

## The protection of human genetic information

*With release of the ALRC/AHEC inquiry report, we are now in a position to develop sound policies*

THE REPORT *Essentially yours: the protection of human genetic information in Australia*,<sup>1</sup> launched in May this year, represents the first comprehensive exploration in this country of the ethical, legal and social implications of the emerging revolution in genetic science and technology. The report is the outcome of a major, two-year, public inquiry conducted by the Australian Law Reform Commission and the Australian Health Ethics Committee of the National Health and Medical Research Council (NHMRC).

Although the central themes of the inquiry were ethical standards, privacy protection and protection against unlawful discrimination, the final report examines the impact of the “new genetics” across a very wide range of social and professional contexts — accounting for the “super-sized” 1200-page document, presented in two volumes and containing 144 recommendations for reform. The inquiry covered obvious issues such as the ethical oversight of genetic research and the increasing use of DNA collection and testing by law enforcement authorities. Other questions considered by the inquiry included:

- the regulation of genetic testing in the workplace;
- the collection and use of genetic information by the insurance industry;
- genetic testing by immigration authorities;
- DNA parentage testing;
- the use of genetic testing as an element in the construction of kinship and identity; and
- the use of genetic testing to identify potential sporting champions.

This may sound like the stuff of science fiction, but the report documents contemporary cases and controversies in all these areas.

In the course of its extensive community consultation effort, the inquiry found significant optimism in Australia about the promised benefits of genetic science for improved diagnostics and therapies. However, there is also an underlying anxiety about the rapid pace of change and the capacity of our institutions to regulate science effectively in the public interest. Thus, the centrepiece of the recommendations is the establishment of a standing Human Genetics Commission of Australia (HGCA). The role of the HGCA would be to provide independent, high-level, technical and strategic advice to Australian governments, industry and the community generally about current and emerging issues in human genetics, and to provide a consultative mechanism for the development of policy statements and national guidelines in this area.

One of the threshold questions for the inquiry was whether to accept arguments in favour of “genetic exceptionalism”. This is the idea that genetic information is so fundamentally different from, and more powerful than, all other forms of personal health information that it requires different or higher levels of legal protection. In contrast, genetic “inclusivists” argue that genetic information is nei-

ther distinctive nor unique in its ability to predict an individual’s health, but indicates only a rough range of probabilities. The inquiry concluded that an exceptionalist approach would be unhelpful to the extent that it would divorce genetic information from the principles, processes and institutions that have been developed over time to provide ethical oversight of research and ensure best practice in clinical medicine. However, the inquiry accepted that genetic information has some special features and issues that necessitate a thorough inspection of existing principles, practices and safeguards, and of the legal, ethical and regulatory landscape, to ensure these are all adequate to the task.

The inquiry concluded that “big law” — an omnibus genetic regulation act — is inappropriate at this time. Nevertheless, the report makes a large number of recommendations for careful fine-tuning of existing legislation in the areas of privacy, discrimination, industrial law, and occupational health and safety, to meet the challenges of the new genetics. For example, it recommends that the federal *Disability Discrimination Act 1992* be amended “to clearly prohibit unlawful discrimination based on a person’s real or perceived genetic status”, and that the federal *Privacy Act 1988* be amended to cover genetic samples as well as data.

The report also strongly emphasises that we need not only adequate protection against the unlawful use of genetic information, but also measures to ensure that, where genetic information may be used lawfully, it will be used fairly and intelligently. As a consequence, the inquiry’s recommendations go beyond simply changing laws — they involve a broad mix of strategies and approaches, including the promulgation of ethical codes, codes of practice and official standards (eg, by the NMHRC and the Federal Privacy Commissioner); industry codes and best practice standards; community and professional education; and better coordination of governmental and intergovernmental programs.

Medical practitioners are well aware of how difficult it is to keep abreast of all the implications of the genetic information explosion. The report calls for all the parties involved in medical education (initial and continuing) to work collaboratively to greatly enhance genetics education for all doctors. The report also makes plain the increasingly important role that genetic counselling will play in everyday clinical practice. For some genetic tests, counselling will be adequately provided by medical practitioners; nevertheless, the report recommends that Australian healthcare authorities give urgent priority to assessing and responding to the need for increased, adequately resourced, genetic counselling services.

The inquiry recognised the powerful “familial dimension” of genetic information — that is, the extent to which an individual’s genetic information can also reveal information about, and therefore have implications for, that person’s relatives, including those in preceding and succeeding gen-

**Some specific recommendations of the Australian Law Reform Commission/Australian Health Ethics Committee inquiry**

- National ethical and privacy guidelines should be developed specifically to cover the use of genetic information held in tissue banks, research databases and genetic registers (including "inchoate" databases, such as Guthrie card collections).
- The support and guidance given to human research ethics committees when reviewing proposals dealing with genetic issues should be significantly strengthened.
- Laboratories that conduct genetic tests for medical, diagnostic or treatment purposes (rather than for research purposes) should be accredited by the National Association of Testing Authorities, and accreditation requirements should be strengthened to deal more broadly with ethical standards in genetic testing, such as proof of consent.
- The Therapeutic Goods Administration should be empowered to more effectively regulate medical devices used in genetic testing, as well as DNA test kits provided directly to the public, whether such kits are marketed for health purposes or for identification (such as for parentage testing).
- Nationally consistent standards should be developed in relation to population genetic screening programs, covering such matters as informed consent, testing standards, quality assurance, cost-benefit considerations, and reporting and data collection.
- Employers should not be permitted to collect or use genetic information in relation to job applicants and employees, except in rare and compelling circumstances. Such circumstances might be when this is necessary to protect the health and safety of workers or third parties, and the action complies with stringent standards developed for this purpose by the HGCA and occupational health and safety authorities.

permitted to disclose confidential information to genetic relatives without the patient's consent, if such a disclosure is necessary to lessen or prevent a serious threat to an individual's life, health or safety. In this sensitive area, the inquiry asks that guidelines be developed to assist healthcare professionals in this task.

Some other recommendations of particular interest to the scientific and medical communities are summarised in the Box.

The inquiry's findings and recommendations have been presented to the two relevant federal Ministers — the Attorney-General and the Minister for Health and Ageing — and the government is expected to respond and outline its plans for implementation soon. However, the pervasive influence of genetic science means that recommendations for change have been addressed to more than 30 bodies across the public and private sectors — many of these organisations do not need to wait for the federal government before they can take action.

We have an excellent opportunity in Australia now to develop policy based on sound principle, rather than managing emerging problems on the run. The area of genetic testing and information is so personal and so sensitive that it is critical we get this right — and do so now — to avoid the crisis of confidence and the public backlash that would inevitably follow from the revelation of poor or unethical practices.

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erations. This leads to a recommendation that, despite the traditional importance of confidentiality to the doctor-patient relationship, there may be exceptional circumstances in which doctors (and familial cancer registries) should be

1. Essentially yours: the protection of human genetic information in Australia. ALRC Report 96. Canberra: Australian Law Reform Commission and Australian Health Ethics Committee, 2003. Available at: <http://www.alrc.gov.au/publications/final-reps.htm> (accessed Jul 2003). □

**MJA/Wyeth Award 2002**

The 2002 MJA/Wyeth Award went to "Sharing the true stories: improving communication between Aboriginal patients and healthcare workers", which was published in the 20 May 2002 issue of The Medical Journal of Australia.

At the recent AMA National Conference, Dr Alan Cass accepted the Award's commemorative plaque from Dr Kerryn Phelps, AMA President, and a cheque for \$10 000 from Ms Erica Mann, Managing Director of Wyeth Australia.

Dr Cass and his coauthors, Anne Lowell, Michael Christie, Paul L Snelling, Melinda Flack, Betty Marrnganyin and Isaac Brown, are from the Cooperative Research Centre for Aboriginal and Tropical Health, Darwin. The award was for their research into factors limiting the effectiveness of health communication between Aboriginal patients and healthcare workers.

Despite the ever increasing influence of science and technology in medicine, what has remained unchanged from antiquity is the patient-doctor consultation. At its centre is effective communication, which is influenced by language, context, and culture, and no more so than in the interaction between Indigenous people and their healthcare workers.



Left to right: Dr Anne Lowell, Ms Betty Marrnganyin, Dr Martin Van Der Weyden, Dr Alan Cass, Ms Erica Mann, Dr Kerryn Phelps

As noted by Ms Mann: "Australian Indigenous culture encompasses a vast tapestry of experiences, language and mythology that both enriches our lives and presents considerable hurdles in communication about healthcare from a traditional non-Indigenous medical perspective".

Cass and his fellow researchers set out to determine barriers that may impede this culturally laden communication, using qualitative research, which included videotaped interactions. Their research showed that, in Aboriginal healthcare, full understanding in the clinical context was rarely achieved. Barriers included lack of control by Aboriginal patients over the timing, content, and circumstances of the predominantly biomedical interactions; cultural and linguistic divides; limited use of interpreters; and lack of training opportunities in cross-cultural communications.

Commenting on the research, Ms Mann said: "There is a great and continuing need for research like this, if improvements in Aboriginal health are to be sustained and [Aboriginal Australians] are to approach the quality of life enjoyed by non-Indigenous Australians".