

Discussion Papers:

1. The Use of Genetic Information for Insurance Purposes
2. Genetic Testing
3. Human Genetic Research
4. Human Genetic Research Databases

The following discussion papers have been prepared by the Ethics Committee of the Western Australian Genetics Council (WAGC). They aim to provide a concise overview of the issues in the areas selected, and have a significant focus on outlining the pertinent ethical issues involved. The need to develop government perspectives on topical issues in human genetics was outlined in the Australian Law Reform Commission (ALRC) Report 96 *Essentially Yours: The Protection of Human Genetic Information in Australia*. Comment on these papers is welcomed and should be directed to the Genomics Directorate.

April 2004

1. The Use of Genetic Information for Insurance Purposes

The increasing use of predictive genetic test information has prompted concerns about the potential for genetic information to be used by third parties, such as insurers, to discriminate against individuals on the basis of their genetic makeup.

The Use of Genetic Information for Life or Disability Insurance

A contract of insurance is one of good faith, as stipulated in section 13 of the *Insurance Contracts Act 1984* (Cth).¹ Persons seeking insurance therefore have a duty to disclose every matter they are aware of that will be relevant to the insurer in making the decision.² Thus, under current disclosure requirements, information gained through genetic testing is treated in the same way as other medical information in that applicants must reveal relevant details from tests they have already undertaken. The failure to disclose relevant information or the making of misrepresentations may invalidate the contract.³ Importantly, there is no obligation to disclose genetic information which comes to light after the insurance contract has been entered into.³

Current anti-discrimination legislation in Australia provides exemptions for insurers which allow them to legally discriminate against an individual on the basis of the medical information they provide, as long as there is statistical or actuarial evidence to support this decision.^{1,3} These exemptions recognise that differentiating between individuals is fundamental to the market in mutually rated insurance products where the decision making process is reasonable.¹

The insurance industry also plays a role in regulating the use of genetic information for underwriting purposes, and industry standards have been developed to this end. The Investment and Financial Services Association (IFSA) have a *Policy on Genetic Testing* (1999) to regulate the collection and use of genetic test results. Under this policy, applicants must disclose any existing test results, and this information can be used in underwriting. However, the policy stipulates that life insurers cannot request applicants to undergo a genetic test, or coerce applicants to take a genetic test by offering preferred risk underwriting to those who have favourable genetic status.⁴

The insurance industry argue that a departure away from a system of equality of information between applicants and insurance companies raises issues of fairness. It has been suggested that a possible implication of allowing high risk individuals to join insurance pools at standard rates is that the expected increased claims by those individuals must ultimately be borne by others.¹

Genetic Discrimination

The concern about the use of genetic information as a component of the underwriting process for life and disability insurance is the potential for discrimination. Discrimination could include unfavourable underwriting decisions, including the charging of higher premiums, longer waiting periods, incentives of lower premiums for those who decide to undertake a genetic test, and the possibility for people who are

shown to be at increased risk through genetic testing to be denied or disadvantaged when accessing insurance.⁵ It is important to note that none of these currently occurs.

Reports of alleged misuse of genetic information for insurance purposes have prompted the development of industry guidelines on the issue, as well as consideration at the legislative level through the joint Australian Law Reform Commission (ALRC) and National Health and Medical Research Council (NHMRC) inquiry into the use of genetic information.¹ To date it is difficult to assess whether the behaviour complained of amounts to unlawful discrimination.⁵ There is uncertainty about the nature and extent of discrimination in this area and a need for further detailed empirical research, which is currently being conducted by the Genetic Discrimination Project Team.^{1,7}

Whilst it may be legal for insurance companies to use genetic information, there is still concern that the information will be used beyond the extent permitted by legislation.⁵ There is evidence that this is occurring in some jurisdictions overseas, and this is largely in response to insurers misinterpreting genetic information.³ Genetic information is complex; many aspects related to precise risk interpretation are often very broad especially with issues concerning penetrance (degree of certainty that a condition will manifest itself in the future), and the degree of severity of a condition, if indeed it should arise.

Impact on Health Care

There is evidence to suggest that the threat of genetic discrimination in insurance, be it real or potential, can impact upon public health outcomes. It has been reported that some patients hesitate to consult clinical genetics services due to fear of negative consequences for insurance.¹ If genetic test information were allowed to be used for the matter of life or disability insurance, it may act as a deterrent for people to find out family health details, possibly to an individual's detriment because they can not access management from which they could well benefit.⁸ It is important that health professionals provide individuals with sufficient information to enable them to understand the implications of genetic test results beyond applicability to their own health outcomes, so that the benefits of genetic testing may be harnessed, and the disadvantages minimised.

Risk Assessment

Insurance companies need to be especially considerate of the manner in which information gained through genetic testing is interpreted and weighed on an actuarial basis. In terms of the scientific reliability of genetic test information, it should be noted that genetic tests, whilst useful in distinguishing between those who have a gene mutation and those who do not, are less accurate at identifying who will actually go on to develop the disease.

In relation to actuarial relevance of the test information, actuaries must take into consideration that the expression of a genetic disorder on an individual's mortality or morbidity is variable, particularly in instances where there are effective treatments available. In calculating this risk estimate, actuaries must calculate the risk that an applicant with a particular condition will make a claim based on health data collected

from large numbers of individuals. At present there is very limited actuarial, statistical or other data available to allow use of genetic test information for underwriting purposes. In future insurance companies may need to become more transparent about the way in which they assess genetic information and how they have used the information to reach their decisions for individual applications. Further, actuarial models which consider the potential preventative health benefits both to the community and the insurance industry would be a positive move forward into the future.

The ALRC Report *Essentially Yours: Protection of Human Genetic Information in Australia* (2003) recommended for establishment in the future, a Human Genetics Commission of Australia (HGCA) to provide independent oversight of the use of predictive genetic test information in insurance. Responsibility for this could be shared between this committee and the government, which would be a similar approach to that taken by the Genetics and Insurance Committee (GAIC) established in the United Kingdom.

Recommendations

In light of these issues, it is recommended that:

- The foremost use of predictive genetic information should be for the benefit of the health and wellbeing of the individual and not be used to cause any social or economic disadvantage;
- Genetics health professionals should counsel patients to understand the potential for any information gained through genetic testing to be used by third parties, particularly insurers;
- The processes utilised by insurance companies to assess genetic test results need to be fair and transparent;
- Ethical guidelines should be developed to ensure that those with a genetic predisposition to disease are not unfairly disadvantaged; and
- If the existence of, or a result of a predictive genetic test has been declared by an individual, such information should not be used to assess the insurance risk of any other family member.

Furthermore, if insurers are allowed to use genetic test information to underwrite policies, it is vital that they also investigate other actuarial models which will allow those with a genetic predisposition to disease to access insurance.

Options could include:⁶

- Introducing a threshold for which applications under the threshold amount are exempt from family history and genetic testing questions;
- having policies that allow coverage for all eventualities other than the genetic illness for which the person is at risk; and
- It may be appropriate to have policies with a time limit in some instances.

References

1. Australian Law Reform Commission and National health and Medical Research Council. (2003). Report 96. *Essentially Yours: the Protection of Human Genetic Information in Australia*.
2. Otlowski, M. (2001a). *Implications of Genetic Testing for Australian Insurance Law and Practice*. Occasional Paper 1, Centre for Law and Genetics. University of Tasmania and the University of Melbourne.
3. Otlowski, M. (2001b). Avoiding genetic discrimination in insurance: An exploration of the legality and ethics of precautionary measures in anticipation of unfavourable test outcomes. *Monash Bioethics Review*, 20 (1), 24-32.
4. Investment and Financial Services Association Limited (IFSA). (1999). *Policy on Genetic Testing*. Available at: <http://www.ifsa.com.au/IFSAWeb/ifsapubl.nsf/CommonShow/Publications+Display?OpenDocument&Headline=IFSA's+Genetic+Testing+Policy&URL=/IFSAWeb\common.nsf/AllDocsNF/17302528EDDC5A8BCA256A470024828C>
5. Otlowski, M. (2001c). Is there scope for lawful genetic discrimination in health insurance in Australia? *Journal of Law and Medicine*, 8 (4), 427-432.
6. Human Genetics Society of Australasia (March 1999). *Policy: Predictive Genetic Testing and Insurance*. Available at: <http://www.hgsa.com.au>
7. Otlowski, M., Taylor, S., Barlow-Stewart, K. (2002). Australian empirical study into genetic discrimination. *Genetic Medicine*, 4 (5), 392-395.
8. Lerman, C., Hughes, C., Lemon, S., Main, D., Snyder, C., Durham, C., Narod, S., & Lynch, H. (1998). What you don't know can hurt you: adverse psychological effects in members of BRCA1 linked and BRCA2 linked families who decline genetic testing. *Journal of Clinical Oncology*, 16 (5), 1650-1654.

2. Genetic Testing

The Australian Law Reform Commission Report (ALRC) 96: *Essentially Yours: Protection of Human Genetic Information in Australia* has outlined a number of areas relating to genetic testing that require short term consideration. These include issues concerning accreditation, the development of ethical guidelines, and access to genetic testing.

Laboratory Accreditation and Ethical Issues

At present laboratory accreditation with National Association of Testing Authorities (NATA) is the desired national standard for laboratories. This accreditation sets forth a minimum set of standards for laboratory practice, which provide assurance to the public that the testing produced meets all technical requirements. Not all laboratories are accredited, and primary concerns regarding non-accredited laboratories include there being no independent assessment about the quality of the testing process, the proficiency of the personnel carrying out the test, or the reliability of the test results¹. Further, non-accredited laboratories do not offer client reimbursement on Medicare rebate items. At present there are only three human molecular gene tests on the Medicare Benefits Schedule (MBS), and others are paid for either by the client or funded through State laboratories, which is a strong commercial disincentive to do most tests.* Therefore, until appropriate regulation measures for testing provision are in place, it is in the community's best interests for tests to remain off the MBS.

Whilst present accreditation standards impose a wide range of general and specific management and technical requirements on laboratories, they do not make recommendations on ethical issues associated with testing. These issues include protocols for obtaining consent and maintaining the protection of privacy for individuals. Guidance on ethical issues involved in testing are provided through the National Health and Medical Research Council (NHMRC) in their document *Ethical aspects of human genetic testing: An information paper*² and through other allied organisation, such as the Human Genetic Society of Australasia (HGSA) and the Australian Medical Association (AMA)⁴.

Areas in need of particular ethical guidance include the adequacy of consent to testing, privacy and confidentiality, and ensuring that individuals are tested only in circumstances where they will be provided with proper information and advice in interpreting the test results, and access to genetic counselling. This is particularly so with *predictive* genetic tests which are carried out in presently well family members, and provide definitive risks of disease in the case where particular gene changes are present. Further guidance is required for these areas, and it would be appropriate for the NHMRC or the proposed Human Genetics Commission of Australia (HGCA) to develop these.

The ALRC has recommended that:

- the Commonwealth, States and Territories should enact legislation to require laboratories to: (a) be accredited for any genetic test that they conduct for medical,

* This does not take into account cytogenetic tests.

diagnostic or treatment purposes; and (b) comply with the relevant accreditation standards; and

- for National Pathology Accreditation Advisory Council (NPAAC), in consultation with the Human Genetic Society of Australasia (HGSA), National Association of Testing Authorities, Australia (NATA) and the Royal College of Pathologists of Australasia (RCPA), should examine how compliance with its accreditation standards in relation to consent, counselling and other ethical considerations in medical genetic testing should be assessed as part of the NATA/RCPA accreditation process.

These recommendations are supported.

Consent to Genetic Testing

Requests for genetic testing will come from a variety of sources from the person to be tested to referral by the health professional. Individuals presenting for testing must be willing to undergo the test, and unless required by law, there should be no compulsion on any person to undertake any genetic test.^{4,6}

Whilst testing has great potential to confer important health information that may lead to positive health outcomes, the information gained through genetic testing also has important psychosocial implications, may result in social stigma, or have important implications for third parties such as insurers or employers¹. In light of these issues, genetic testing should not be undertaken in the absence of appropriate pre- and post-test counselling by a qualified professional. Such counselling often requires appropriate time, training and facilities and is also an important vehicle through which information about the test, its results and implications is imparted. Laboratories should be satisfied that all the appropriate pre-test requirements have been fulfilled before commencing any test procedure.

Testing of children and adolescents raises particular ethical issues and the HGSA position paper on this should be consulted³. Of particular concern with regard to the testing of minors is the potential for non-consensual testing in which the children's interests are not adequately represented. The ALRC has recommended the introduction of legislation to protect against non-consensual genetic testing, and this reflects the seriousness of the issue, and how this type of testing violates individual autonomy and privacy.

Additionally, it is currently possible for laboratories who are NATA accredited to conduct non-consensual testing. It is recommended that NATA develop a policy to address this issue, and that this policy prevents laboratories who are NATA accredited from performing non-consensual genetic testing.

Access to Genetic Testing Services

Access to genetic testing services should be equitably available for all people.^{2,4}

Direct to Public Testing

Genetic test kits or results provided direct to the public raise particular ethical concerns and warrant considerable caution. There is increased potential for harm from individuals

accessing genetic information in this manner, as there is no counselling provided, no assistance in interpreting results and a lack of credibility for the results. Further there is no quality assurance measures undertaken to Australian standards for these laboratories¹.

The ALRC has recommended that the HGCA develop codes of practice and advice relating to technical and ethical standards for genetic testing services provided directly to the public. This recommendation is supported.

Other Ethical Issues

Genetic testing raises a number of other ethical issues that need to be considered. Some of these are outlined below.

Storage

Information gained through genetic testing, either from research or clinical purposes, will be stored indefinitely by laboratories, as a component of good laboratory practice.⁵ Individuals having predictive, presymptomatic, susceptibility and carrier tests should be informed that the test result will be retained by the testing laboratory².

Institutions or organisations wishing to conduct research on genetic material collected for clinical testing should develop and disseminate a general policy which informs patients that such material may be used for future research following HREC approval. Refer to NHMRC National Statement with this section².

When genetic material was collected initially for research, the individual concerned should be informed that storage is planned and consent should be obtained for any future uses other than that for which the genetic material was obtained².

Privacy

Information gained through genetic testing has important implications for the individual. The Department of Health's *Code of Ethical Practice for the Provision of Genetic Services in Western Australia* makes the following recommendations with regard to protecting privacy of genetic test information:

- The privacy and confidentiality of genetic information should be respected and disclosure of this information to third parties without the consent of the individuals concerned is not generally ethically acceptable.
- In some situations, the genetic information about an individual may be of clinical significance to their genetic relatives. Those undergoing testing should be advised to carefully consider with whom the test result should be discussed before testing takes place. Health professionals should encourage, to the greatest extent possible, the sharing of this information.
- It may be ethical for health professionals to share genetic information with genetic relatives of an individual without that individual's consent in instances where the health of others is at serious risk, and / or where disclosure is likely to prevent serious harm, and where treatment or prevention is available.
- Health professionals need to work within the regulations set forth by appropriate state legislation.

- There is no formal duty for health professionals to warn genetic relatives of any risks associated with the genetic status of their relatives.

Further guidance on these areas is provided by the NHMRC *Ethical aspects of human genetic testing: An information paper*.

Relevant guidelines and regulations

National Association of Testing Authorities, Australia (NATA)

<http://www.nata.asn.au/>

Royal College of Pathologists of Australasia (RCPA)

<http://www.rcpa.edu.au/public/default.cfm>

Therapeutic Goods Administration

<http://www.health.gov.au/tga/>

National Pathology Accreditation Advisory Council (NPAAC)

<http://www.health.gov.au/npaac/>

- Standards for Pathology Laboratories
- Quality Systems for Medical Laboratories
- Requirements for Supervision of Pathology Laboratories
- Laboratory Accreditation Standards and Guidelines for Nucleic Acid Detection Techniques

Human Genetic Society of Australasia (HGSA)

<http://www.hgsa.com.au/>

- Prenatal Diagnosis Policy (July 2001)
- Privacy Implications of Genetic Testing (March 1999)
- Predictive Testing in Children and Adolescents (2003)

National Health and Medical Research Council (NHMRC)

<http://www.nhmrc.gov.au/>

- Ethical Aspects of Human Genetic Testing: An Information Paper.

References

1. Australian Law Reform Commission and National health and Medical Research
2. Council. (2003). Report 96. Essentially Yours: the Protection of Human Genetic Information in Australia.
3. National Health and Medical Research Council. (1992). Ethical aspects of human genetic testing: An information paper.
4. Human Genetic Society of Australasia. (2003). Predictive Testing of Children and Adolescents.
5. Australian Medical Association Position Statement: Human Genetic Issues. (1998).
6. National Health and Medical Research Council. (1998). Retention of laboratory records and diagnostic material.
7. Human Genetics Society of Australasia. (March 1999). Privacy Implications of Genetic Testing.

3. Human Genetic Research

Human genetic research is an important avenue through which advancements with the potential to improve health are made. Whilst the completion of the Human Genome Project will provide many research opportunities, research also raises ethical issues which highlight the need for research to be conducted in a manner which is transparent and accountable, to ensure that ethical practice is observed and potential for harm is minimised.

National Statement on Ethical Conduct in Research Involving Humans

The National Health and Medical Research Council (NHMRC) policy entitled *National Statement on Ethical Conduct in Research Involving Humans* ('National Statement') sets standards for ethical practice of research. This document specifically outlines issues concerning human genetic research. Key issues include:

- Social significance of the research- genetic research results may be of significance to the health of genetic relatives, including some who have not participated in the research, and for collectives;
- Privacy and confidentiality- there exists potential for harm, including stigmatisation or unfair discrimination, and researchers need to exercise special care to protect the privacy and confidentiality of human genetic information; and
- Counselling- there may be instances where research will reveal important health information for identifiable or potentially identifiable individuals and in these cases counselling must be provided for in the research protocol.

The National Statement is due to be formally revised by NHMRC during the 2003-2005 triennium, consistent with NHMRC policy of revising guidelines that have been in place for five years. A number of specific areas in the National Statement which require re-examination include the protection of participant privacy, consent procedures, de-identification of genetic data, and the storage of data.¹

There are several areas of human genetic research which are being extensively debated at the present time. These include compliance with the National Statement, the maintenance of best practice in research and the protection of participant privacy.¹

Compliance with the National Statement

At present only publicly funded research organisations are formally required to comply with the National Statement, and there exists the potential for private sector research to be conducted outside of the ethical parameters promoted by the NHMRC. It should be noted that the importance of public sector investment in research is significant and most research conducted in the private sector does comply with requirements in the National Statement.¹

Nevertheless, it would make for stronger ethical practice to ensure that all human genetic research is conducted in accordance with nationally accepted guidelines, these being the National Statement, and the mechanisms by which compliance with the National

Statement is enforced should be strengthened to require that all researchers and research organisations comply.

Furthermore, additional guidance on specific protocols, such as mechanisms for coding samples, to guide Human Research Ethics Committees (HREC) are welcomed if developed by the NHMRC.

Consent and the Protection of Privacy

Consent is a basic and essential component of ethical conduct in research. Consent must be given by the participant, where competent, or by a person acting with lawful authority on behalf of a participant who lacks competence. Consent must be based on information about the purpose, methods, demands, risks, discomforts and outcomes of the research and it must be voluntary and not impaired by any coercion, inducement or influence.²

It is important that consent requirements achieve the appropriate balance between protecting participant rights and not inappropriately constraining research.

Waiver of Consent

In limited circumstances, and taking into consideration a number of factors, both the National Statement and the Commonwealth *Privacy Act* (through sections 95 and 95A which relate to information only, not tissue samples) recognise that the use of human genetic information without consent for research purposes may be justifiable. Waiver of consent issues often arise where researchers propose to access and use material collected for therapeutic or diagnostic purposes (such as pathology samples) in circumstances in which the individuals from whom the samples came may not have consented to the use of their samples in research.¹ Ideally a waiver of consent should only take place in exceptional circumstances, however the practicality of conducting research also needs to be considered by the HREC.

In terms of reporting waiver of consent cases, it would be appropriate for HRECs to report to the Australian Health Ethics Committee (AHEC) with respect to human genetic research proposals for which waiver of consent has been granted under the National Statement, and not just for those granted under s 95 and s 95A guidelines of the *Privacy Act*.

Consent for Unspecified Future Research

The National Statement provides that consent to the collection of human tissue for research purposes should be specific to the purpose for which the tissue is to be used. Where it is proposed that human tissue samples previously collected and stored with consent for research be used for a different research purpose, separate consent for the different research should be obtained.²

The key ethical issue in consent for unspecified future research is the validity of obtaining consent where people cannot be informed in any detail about the nature of the research. It should be ensured that future research is related to the research to which individuals donated their information in the first instance. This is a particularly

challenging issue for ethics committees to resolve and in these instances HRECs are encouraged to access relevant information on the topic to inform their decisions.

The ALRC Report 96 has recommended that the National Health and Medical Research Council, as part of its review of the National Statement in the 2003-2005 triennium, should review the mechanisms for achieving compliance with the National Statement, with particular regard to human research conducted wholly within the private sector.

Guidelines and Regulations

The current key relevant pieces of Australian guidance and information for human genetic research may be found on the NHMRC website (<http://www.nhmrc.gov.au>) and include the following policies:

- Guidelines for Genetic Registers and Associated Genetic Material (2000)
- National Statement on Ethical Conduct in Research Involving Humans (1999)
- Ethical Aspects of Human Genetic Testing- and information paper (2000)
- Human Research Ethics Handbook (2001)
- Guidelines of Ethical Matters in Aboriginal and Torres Strait Islanders Health Research (1991)
- Guidelines for Ethical Review of Research Proposals for Human Somatic Cell Gene Therapy and Related Therapies (2000).

References

4. Australian Law Reform Commission and National health and Medical Research Council. (2003). Report 96. *Essentially Yours: the Protection of Human Genetic Information in Australia*.
2. National Health and Medical Research Council. (1999). *National Statement on Ethical Conduct in Research Involving Humans*.

4. Human Genetic Research Databases

Human genetic research databases are increasingly becoming valuable sources of information for researchers. The types of research conducted using human genetic databases include:¹

- Linkage studies to identify the gene sequences associated with inherited diseases
- Association studies to find correlations between a disease and a genetic change where there is no obvious pattern of inheritance
- Genetic epidemiology studies of the interaction between genes and environment
- Pharmacogenetic studies to determine if there is a genetic basis for certain adverse reactions to drugs.

The possibilities opened up by the storage of large quantities of genetic information on research databases raise numerous ethical issues and may cause concern for many people. Concerns relate primarily to the way in which personal information is collected, used, stored, disclosed and transferred.^{1, 2} Strengthening current regulations would go some way towards ensuring high ethical standards are maintained in the administration of databases.

Regulation

Regulation of human genetic research databases currently consists of:

- The Commonwealth *Privacy Act (1988)* and similar state privacy legislation;
- State and territory Human Tissue Acts;
- Ethical guidelines contained in the National Health and Medical Research Council (NHMRC) *National Statement on Ethical Conduct in Research Involving Humans* ('National Statement'); and
- Human Genetic Society of Australasia (HGSA) *Guidelines for Human DNA Banking*.

The National Statement sets the current standard for ethical administration of research databases. This document outlines the necessity of maintaining respect for persons, the situations where consent would be required or could be waived, the importance of confidentiality, and the responsibility of institutions where the databases are held to develop policies about the conduct and ethical approval of research which conform to relevant legislation and are consistent with the National Statement.³

However, as outlined below there are some areas not covered by current regulation, and ethical guidelines need to be formalised in order to ensure that databases are subject to appropriate governance and accountability.

Ethical Issues

The ethical concerns which most urgently need addressing relate to the protection of privacy, consent including for unspecified future research, and the use of stored samples and information for secondary purposes.

Privacy

Human genetic research databases raise many privacy concerns due to their storage of large quantities of genetic samples and information that may be accessed by many different researchers, over many years, and for many different research purposes.¹ Key privacy concerns relate to issues such as consent to storage, use and re-use, linking of information to samples, and the extent of disclosure of samples and information.¹

At present a primary issue is that in some circumstances it may not be clear which privacy legislation, if any, will apply where researchers affiliated with hospitals, or universities are funded by an external source or work in collaboration with private sector organisations.¹ It may be assumed that where the organisation who holds the database is covered by privacy regulations, the database comes under the same regulations, regardless of funding sources. There is clearly a need for clarification on this issue.

As genetic databases store personal information that could be predictive of an individual's health, they must be regarded as health databases and come under privacy legislation covering that researcher or organisation. They should be declared in the privacy statement of that researcher or organisation and access provisions for individuals should apply.

There has also been suggestion that greater national consistency needs to be achieved in privacy regulation covering human genetic databases. This may be an achievable aim in the near future.

It is also important that consent requirements achieve the appropriate balance between protecting participant rights and not inappropriately constraining research that is deemed to be in the public interest.

Consent

There is a presumption that an individual should normally give consent to the collection and use of their genetic data, and that no data will be collected, stored or used without consent or used for purposes other than those for which consent had been obtained. For existing genetic databases where consent to use the material or information for research purposes has not been given, access should only be granted to identifiable information with the consent of the donor, or under the usual circumstances, with the approval of the HREC.

Whilst many human genetic research databases already operate by obtaining consent to unspecified future research, further guidance is required on the mechanisms for obtaining consent to unspecified future research, particularly in the context of standing research databases.¹

There will also be many instances in which retrospective consent must be sought. This is likely to arise where databases have been developed from existing collections, from samples and information left over from clinical use, and from the conversion of clinical databases to research use.

Use of genetic samples and information for secondary purposes

In some instances there may be a call to use the samples and information stored in research databases for secondary purposes, such as law enforcement. The key ethical issue here is that the individuals donating this information did not consent to these secondary purposes. The use of any genetic samples or information for secondary purposes, such as law enforcement, should be regulated by national policy to ensure ethicality is maintained. Further, where such disclosure without consent takes place, it should be recorded and monitored.

Recommendations

The National Statement is due to be formally revised by NHMRC during the 2003-2005 triennium, consistent with NHMRC policy of revising guidelines that have been in place for five years. It would be appropriate for revisions of the National Statement to include more guidance on some of the distinct privacy and ethical issues raised by human genetic research databases in order to deal more adequately with these issues.

The Australian Law Reform Commission (ALRC) has extensively researched this area and its final report *Essentially Yours: Protection of Human Genetic Information* concludes that current regulation is inadequate and requires reform. This report makes a number of recommendations for reform, primarily through the National Statement, the most salient being:

- There should be uniformity across States and Territories in the registration, use and auditing of human genetic research databases;
- Clarification is required on the mechanisms for obtaining consent for unspecified future research;
- Guidelines on the circumstances on which it may be acceptable for the genetic information stored in research databases to be used for secondary purposes should be added;
- Provisions that require institutions to register their human genetic research databases with the NHMRC should be included. This would enable comprehensive information on databases to be available to the Australian Health Ethics Committee, and therefore ensure that transparency is maintained;
- There should be greater consultation with stakeholders regarding the proposal to appoint an independent intermediary to control information used to identify data and samples held within a database.

There should be a nationally agreed set of criteria under which a gene trustee *must* be used, for example, for excess pathology specimens. For other uses, the relevant HREC should have the power to determine when a gene trustee *should* be used. The

gene trustee would have the authority and resources to monitor compliance, reporting and auditing of individual human genetic research databases.

References

1. Australian Law Reform Commission. (March 2003). Report 96: *Essentially Yours: Protection of Human Genetic Information in Australia*.
2. UNESCO. (2003). *Draft International Declaration on Human Genetic Data*.
3. National Health and Medical Research Council. (1999). *National Statement on the Ethical Conduct of Research Involving Humans*.