Fact sheet **19**

ETHICAL ISSUES
IN HUMAN GENETICS AND GENOMICS

This fact sheet talks about some of the ethical issues to think about when we have a DNA test.

IN SUMMARY

- The type of results from a genetic test may raise ethical issues in a different way to other types of health information
- Laboratories undertaking the testing should be accredited (approved or certified).

WHY IS GENETIC INFORMATION DIFFERENT?

For a number of reasons, genetic information is often considered different when compared to other medical information about an individual.

The shared nature and ownership of genetic information

Genetic conditions are family health problems. A diagnosis or an increased chance of developing a health condition because of an inherited DNA change has implications for a person and their family. Health professionals have an ethical responsibility to prevent harm or avoid risks to the health of others (the duty of care). Similarly, people having a genetic test have a responsibility to consider not only what it means for their own health, but also what the information may mean for their relatives.

Geographic distance or discord in families can sometimes mean that sharing genetic test results important for other family members is more challenging. Responsibility, however, needs to be balanced with the right of a person to choose to know their personal genetic information or, equally, not to know.

Therefore, a person has the right to choose. Genetic counselling is important both before and after genetic testing so that all the implications of having a test can be understood.

Limitations of genetic testing

While sometimes, genetic tests provide reliable and accurate information for people to make decisions, at other times it may not be possible to get a result that is certain.

We are each much more than the sum of our genes: our environment can influence our health and development and many health factors are not genetic.

The discovery of a variation in a particular gene may provide some information about the nature of a health condition that a person has, will develop, or for which they may be at increased risk. Rarely can it predict how severe the condition will be, or the age at which symptoms will first start.

How our health or development shows up (called the phenotype) and whether this links with our genetic make-up (called the genotype) can help inform how we act on the genetic information.

This is particularly so for testing in pregnancy for a genetic condition. Genetic counselling is important to help families with decision-making so that a decision about managing health 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 found and can be used to identify currently healthy family members that are at-risk, if they wish to know this.

Further information may be found in the Position Statement, Predictive and Pre-symptomatic Genetic Testing in Adults and Children (Human Genetics Society of Australasia).

Pre-test counselling is important and aims to give accurate information so that a person can make an informed decision about whether or not to have testing.

This is called **informed consent** and means that the person having the test is doing so on a voluntary basis and understands the implications. There may be a possibility of coercion, for example, an enthusiastic researcher or a member of a family may try to persuade others in the family to have testing even when they feel uncomfortable about this.

Discussion of the potential **emotional impact on family members** of finding out test results should also be talked about before testing. This may be significant whether the results are favourable or not. For example, there may be feelings of guilt felt by 'survivors' who have not inherited a particular gene variation. Talking about the relevance of genetic information to other family members and importance of letting them know, as well as the potential interest of third parties, such as insurance agencies and employers, are also important.

The potential for discrimination

Genetic testing may influence ability to get life insurance and employment in certain professions. This is especially when having predictive/presymptomatic testing, which provide information about someone's future health.

Reproductive choices/Prenatal testing

Planning a pregnancy may be more difficult where one or both of the prospective parents knows or suspects that they may have a gene variant linked with a health problem, which could affect their children.

Decisions may be made regarding genetic testing of the embryo/fetus during the pregnancy. Limitations of such testing are the same as those discussed before. In particular, finding a gene variation or a chromosome change may not provide all the information about the potential or quality of life for the child, or severity of a particular condition.

When a problem with a developing baby is found, support is essential for whatever decision is made.

Some expectant parents decide to continue a pregnancy and may put in place the professional, medical and social support they need. Others may choose to terminate the pregnancy. This decision may conflict with personal moral, religious and cultural beliefs.

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

Inappropriate ways of using genetic testing

Genetic testing can be used in many ways. Some of these are, however, in conflict with what might be considered ethical. These include the use of genetic testing to confirm paternity without the informed consent of all individuals involved, or sex selection of a fetus for family balancing reasons.

Setting boundaries on how genetics technology is used

Philosophers of 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 new advances in genetics, as with any powerful new scientific tool, there is a potential for abuse. Controversial ways that genetic testing may be used such as reproductive cloning and genetic testing for enhancement or improvement rather than preventing health conditions create a challenge worldwide. International regulations may help manage some of these issues. Moral, religious and cultural beliefs may motivate 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 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 make sure that samples are used for the reason they were collected, and there is protection 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 in 2004, the Senate Community Affairs Reference Committee in 2010 and the Advisory Council on Intellectual Property in 2011.

The Australian Government issued a response in 2011 confirming that the government does not support the absolute prohibition of gene patenting, however, it aims to make sure 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 symptoms get worse over time. Symptoms include deterioration in movement and cognition. Death usually results from respiratory illness.

HD is an inherited condition. A child of an affected person has a 50% chance of inheriting the non-working gene that causes the condition. Genetic predictive testing is now available for persons able to provide consent, who have an affected parent or relative. Testing will tell them in almost all cases whether they will develop the disease at some stage in their life.

Worldwide, of those able to test, only around 15% of people choose to take this up.

Mr H. is a 25 year old man whose grandfather died some 10 years ago from Huntington disease. Mr H's mother therefore has a 50% chance of developing HD.

She decided to have the genetic test and has been shown to have the non-working gene copy. She will definitely develop HD at some time and Mr H. is now at 50% chance 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 non-working gene copy 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 or presymptomatic testing is available for conditions that may impact on public safety, do employers have a right to genetic 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 change, then he must have inherited from his mother)?
- Impact on Mr H.'s reproductive choices.