

ESSENTIALLY YOURS: THE PROTECTION OF HUMAN GENETIC INFORMATION IN AUSTRALIA – THE IMPACT ON CLINICAL PRACTICE AND THE ‘NEW GENETICS’

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I BACKGROUND

The ‘new genetics’ refers to the application of DNA-based knowledge to clinical practice. It is only 50 years since Watson and Crick reported in April 1953 that DNA was a double helix, and in this form it could function as the source of genetic information in the cell. From this emerged molecular biology and the ‘new genetics’. Today, health professionals and the community are faced with many new discoveries in genetic medicine, as well as the challenges which go with these developments.

Another milestone was the Human Genome Project (‘HGP’) which started in 1990, and finished in 2000. The HGP was considered unachievable by many because of the difficulties associated with sequencing 3.3×10^9 bases which make up our DNA. However, technological advances, including partnership with industry, produced a complete sequence of the human genome as well as those of many model organisms five years earlier than originally planned. These sequences are now in the databases waiting for researchers to analyse them. The information coming from the HGP will reshape future clinical practice, not just in clinical genetics, but in a wide range of clinical and para-clinical disciplines.¹

Prior to the HGP we spoke about the new ‘genetics’. Today this term is becoming outdated as ‘genomics’ takes over. With genetics, the focus was on single gene disorders and single genes. With genomics, we now have the tools to look at the complete genome and how it interacts. Some extend the definition of genomics to include the study of genes and their interactions with the environment. The rapid developments will provide many opportunities but also uncertainties. Challenges that must be addressed to maintain high standards of clinical care will include the education of the community and health

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1 Francis S Collings, Eric D Green, Alan E Guttmacher, Mark S Guyer, ‘A Vision for the Future of Genomics Research’ (2003) 422 *Nature* 835.

professionals. The legal profession will need to be informed if it is to arbitrate in times of dispute. For the future it is essential that bright minds as well as the best motivated graduates are attracted to clinical practice in the genomic era. How is this to be achieved?

A start has been made with the Australian Law Reform Commission's ('ALRC') final report entitled *Essentially Yours: The Protection of Human Genetic Information in Australia* ('ALRC Final Report').² The recommendations in the ALRC Final Report have far reaching effects, and if implemented appropriately, will ensure that the genomic era provides Australians with better and more affordable health care. The Report has produced 144 recommendations, and was tabled in Parliament in May 2003. It is now being considered by the Commonwealth Government. Two major themes have emerged from the Report, and these will be discussed separately before recommendations of particular relevance to clinical practice are reviewed.

II MAJOR THEMES

A Human Genetics Commission of Australia

The Report recommends the establishment of an independent statutory authority, the Human Genetics Commission of Australia ('HGCA'). In view of the rapid changes occurring in genomics, and the Australian Federal structure, there is little doubt that such a body could make a substantial contribution to community and professional awareness of genomics, as well as define clearly the standards and guidelines by which clinical practice will proceed when genetic information is involved.

In Australia, there are many active groups in genetics. These include the various societies and professional colleges. There are strong genetic support groups represented by the Association of Genetic Support of Australasia. These bodies have lobbied governments, both State and Commonwealth, to address various 'hot topics' in genetics. A difficulty with the Federal system is the delineation of responsibility between the Commonwealth and States, increasing the potential for difficult issues to remain unresolved. An example of this was the recent *BRCA1* (breast cancer gene) patent dispute when Genetic Technologies, an Australian company, indicated that *BRCA1* testing in this country could only be carried out in the company's laboratory in Melbourne or Myriad Genetics' laboratory in the United States ('US'). Similar prohibitions were being enforced in other countries with variable responses – some acquiesced whilst others threatened legal action because patient care was being compromised. The Australian response was slow and initially uncoordinated. In NSW, laboratories

2 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia*, Report No 96 (2003).

involved were adopting different stances. Finally, some forward progress was initiated and today the ALRC is undertaking an enquiry.³

A long-standing issue which remains unresolved is the funding of DNA tests since the vast majority are not covered by Medicare. The user-pay option is a poor model for genetics since inappropriate use of DNA tests is harmful not only to the patient but to others in the family. As well as determining a mechanism that will fund DNA tests, it is equally important to link this mechanism to a realistic and health-relevant approach in deciding which DNA tests are needed. On current estimates there are about 35 000 human genes. The genes that are clinically relevant and worthwhile pursuing to improve health outcomes will need to be determined.

B Substantial Legislative Changes are not Needed

The ALRC Final Report recommends against the introduction of major legislative changes, but indicates that current laws and frameworks can be amended to cover most issues that would emerge. This is particularly relevant to the genomics era because there will be substantive changes in our understanding of genes and how they work. New innovative technologies will be developed. The Internet will be an option for disseminating information or soliciting samples of DNA. A lot has been said about the potential for DNA tests to discriminate against individuals. However, the extent of this problem remains to be determined. Perhaps just as important is the question of whether the occurrence of discrimination constitutes malicious misuse of genetic information or ignorance? A frequently quoted example of discrimination by an employer is *Equal Employment Opportunity Commission v Burlington Northern Santa Fe Railroad*, a case in which DNA testing was started to detect employees with a genetic predisposition to carpal tunnel syndrome.⁴ However, if the case is examined from the genetics perspective, what the employer did had no scientific validity. The better educated employer would have saved the court's time, and the company's money.

Recently, there was a dramatic news item indicating that a US-based fertility expert had cloned a human. Immediately, the reporter on television indicated that this could not happen in Australia because we have a law prohibiting cloning. However, globalisation is not constrained by laws. What the reporter should have said was that this was an unfortunate development, if correct, because there is increasing evidence that animals cloned by somatic cell nuclear transfer ('SCNT') have developmental malformations thought to reflect the inability of SCNT to reprogram complex regulatory controls of genes.⁵ Without a well

3 ALRC, *Intellectual Property Rights over Genetic Materials and Genetic and Related Technologies*, Issues Paper No 27 (2003).

4 The railroad company settled out of court. The agreement provided that the railroad company would no longer require its employees to submit blood for genetic tests and would pay US\$2.2 million in damages: <<http://www.eeoc.gov/press/4-18-01.html>> at 20 November 2003. See Wylie Burke, L Pinsky and Nancy Press, 'Categorizing Genetic Tests to Identify their Ethical, Legal and Social Implications' (2001) 106 *American Journal of Medical Genetics* 233.

5 Ramiro Alberio and Keith Campbell, 'Epigenetics and Nuclear Transfer' (2003) 361 *Lancet* 1239.

informed community, laws will not be effective because individuals can get on a plane to have a procedure, or send DNA in the mail to have a DNA diagnostic test. The latter is already available through the Internet, in some cases without the requirement for medical referral.⁶ A further consideration is that globalisation disadvantages those who cannot afford to travel and/or pay for the procedure or test.

III SPECIFIC RECOMMENDATIONS

A Privacy and Duty of Care

The conventional clinical environment is exclusively focused to one individual – the patient. Genomic medicine is different. Any information obtained about an individual's genes must apply to other family members who will invariably share some of the genetic information. Hence, the concept of genetic privacy needs to be revisited. Related is the health professional's duty of care. The doctor advising a patient with a 50 per cent risk of an autosomal dominant genetic disorder has a duty of care to that person. However, upon finding that the patient has the relevant mutation in the DNA (and so will develop the disorder), the health professional is now faced with the dilemma that first degree relatives (in particular, siblings and children) of that patient are now at the same risk (50 per cent) that the patient had when he or she first walked into the consulting room. How wide does the health professional's duty of care extend, and what would be expected of the health professional in terms of follow up of family members? Follow up is clearly important but has the potential to impact on the privacy of many individuals in the family (including the patient who is seeing the doctor). These are not theoretical issues but a part of everyday clinical practice. The ALRC Final Report provides the first opportunity to address potential dilemmas outside the more difficult medico-legal environment.

B Regulation of Human Genetic Research

The cornerstone of genetic research in Australia is the Human Research Ethics Committees ('HREC'). In terms of genetics these committees face two major challenges: first, the volume of work that is expected of them; and secondly, the potential ethical and social issues that some research protocols raise. In this environment, the institutionally based HREC has a lot on its plate. Cutting-edge research related to new genome discoveries will place additional demands on the HREC. The ALRC Final Report has identified some of the pressures on HRECs and appropriate recommendations have been made, some of which are already being implemented.

6 Bryn Williams-Jones, 'Where There's a Web, There's a Way: Commercial Genetic Testing and the Internet' (2003) 6 *Community Genetics* 46.

C Practice Guidelines and Standards

In Australia there are various national accreditation bodies that oversee standards. In particular, the National Association of Testing Authorities ('NATA') and the National Pathology Accreditation Advisory Council ('NPAAC') define laboratory practices. There is formal accreditation of hospitals, and the various professional colleges monitor continuing medical education. What is lacking in the clinical practice area, particularly in genetics, are guidelines for common scenarios particularly in relation to DNA testing. For example, how many cystic fibrosis mutations are reasonable to test for in determining a couple's risk for having an affected child? (There are over 1000 in theory but it would not be feasible to test for all.) While this may seem a trivial question it remains an uncertainty that is not needed in an area of medical practice that is already moving so fast that tension is likely. The ALRC Final Report makes recommendations about practice guidelines, and the implementation of these will reduce the potential for litigation as well as unnecessary distress to patients and families because something could have been done, but it was not made available for various reasons.

A topic to address in the near future will be community DNA screening. At present, there is considerable debate about this issue. However, what is relevant in the US may not apply to Australia with our particular health care system and the very mixed ethnic communities in some geographic regions. The latter is relevant since risks for various genetic diseases as well as the DNA mutations likely to be found are often linked to ethnicity. In this environment, forward and effective planning and dialogue with the community will avoid the unfortunate earlier experiences of the sickle cell screening programs in African Americans and the stigmatisation that these produced.

D Education

A key recommendation relates to the education of the medical students, medical practitioners and other health professionals likely to be involved in caring for patients and families with genetic disorders. This is a complex issue. The reason for saying this is that genetics presently focuses on single gene disorders such as Huntington's disease, haemochromatosis and cystic fibrosis. In these cases, the modes of inheritance and the basis for disease are straightforward. The genomics era will complicate this by taking on the multifactorial disorders where genes and environment interact to produce the important contemporary health issues mentioned earlier. These conditions will require diagnosis, surveillance and treatment as well as an understanding of the genetics. The only way this will be possible is via an educated community and educated health professionals working as a team.

Bland statements that all health professionals need to know more about the new genetics are not enough. New skills will need to be developed whether the

health professional is a GP, specialist physician, obstetrician, or counsellor.⁷ Once acquired, these skills will need regular updating. The colleges and the societies must be involved, but from past experience the driver will need to come from outside. The HGCA will certainly have many challenges but none greater than education. Novel ways will be needed since the information from genomics is too rapidly changing for traditional educative tools. The Internet is vital and already there are key resources available. For example, most professionals will refer regularly to the latest update in Online Mendelian Inheritance In Man, a resource that is reputable and updated as soon as there is a new development.⁸

7 The *Medical Journal of Australia* has published six articles between 21 April and 4 August 2003. These represent the opinions of a wide range of health professionals on the impact that the new genetics will have on their practice.

8 Online Mendelian Inheritance in Man <<http://www.ncbi.nlm.nih.gov/Omim/>> at 20 November 2003.