) where information about a person's future - even though imprecise or exaggerated - could be of importance. Genetic support groups, for example, have related many instances to the inquiry in which test information or the nature of a particular condition is misunderstood or mistakenly applied - even by health professionals - resulting in adverse consequences for the person concerned.

At the same time, genetic information has the corresponding potential to empower people to make choices about health for themselves and their families to a much greater extent than is the case with most other health information. Genetic testing for haemochromatosis, glaucoma, some cancers and other medical conditions can alert the individual to begin preventive measures before the disease causes harm.

Precisely because genetic information is familial in nature, much of it will come as no surprise; indeed, it can often provide great relief to those who receive the data. It is relatively rare that individuals learn of a risk through genetic testing that they did not already anticipate.

Further, there should be no implication that 'genetics' is about bad things; in truth, there is no such thing as an 'HD gene' or a 'breast cancer gene' - these genes are (in most of us) genes for health. Community and professional education and the ready availability of information when needed can minimise misunderstanding of, over-reaction to, and misuse of, genetic information.

BUILDING BLOCKS

As noted above, the inquiry is examining the current regulatory framework in relation to ethics, the protection of personal privacy, and protection against discrimination - in a number of specific contexts in which the use of human genetic information is, or may become, important.

THE TRENDS TOWARDS A NATIONAL APPROACH

One fundamental question is the extent to which a national approach to biotechnology regulation may be required, rather than relying upon the traditional mix of federal, state and territory laws.

To some extent, this shift is already taking place. For example, the extension of privacy protections to cover the private sector is being achieved through federal law, regulations and processes, and will be overseen by the Federal Privacy

Commissioner. Aspects of federal anti-discrimination law and industrial law already cover the field. Intellectual property rights for advances in genetics are determined according to federal laws and international agreements.

The location of regulatory authority in a federal system is always a matter of some contention. Uniformity has obvious advantages in terms of clarity and certainty. However, in a rapidly developing area of science and technology, there may also be something to be said for allowing innovation and experimentation on a state-by-state basis.

Public consultations and submissions thus far have consistently favoured the view that the protection of human genetic information is an area in which the public interest would best be served by a more uniform, national approach to regulation. However, given the wide array of activities covered by this inquiry, and the constitutional limitations on the exercise of federal legislative power,⁶⁸ only a cooperative approach involving the Commonwealth, the States and the Territories would assure the successful establishment of a comprehensive national scheme.⁶⁹

COMMUNITY CONSULTATION PROCESSES AND EXPECTED OUTCOMES

One of the main objectives of this inquiry is to promote community education and debate about the social, ethical and legal implications of developments in genetic research and technology. The inquiry is taking the approach that this is not an area to be left exclusively to experts and well-organised industry, professional or interest groups. The ALRC and AHEC have taken steps to raise community interest, public debate, education and promote public participation, by conducting public meetings which give the general public an opportunity to have direct input in the formulation of recommendations tabled by the inquiry. Following the publication of the Issues Paper, the inquiry conducted 16 public meetings in capital cities and major regional centres.

Public and media interest in this inquiry has been very high to date, with significant attendance at the public forums, a substantial volume of requests for the Issues Paper and other related materials, heavy use of the ALRC's website, many requests

⁶⁸ Section 51 of the Constitution specifies the areas in which the Commonwealth Parliament may legislate, such as with respect to interstate and international trade and commerce, taxation, defence, immigration, insurance, and intellectual property rights. Areas not specifically mentioned are reserved for the States - except to the extent to which the Commonwealth might hang the exercise of power on a general peg within s 51, such as the enforcement of treaty obligations under the external affairs power: s 51(xxix). The advantages of a dispersed, rather than a centralised, legislative power are outlined in Geoffrey de Q Walker, *Ten Advantages of a Federal Constitution* (2001) Vol 49, Centre for Independent Studies, Sydney.

⁶⁹ There are a number of ways in which this could be achieved technically, ranging from a referral of powers (most unlikely) to the adoption of uniform laws by each jurisdiction. The recent difficulties in achieving a national approach to corporate regulation that also survives constitutional scrutiny by the High Court indicates the traps in this area: see *Re Wakim; Ex parte McNally* (1999) 198 CLR 511.

for targeted meetings with expert and community groups, and many written submissions. As of 31 March 2002, the response to the Issues Paper has been encouraging, with over 150 written submissions received by the inquiry and quite a few more organisational submissions expected to arrive soon.

It is important to note that the final report of the inquiry will not be a self-executing document — the inquiry may only provide advice and recommendations about the best way to proceed, but implementation is a matter for others.⁷⁰

In an earlier era, the centrepiece of any significant law reform effort was the recommendation of a major new piece of legislation. However, in a more complex environment in which authority is much more diffused, modern law reform efforts are likely to involve a mix of strategies and approaches, including legislation and subordinate regulations; official standards and codes of practices (such as those promulgated by the NHMRC and the Privacy Commissioner); voluntary industry codes; education and training programs; better coordination of governmental (and intergovernmental) programs, and so on.

Similarly, although the inquiry's final Report will be presented to the Attorney-General and the Minister for Health and Ageing, it is likely that some (or many) of the recommendations will be directed to government departments and agencies; the NHMRC; the Australian Health Ministers' Conference; the Standing Committee of Attorneys-General; industry associations (such as IFSA); hospital and public health authorities; some of the Royal Colleges of medicine, as well as the various Divisions of General Practice; individual health practitioners; educational authorities; employer organisations and trade unions; and statutory authorities with responsibility for privacy and discrimination matters, among others.

It may also be the case that major advances in genetic science and technology will be so rapid that some of the bases for our policy-making in the report will be out of date in a relatively short span of years. Public consultations and submissions thus far strongly favour the establishment of a standing advisory committee to government on genetic science, along the lines of the UK Human Genetics Commission, the European Life Sciences High Level Group, and the Canadian Biotechnology Advisory Committee.⁷¹ If no standing body has been established to advise governments on these matters, it will not come as a surprise if the joint inquiry has to be reconstituted in future to revisit the issues.

⁷⁰ However, the ALRC has a strong record of having its advice followed. About 60% of the Commission's previous reports have been fully or substantially implemented, about 20% of reports have been partially implemented, and the remaining 20% have not been implemented or are sufficiently recent to be still under consideration.

⁷¹ See IP 26 at 94-99.