

## Review Article

# Ethical Considerations in the Use of DNA for the Diagnosis of Diseases

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### **Abstract**

Scientific advances in genetics have recently provided new information and enabled new interventions that are challenging existing ethical conventions. ISO 15189:2003<sup>1</sup> obliges the laboratory to consider its ethical responsibilities and the AACB (through membership of the IFCC) has taken a leading role in the discussion of evolving new ethical frameworks.

This paper discusses the ethical implication of many of these recent advances in genetics and highlights some of the still unresolved ethical issues.

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### **Introduction**

The identification of genes and their location are being added to the databanks at a rapid rate: currently around 19,500 genes of the estimated 30,000 genes in the human genome have been mapped to individual chromosomes.<sup>2</sup> Of these, 1,647 have now been identified as being involved in a genetic condition.

From these findings, genetic tests have been developed, many of which have now moved from research to clinical laboratories although the number of available genetic tests is still quite small. Within Australia, almost all genetic testing carried out for health reasons is conducted in a medical setting with the assistance of professional genetic counselling to interpret the results and ensure that the testing was undertaken with informed consent. Some of these tests can determine, with some limitations, if a person is at increased risk of developing certain conditions later in life, such as some forms of cancer, haemochromatosis<sup>3</sup> and some neurological conditions such as early onset Alzheimer disease. In rare cases, such as with Huntington disease,<sup>4</sup> testing can determine whether or not the person will definitely develop the condition later in life (if they live long enough).

### **Implications of Genetic Testing**

The application of genetic testing can offer individuals and families new information and, with that, potential new

freedoms. Even where there are few or no current treatment options, for example, in conditions such as Huntington disease, genetic tests can still enable those who are at risk due to their family history to make life choices. For conditions such as breast and bowel cancer, a positive test means that early detection strategies can be considered. For example, an individual with a positive genetic test for predisposition to haemochromatosis may be advised to regularly monitor their iron levels. Genetic testing may also offer reassurance where someone who is concerned that they are at high family risk of developing a condition is given genetic information confirming that they are not at such risk after all.

Along with these freedoms there are potential burdens that can come from knowing one's genetic information, as well as new choices that may need to be considered. Decisions may need to be made about the termination of a pregnancy or about lifestyle changes. A person may be found to be at risk of an untreatable condition or a condition that requires intensive surveillance to enable early detection for treatment. Other potential burdens include the creation of a new patient group called the 'worried-well', who may experience increased anxiety in spite of the fact that they may never develop the genetic condition about which they worry. Genetic knowledge may also impact on family dynamics since test results on an individual may mean other family members are, or are not,

identified as being at risk. Those who have a positive genetics test result may also have to communicate sometimes complex and difficult information to other family members. Other burdens include the potential impact on their insurability and employability.

Genetic conditions are family health problems, for the identification of an individual as being affected or being at risk means that other blood relatives may now also be known (or discoverable) to be similarly at risk. A tested individual may refuse to share their genetic information or may choose to be selective in how it is shared. Commercial organisations such as insurance companies and employers may also have strong interest in the future health of a currently asymptomatic person who has had a genetic test and they may seek, or even be able, to exert power over such individuals in certain circumstances. This raises the spectre of 'genetic discrimination', defined as unjustified treatment on the basis of an asymptomatic person's genetic make-up. The need for public policies and regulation in regard to this issue were comprehensively addressed by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) in their report 'Essentially Yours'.<sup>5</sup>

Both positive and negative emotional and psychosocial responses can occur as a result of a genetic test.<sup>6</sup> Positive responses include the feelings of reassurance where they have not inherited the faulty gene involved, or often relief and empowerment even if they have. Removal of uncertainty is often liberating. For many, finally having a name or label to explain their constellation of symptoms and therefore knowing even to some degree what the future may hold, brings enormous relief. In our world of scattered families, the need to communicate even bad news can facilitate and strengthen communications that have not formerly been fostered.

The availability of a prenatal diagnostic test, or the receipt of a negative genetic carrier, predictive or presymptomatic test result can restore reproductive confidence. Such confidence can also be restored where the prospect for treatment or surveillance is available or possible. Similarly, utilisation of genetic testing is more acceptable when treatment is available. In the case of Huntington disease, for which there is currently no treatment, uptake of testing is only about 20% worldwide<sup>7</sup> while with testing for haemochromatosis, where treatment, surveillance and prevention is available and simple, uptake is increasing.

On the other hand, a genetic diagnosis or a positive genetics test result can generate grief: blame, guilt, shame, denial, anger and depression are often expressed. Family dynamics can be severely impacted: "It didn't come from my side of

the family"; "there are bad genes in her/his side of the family". Even when a person is shown to be negative for a particular genetics test result, they can suffer from the guilt of having 'escaped' and can feel that they are no longer part of the family sharing a bond of being 'at risk'. Limitations can be placed on expectations of a child or family member and having a label or name for a condition and therefore the range of symptoms that can occur can sometimes be a self-fulfilling prophecy. Most damaging can be the removal of hope and dreams for the future. Treatment and surveillance may be available but involve difficult choices: a preventive mastectomy will necessarily impact on a woman's perception of self. For some, a positive genetics test result does not deliver them from uncertainty: there is most often lack of correlation between genotype and phenotype. If the condition is rare, little information may be available about the prognosis or path that the condition will take. Uncertainty is compounded when clinical signs indicate that, for example, cancer is running in the family, but a mutation cannot be found in the genes analysed.

Social issues can also impact on this decision: termination of pregnancy is still difficult to talk about openly and the community may be polarised on the ethics of this choice. A genetic diagnosis in one family member means that the affected person, or a parent, becomes the bearer of bad news: decisions must be made as to who to tell (children, grandparents, other family members, work colleagues for example), when to tell and how much to tell. Genetic counselling needs to include discussion of the possible reactions that the individual may experience as well as how others may react. The support systems that are in place or may need to be developed need to be explored and may include discussions of respite, the financial implications for the family and the potential for discrimination. Information about support groups is essential, as they are an excellent resource in this area.

Conflict may exist between the scientific ('biomedical') information provided relating to the genetic causes of the condition or its heritability and the beliefs and values held by the person or family: for example, karma, fate or ancestral retribution may be given as explanations for the cancer in the family.<sup>8</sup> This conflict may be a barrier to dissemination of the information to 'at risk' relatives or acceptance of recommended surveillance strategies or treatment. Reluctance to discuss particular illnesses, such as cancer, or to document relatives who may have been affected for fear of contagion may lead to inaccurate documentation of a family history and inaccurate triaging for genetic counselling.<sup>8</sup> Non-Western concepts of kinship may also inaccurately inform family history.<sup>8</sup> In addition, perception of disability is different in different communities and choices made about termination of pregnancy for disabilities unacceptable in certain cultures

may challenge the values held by the genetic counselling team. However the basic tenet of genetic counselling is non-judgemental support of the client's decisions. Cultural beliefs about the time at which life begins will also impact on the stage of pregnancy at which termination of pregnancy will be accepted.

### **Why is Clinical Biochemistry different to Clinical Genetics?**

Although the origins of clinical biochemistry are strongly linked to the practice of clinical medicine, and notwithstanding that many clinical biochemistry departments retain clinical consultative services, the reality is that the vast majority of modern diagnostic biochemistry is performed in a laboratory without direct contact with the patient.

Clinical biochemists perform their analysis in an environment of some isolation, often relying on the referral letter or the request form, deficient of clinical information in many cases. It is not unusual for significant analysis to be performed at the level of analyte, enzyme, gene product or even at the level of the gene itself, relying on the original request form and the clinical assessment of the primary consultant and initiating referrer of the specimen. Perhaps for this reason, the prevailing culture in clinical biochemistry is that there is nothing 'special' about genetic testing, as distinct from other types of testing. After all, a genetic diagnosis (such as Wilson disease or cystinuria) can be made using classical biochemistry without resorting to direct genetic testing, and it does not seem sensible to accord a different level of 'specialness' if the diagnosis was made using genetic analysis compared to making the same diagnosis using biochemical techniques.

There will probably be a convergence of these views in the near future: clinical biochemists will need to understand the shared nature of genetic information, and the greater sensitivity and privacy issues associated with genetics information. Conversely, the practical experience of the clinical biochemistry community, and the demonstrated lack of harm (and indeed, the considerable benefit) that has often arisen from prompt, dispassionate laboratory analysis will probably remove the 'preciousness' attributed to genetics testing merely because it is genetics.

### **Ethical Issues Associated with Genetic Testing**

The same genetic test can be used clinically for different purposes and the implications and considerations will differ according to that purpose. The same test may be used to confirm clinical symptoms (diagnostic) or determine if a person with a family history of the condition will definitely develop the condition in later life (presymptomatic) or will be at increased risk of developing the condition (predictive). Genetic tests

can also be used in pregnancy. The general principles that underpin genetics testing in all of these contexts include:

- Patient identification
- Consent
- Understanding the limitations of the test
- Testing of children
- Confidentiality
- Duty to inform

These are discussed in more detail in the following sections.

#### **Patient Identification**

Specimens collected from patients need to meet minimum standards of identification. In Australia, the National Pathology Accreditation Advisory Council (NPAAC) standards require two forms of identification, usually name and date-of-birth, or name and medical record number (or similar hospital identifier). However, standards for specimen collection and patient identification<sup>9</sup> are higher for Blood Transfusion than for other forms of diagnostic pathology.

The Royal College of Pathologists of Australasia (RCPA) is currently reviewing its policy on specimen identification. The key issue is that a genetics diagnosis may have life-long significance, and may impact on other members of a family beyond the original patient. The question then arises whether standards for sample collection for genetics testing should be of a higher standard than for 'ordinary' pathology testing? Or should the standards for specimen collection for *all* specimens be at least the standard required of, for example, a transfusion specimen. In considering this question, it must be realised that the diagnosis of genetic conditions can be made by other than a genetics test, and this in turn implies that the standards of specimen identification for a sample not intended for a genetics test may need to be raised to the same standards as for a genetics test. Note that the ability to extract DNA from stored clinical specimens will also raise a question in the future of the standards of patient identification and specimen collection that applied at the time the original specimen was collected, not merely the contemporary standards that may apply at a future time.

#### **Consent**

Consent must be given by a legally competent person and signed consent will be required if testing is conducted through a clinical genetic service. Testing should only be undertaken when the individual has been fully informed about the purpose of the test or the procedure and the possible implications of the results. The consent must be given freely without coercion by professional staff, family members, employers, insurers or others; and the person must be adequately informed about all relevant issues including available future options. The person may withdraw consent for the test at any time. Laboratory

accreditation in Australia requires adherence to NPAAC Standards,<sup>17</sup> which define in greater detail the nature of informed consent, and how this may differ depending on the nature and implications of the genetics test being requested.

#### Understanding the Limitations of the Test

An essential component of the consent process is addressing the limitations of the genetic tests. Presymptomatic or predictive tests do not give information about the age of onset or the severity of the tests. Where there is low sensitivity of the genetic test, or not all the genes involved have been identified, the interpretation of a negative result may be limited.

#### Testing of Children

Guidelines of the Human Genetics Society of Australasia state that children, in general, should only have presymptomatic testing when the resulting information will be used to help with their health management in the immediate future, and not simply because parents wish to know.<sup>10</sup> However, the age at which presymptomatic/predictive testing can be offered to a child should be given flexible consideration by the testing team. Where risk status for a condition has been established for a child, either prenatally or after birth, the child should be informed that the information is available once he/she has reached a level of maturity consistent with understanding its implications.

An asymptomatic at-risk child's DNA should not be collected and stored for research or for possible future use by the child or the family.

#### Confidentiality

The results of a genetic test may be of interest to third parties including other family members. The highest levels of confidentiality need to be applied regarding the results, and the fact that a person has undergone testing.

There is a common practice in genetics laboratories to regard the results of laboratory testing as being confidential to the patient and their immediate attending medical practitioner. The referral laboratory may often decline to send the results of genetic testing to a referring laboratory, instead merely reporting that "analysis has been completed", and instead choosing to send the report directly to the attending medical practitioner. This will be discussed in more detail later in this article under the section discussing the appropriate addressee of the laboratory report.

#### Duty to Inform

Genetic conditions are family health problems as the identification of an individual as affected or at risk means that other blood relatives may be at risk. This shared nature of

genetic information means that the outcome of genetic testing can have a significant impact not only on the individual being tested but also on other members of their families. The family however is responsible for informing their members of the genetic test results and their implications.

This may be difficult and patients may need assistance and counselling on how best to approach and inform relatives of their potential risk. In rare cases, a tested individual may refuse to share their genetic information or be selective in how it is shared. However, current policies limit disclosure of confidential information by health professionals only where the condition is seen to be serious and imminent<sup>3</sup> and genetic conditions do not meet these requirements.

#### Laboratory Issues Associated with Genetic Testing

In Australia, the standards for laboratory accreditation are set by the NPAAC. Since 1<sup>st</sup> July 2005, all medical testing laboratories are required to be certified to ISO 15189:2003<sup>1</sup> (also known as AS4633-2004) as a condition of eligibility for medical reimbursement. This requirement is not as enforceable as it might appear at first, for very few genetics tests are eligible for medical reimbursement at present. The Australian Health Ministers Advisory Council (AHMAC) has recently authorised a consultancy study to advise the Governments of Australia whether *all* laboratories should be required to be accredited for genetic testing, and not just those seeking medical reimbursement.

ISO 15189:2003 contains an Annex (Annex "C") that relates to ethics in Laboratory Medicine. It requires the laboratory to place the patient's welfare as its primary consideration, but recognises that the laboratory may be in a third party relationship to the patient due to the referral of the specimen to the laboratory. The Standards specifically state that this referral relationship does not allow the laboratory to abrogate its responsibilities to the patient.

#### Access to Testing

Access to genetic testing should normally be through a clinical service, such as a clinical geneticist, other medical practitioner or a genetic counsellor. Laboratories are not permitted to tout genetic tests directly to the public, or even to accept referrals directly from the public without the patient having the benefit of access to an appropriate clinical service.<sup>17</sup> An important exception to this principle are Community Genetics Programs,<sup>11</sup> in which genetic tests are offered directly to the public, but as part of an integrated education, laboratory and clinical support program.

#### Types of Genetic Tests

NPAAC has proposed classifying all molecular genetic tests

into two broad groups<sup>17</sup>: Level 1 (standard DNA test) and Level 2 (DNA test with potential complex issues) (Table).

Level 1 tests are “standard” tests (such as testing for haemochromatosis, which is now commonplace in many clinical biochemistry laboratories), while Level 2 tests are those that may lead to more complex ethical or clinical circumstances.

For Level 2 tests, the primary health professional attending the person seeking genetic advice is responsible for ensuring that informed written consent is obtained and for providing

or referring to appropriate pre-test and post-test professional genetic counselling.<sup>17</sup> In turn, this creates an obligation on the laboratory to ensure that it is satisfied this is likely to have been done. For example, where the specimen has been referred from a consultant known to be expert in the field, it is likely that they would be aware of their obligations in regard to such informed consent, but where the referral has come from a new, less experienced or casual referrer not likely to be familiar with such matters, the ethical obligation on the laboratory would require it to satisfy itself that such consent has been obtained.

**Table.** Draft classification of types of genetic tests by the NPAAC in guidelines for laboratory accreditation<sup>17</sup>

Classification*	Description
Level 1 DNA test (standard)	Included here would be: a) DNA testing for diagnostic purposes (e.g. the patient has clinical indicators or a family history of an established inherited disorder, and DNA testing is being used to confirm the disorder) or any other DNA test that doesn't fall into level 2. b) Neonatal screening programs.
Level 2 DNA test (i.e. the test has the potential to lead to complex clinical issues)	DNA testing for which specialised knowledge is needed for the DNA test to be requested, and for which professional genetic counselling should precede and accompany the test. Predictive or presymptomatic DNA testing, for conditions for which there is no simple treatment, would usually be included in this grouping. Specific written consent and counselling issues are associated with this grouping.

\* The distinction between Level 1 (standard DNA test) and Level 2 (DNA test with potential complex issues) would usually be made by the doctor ordering the test, since that individual will be best placed to appreciate the short-term and long-term implications of the test for the patient and other family members.

1. The previous description of DNA testing for inherited genetic disorders — which used Class A and Class B categories — has not been followed in this version of the NPAAC standards and guidelines. Although some type of categorisation is necessary, feedback indicated that Class A and Class B were confusing, particularly when examples were given, because there were inconsistencies in reasons for choosing a particular class and an example could move from one class to the other, depending on use and circumstances. It is not possible to rigidly assign disorders to either Class A or Class B, and the definition of a ‘serious genetic disorder’ can vary, depending on the circumstances.

2. In using the above Level 1 (standard DNA test) versus Level 2 DNA test (complex issues) categorisation, no examples are given, because the implication of the test will vary. Thus, a test may be standard in one particular circumstance but complex in another. This confusion will only be resolved as knowledge about DNA genetic testing and its implications increases in the health profession.

For Level 1 tests, the laboratory may often be the party collecting the specimen, and the standards of informed consent are broadly similar to those of 'ordinary' biochemistry tests. However, as genetic tests are relatively new, health professionals may not be as knowledgeable of these as for many other tests, and the laboratory may need to establish educational and training procedures to ensure its referrers and its collecting staff have access to information or other resources sufficient to be able to answer questions that the patient may wish to ask, so as to ensure informed consent has been obtained.

### **Professional Genetic Counselling**

Genetic counselling is more than simply providing genetic testing. The counselling process guides the patient in making use of the information generated by their family history or a test; enables discussion of the options presented in the context of their beliefs and values and facilitates decision-making considering the perception and understanding of the risks and burden of the condition.<sup>12</sup>

There are three main areas where professional genetic counselling is essential: involving or following the diagnosis of a genetic condition that may be at birth, in childhood or adulthood; before and after predictive or presymptomatic genetic testing for conditions such as cancer and neurological conditions for which treatment and/or prevention may not be available or is complex or where family implications of the test result need to be considered before testing is commenced; diagnostic testing in pregnancy following an increased risk result on prenatal screening or abnormality diagnosed prenatally or where there is a family history of a genetic condition.

### **Laboratory Reports of Genetic Testing**

Laboratory reports containing results of genetic tests may need to be more complex and detailed than standard biochemistry reports.

The shared nature of genetic information may require that a family pedigree be described, so that it is clear to which other members of the family the patient is related, and the nature of that relationship. Sometimes, genetic results can only be interpreted by the combination of results from more than one family member.

Testing should also describe in significant detail the nature of the testing performed. For example, it may not be sufficient to merely state the gene that has been examined, as different Polymerase Chain Reaction (PCR) primers may result in examination of different parts of the same gene, and such differences may be clinically significant. Similarly, it may be

important to know whether the testing process has used DNA sequencing or not. Finally, it may be important to know *which* mutations have been specifically searched for and excluded, as some mutations may not have been considered in the analysis. It is not uncommon for a single genetics result to occupy several paragraphs of description in the report, and this type of format may not lend itself to the 'spreadsheet-like' layout of many biochemical reports in common use today.

### **Addressee of the laboratory report**

Normal practice of most pathology laboratories is to provide a copy of the test result to the referring doctor. Where one laboratory has referred the specimen to another laboratory, the referral laboratory will send the report to the referring laboratory which will, in turn, send the report back along the referral chain to any earlier referring laboratory or to the originating requesting doctor.

Genetics laboratories have traditionally operated in a different reporting paradigm. Instead of the referral laboratory informing the referring laboratory of the result, they often merely inform the referring laboratory that testing has "been completed", and send the complete report directly to the primary referring doctor. This practice has arisen as a means of protecting the privacy of genetics information, to ensure that as few people as possible are in possession of genetic information.

The recent publication of ISO 15189:2003<sup>1</sup> and of the NPAAC Standards and Guidelines for Nucleic Acid Detection and Analysis<sup>17</sup> has made it clear which of these two reporting paradigms must be observed. Both have mandated that the referral laboratory *must* issue the report to the referring laboratory, although the practice of the genetics laboratory informing the primary referring doctor is permitted, but only in addition to, and not instead of, informing the referring laboratory.

### **Trusted Third Party Reports**

Genetic testing may be performed on 'well' individuals, who may wish to know their genotype for reasons ranging from family history of autosomal recessive disorders, membership of an ethnic or geographic group at increased risk or prevalence for specific genetic mutations, through to mere curiosity as to their genotype. As a result of this wide range of motivating factors, the sample is obtained from a well individual who is perhaps more correctly referred to as a client, rather than a patient.

Some of these well and symptom-free clients may not wish to have the results of their genetics testing forwarded to their medical practitioner. While it is not permitted in Australia for a laboratory to market genetic tests directly to the public in the

absence of an integrated clinical service,<sup>17</sup> it is permissible to offer testing directly to the public as part of a comprehensive community genetics testing program (for example, ref. 11). Laboratories wishing to offer testing as part of a community genetics program may need to develop novel result reporting strategies (reviewed in reference 5, and examples described in reference 11), in which results are reported directly to the client, and without copies of the report being provided to intermediary medical practitioners or laboratories. The NPAAC Standards<sup>17</sup> recognise this form of ‘trusted third party’ reporting,<sup>5</sup> although ISO 15189:2003<sup>1</sup> accommodates this only indirectly, through recognition of the need to observe “local” regulations in the reporting of laboratory results to other than the referring party.

### Specific Applications of Genetic Tests

The ethical and psychosocial implications of the genetic test will have particular application according to the context in which it is carried out. The distinction between the two levels in the NPAAC Standards and Guidelines<sup>17</sup> (and Table) would usually be made by the doctor ordering the test, since that individual will be best placed to appreciate the short-term and long-term implications of the test for the patient and other family members. Within these levels are tests that are used for a range of purposes that include:

#### 1. Late-onset Neurological Conditions (untreatable)

##### *Diagnostic testing*

In Huntington disease, where there are clinical symptoms, the diagnosis is confirmed according to the number of triplet repeats in the IT15 gene increased over the usual number.

Relevant issues are consent and discussion of result disclosure to other family members.

##### *Presymptomatic Testing*

The genetic diagnosis of Huntington disease in a family member can then enable unaffected family members to be tested to determine if they have the mutation. If the mutation is present (a positive result) the person will develop the condition if they live long enough. Testing can provide certainty regarding their risk and enable life planning. However both positive and negative results can impact on family dynamics.

Again, informed consent is essential, as are the issues of ‘whom to tell’ and ‘when to tell’. An asymptomatic person with a positive test result must disclose this result in an application for a life insurance product, which may impact on their insurability. On the other hand, a negative test result will negate the impact that the family history had on their life insurance accessibility. Consideration of timing of the test needs to be given as the result of the presymptomatic test would

have no health benefit for a child although testing in pregnancy is available. Testing of embryos prior to implantation using in vitro technologies may also be available.

#### 2. Familial Cancers (breast, ovarian, colorectal and prostate cancers and melanoma)

##### *Diagnostic Testing (mutation searching)*

Where there is a strong family history of some specific cancers, genetic testing may confirm that the onset of cancer involves inherited susceptibility. For these conditions, the test involves searching for the family specific mutation in the genes that have been identified as associated with the condition. A negative result on a mutation search only means that a genetic basis has not been confirmed. A positive result however means that asymptomatic blood relatives can have the genetic test that would look for the specific mutation.

Specific issues include consent and family communication and dynamics.

##### *Predictive Testing*

A positive test for the family specific mutation means that the asymptomatic person is at increased risk for the cancer or associated cancers but they may never develop the condition. A negative result means that the person is now at population risk.

Specific issues include consent, disclosure to third parties, screening and early detection strategies. For some colorectal cancers for example, testing may need to be done in childhood.

#### 3. Conditions with reproductive implications e.g. cystic fibrosis

##### *Diagnostic Testing*

Cystic fibrosis is one of the conditions covered by newborn screening carried out a few days after birth. There are over 1,000 mutations in the CFTR gene identified to date<sup>13</sup> but of these, only about 29 mutations are most commonly found in the Australian population. Identification of the genetic basis of a child with clinical symptoms may take some time if the mutations are not within this ‘panel’. However, once the mutations have been identified, testing can be offered to the parents in future pregnancies or preimplantation of a tested embryo.

Issues include careful consideration of the risks to the pregnancy associated with the prenatal test itself and the decision making engendered by a positive prenatal test result.

##### *Genetic Carrier Testing*

###### (a) Cascade Testing

Where the mutation causing cystic fibrosis in the family has been identified, testing of blood relatives can be offered to determine if they are carriers of the mutation.

Informed consent requires addressing the meaning of the result for their personal health and their reproductive options when considered with their partner's result. Issues of family communication also need to be addressed.

#### (b) Population Testing

As the genetic carrier rate for cystic fibrosis in Australia is one in 25 amongst Australian of Northern European ancestry, and the autosomal recessive pattern of inheritance means that a family history for the condition is not common, population genetic carrier screening has been proposed.<sup>14</sup> Testing in this context would only involve the more common mutations.

Issues include offering testing for a condition that is not present at the same rate across all the communities in multicultural Australia. The challenges for informed consent include addressing the variable sensitivity, understanding the meaning of the positive result, family disclosure and the implications for reproductive planning.

#### **AACB, IFCC and an Ethics Framework for Laboratory Medicine**

The AACB<sup>15</sup> is a member of the International Federation of Clinical Chemistry and Laboratory Medicine (IFCC).<sup>16</sup> Through the IFCC, the AACB has played a major role in the development of an over-arching Ethics framework for Laboratory Medicine. This framework has recently been completed and has been submitted for publication.

Recognising that the IFCC is formed by representatives from Clinical Chemistry and Laboratory Medicine in more than 70 countries plus more than 30 corporate members, it is not intended that the IFCC Ethics Framework will necessarily have the complete agreement of all IFCC member States, Societies and organisations. The objective is to produce a statement with widespread support from the members of the Federation.

Many IFCC member Societies and States may already have appropriate policies in place addressing various ethics issues. These may have been developed by regulatory bodies or by various national professional bodies and associations. These existing national policies may be able to support the IFCC policies as bibliographic references to inform and improve international frameworks. In turn, these IFCC policies may assist members that do not yet have such formal national policies, or that are planning to review existing national policies.

#### **Conclusion**

In this brief overview, we have sought to discuss the ethical implication of many of the recent advances in diagnostic genetics testing and to highlight some of the still unresolved ethical issues.

Clinical biochemists in Australasia, through the AACB and its membership of the IFCC, have already played a major role in defining and beginning to address these issues. It is important that we continue these activities. Genetics is not always viewed benignly by the lay public at large. Popular Science Fiction movies (such as "GATTACA"), combined with sensationalist media coverage, sometimes presents genetics testing in a malevolent light. Without positive and proactive efforts to counter these incorrect impressions, the mood of society may well shift and evolve in the future into one in which governments choose to regulate genetics testing in ways that the knowledgeable scientific community can see may not be in the public's long-term interests.

The laboratory community has the knowledge and the ethos to be able to undertake genetics testing of a high standard and in a responsible way. The challenge will be ours to take this message out of the comfort of our back-room laboratories and into the daylight of wider scrutiny and public policy.

We live in exciting times.

Competing interests: None declared

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