

This fact sheet describes some of the ethical issues that can arise because of the use of genetic testing.

### In summary

- The nature of the information gained from a genetic test raises additional ethical issues compared to other health information
- Genetic testing is best offered by specialised services such as a Clinical Genetics Services
- Laboratories undertaking the testing should be accredited.

### WHY IS GENETIC INFORMATION SPECIAL?

Genetic information is often considered exceptional when compared to other medical information about an individual for a number of reasons.

#### The shared nature and ownership of genetic information

Genetic conditions are family health problems. A diagnosis or a finding of inherited predisposition in a family member has implications for other family members. Health professionals have an ethical responsibility to prevent harm or avoid seriously jeopardising the health of others (the duty of care). Similarly, individuals undergoing genetic testing have a responsibility to consider not only what it means for their own health, but also what the information may mean for their relatives, and their responsibilities towards those relatives.

Geographic distance or discord in families can sometimes lead to difficulties in revealing genetic test results that may be important for other family members. Responsibility and obligation however needs to be balanced with the right of an individual to choose to know their personal genetic information or, equally, not to know.

The emphasis needs to be on the right of the person to choose. Genetic counselling is essential both before and after genetic testing so that all the implications of undertaking testing including having information which might be of interest to others can be understood.

### Limitations of genetic testing

While in some cases, genetic tests provide reliable and accurate information on which people can make decisions, in other cases it may not be possible to obtain a definitive result.

An individual is much more than the sum of their genes: the individual's environment can modify the expression of genetic messages to the body and many health factors are not genetic.

The discovery of a variation in a particular gene may provide some information about the nature of the condition that the person has, will develop or for which they may be at increased risk, but can rarely predict the severity of the condition or the age at which symptoms will first onset.

This lack of precision in relating the expression of the condition (called the **phenotype**) to an individual's genetic make-up (called the **genotype**) can make the decision-making process in regard to acting on the information very challenging.

This is particularly so when the genetic testing is done for prenatal testing of a condition. Genetic counselling is essential to assist families in that decision making process and ensure that the decision is as informed as possible.

#### Predictive/Pre-symptomatic testing – generally for adult-onset conditions

This type of genetic testing applies to families in which an underlying genetic cause for their condition has been identified and can be used to identify currently healthy family members that are at-risk, if they wish to do so.

Pre-test counselling is important in these cases and aims to provide accurate information so that the individual can make an informed decision about whether or not to have testing.

This is called **informed consent** and means that the person undergoing the test should only do so on a voluntary basis and with a full understanding of all the implications. There can be a danger of coercion, for example, an enthusiastic researcher or a member of a family may try to persuade others in the family to undergo testing about which they feel uncomfortable

Discussion of the potential **emotional impact on family members** of finding out test results should also be undertaken before testing. This can be substantial whether the results are bad or good, for example the feelings of guilt often felt by 'survivors' who have not inherited the gene fault. Discussion of implications for other family members and obligations to inform, as well as the potential interest of third parties in genetic information revealed by testing such as insurance and employment, are also important.

#### **The potential for discrimination**

Genetic testing may impact an individual's ability to obtain life insurance and employment in certain professions. This is especially the case with predictive/presymptomatic testing which provide information about an individual's future health.

#### **Reproductive choices/Prenatal testing**

Whether or not to have children is a major decision for any individual. It is even more difficult where one or both of the prospective parents knows or suspects that they may carry a faulty gene associated with a health problem which could affect their children.

The decision to have a baby may lead to a number of further decisions to be made in regards to the possible genetic testing of the embryo/fetus during the pregnancy. Limitations of such testing are the same as those discussed previously, in particular detection of a faulty gene or a chromosomal abnormality may not provide all the information about the potential or quality of life for the child or the severity of a particular condition

When a fetal abnormality is detected, support is essential for whatever difficult decision is made.

Some expectant parents will decide to continue the pregnancy and try to put in place the professional, medical and social support that will be required. Others may choose to terminate the pregnancy. This decision may conflict with moral, religious and cultural beliefs.

Different individuals, communities, cultures and religions have different perceptions of disability and this may raise additional issues.

#### **Inappropriate applications of genetic testing**

Genetic testing has many potential applications, however some of these are in conflict with what we could consider ethical. These include use of genetic testing to confirm paternity sex selection of a fetus for family balancing reasons without the informed consent of all parties involved.

#### **Setting boundaries in applications of the genetics technology**

Philosophers on science have put the view that science is morally neutral. It is the uses to which the science is put that might be good or bad. With the new advances in genetics, as with any powerful new scientific tool, there is a potential for abuse. Controversial applications of genetic testing such as reproductive cloning and genetic testing for enhancement create a huge challenge worldwide and require implementation of international regulations on the boundaries within which these applications can be applied. Moral, religious and cultural beliefs underpin decision-making by individuals, couples, families and communities and may challenge such boundaries.

#### **Forensic DNA databanks**

The use of fingerprints (more accurately known as dermatoglyphic fingerprints) for forensic identification purposes has been in place since the 1890s. One hundred years later, **DNA fingerprinting** is being used to complement the traditional system, or is being used in isolation for identification. The public has also contributed to investigations of unsolved crimes by volunteering genetic samples. Overall there is a need to ensure that samples are used for the purpose for which they were collected and protected from misuse.

## Patenting of genes

The issue of patenting genes as recognition of the intellectual achievement required to isolate a single gene from the 20,000 or so genes in the cell is contentious.

In Australia there have been three inquiries into the issue of gene patenting and human health, including the Australian Law Reform Commission ([www.alrc.gov.au](http://www.alrc.gov.au)) in 2004, the Senate Community Affairs Reference Committee in 2010 ([www.aph.gov.au](http://www.aph.gov.au)) and the Advisory Council on Intellectual Property in 2011 ([www.acip.gov.au](http://www.acip.gov.au)).

The Federal Government issued a response in 2011 confirming that the government does not support the absolute prohibition of gene patenting, however will aim to ensure that gene patents do not lead to patients being denied 'reasonable access to healthcare'.

## Case study

*Huntington disease (HD) is a neurological degenerative disease that has an onset in most people between the ages of 30 and 50. There is no cure for this condition and it is progressive. Symptoms include deterioration in movement, cognition and generalised functioning. Death usually results from respiratory illness.*

*HD is an inherited condition. A child of an affected person has a 50% chance of inheriting the faulty gene that causes the condition. Genetic predictive testing is now available for persons over the age of 18 who have an affected parent or relative which will tell them in almost all cases whether they will develop the disease at some stage in their life.*

*Worldwide, of those eligible for the test, only around 15% of people have taken up the option of testing.*

*Mr H. is a 25 year old man whose grandfather died some 10 years ago from Huntington disease. Mr H's mother has therefore a 50% chance of developing HD. She decided to have the genetic test and has been shown to have the faulty gene. She will definitely develop HD at some time and Mr H is now at 50% risk of developing HD.*

*Mr H. is an air traffic controller. He loves his job and he feels he could perform his duties most adequately for many years, irrespective of whether he carries the faulty gene for HD or not. He does not wish to have the genetic test. His employer is unaware of his family history.*

## Dilemmas

- To know or not to know? When is the right time to decide to have predictive/pre-symptomatic testing?
- Do employers in industries involving public safety have the right to demand family health history information? In cases where genetic predictive testing is available for conditions that may impact on public safety, do employers have a right to predictive testing information about an individual whose current health status is excellent?
- Who actually 'owns' this information and who should decide who can access it?
- What if the situation was reversed and Mr H wanted testing but his mother had refused? What responsibility is there to offer testing to an individual when the result may indirectly reveal the genetic status of a relative (if Mr H carries the HD gene fault then he must have inherited from his mother)?
- Implications for Mr H's reproductive choices.