ACI NSW Agency for Clinical Innovation

INFORMATION BOOKLET

Genetic/genomic testing

Clinical Genetics Network

This information booklet is to accompany the consent forms for **Genetic/genomic testing**.

This information booklet provides further information so an informed consent can be provided if you are consenting for; yourself OR a parent or guardian consenting for a child OR the Person Responsible consenting for a patient under the Guardianship Act.

Collaboration. Innovation. Better Healthcare.

Our obligations

We are committed to providing you with information that clearly explains genetic testing so you are able to provide an informed consent.

Section A – Testing and results

What is genetic testing?

DNA is the chemical that makes up your chromosomes and genes in the cells of your body.

Genes provide genetic information that the body uses to develop, grow and work.

Genetic testing looks at the DNA in your genes, usually examining just one gene at a time rather than all of your 20,000 genes.

However, sometimes a number of genes known to be associated with the condition are tested at the same time. This is called panel testing.

What are doctors looking for in DNA?

Doctors look for changes in the genetic information that are known to make some genes faulty.

What is a gene fault?

A gene fault is a change in a person's DNA code that may cause or increase the risk for developing a condition.

A gene fault is also called a **pathogenic variant** or a **mutation**.

What does it mean to be born with a gene fault?

We are all born with a number of differences in our 20,000 genes. That is why we are all unique.

These gene changes may be inherited or occur for the first time in you. Usually these gene changes do not cause a problem.

However sometimes a gene change in one or more genes can cause the gene/s to be faulty which can:

- Directly cause a condition to develop in a baby, child, adolescent or an adult;
- Make a person at increased risk of developing the condition, but noting that they might never develop it at all.

What is genomic testing?

Genomic testing, like genetic testing, looks for gene faults to find a cause of a genetic condition.

However, instead of testing one or several genes at a time, it is now possible to test most or even all of the 20,000 genes at the same time.

Genomic testing may be able to detect the cause of a genetic condition within a shorter period of time.

Why is genomic testing done?

If no gene faults are found by testing one gene or a panel of genes, or if you and the doctor choose, testing of all the 20,000 genes can be done to see if the cause of the condition can be found.

What is required for genetic testing?

Your doctor will explain the test and you will have an opportunity to ask questions and receive answers. The doctor will then ask for your written consent before testing.

Doctors can take DNA from the blood or other tissues to examine in the laboratory. If a blood sample is needed, 5–20 mL (1–2 tubes) is taken for testing.

In some cases, testing blood samples from other genetic relatives may be needed to help understand the results.

You have the right at any time to withdraw your consent, without affecting the quality of your treatment.

What will the test results mean?

Sometimes testing does not give you an answer, or it may reveal information you may not want to know.

Testing may also find DNA changes which are not yet fully understood and therefore doctors may not yet understand what they mean for your health.

What are the possible outcomes of a genetic test?

- **A positive result:** A gene fault is found that is the cause of the genetic condition. This is called a pathogenic variant or mutation. This may be important for you or your genetic relatives and future children.
- **A result of uncertain significance:** One or more gene changes have been found, but it is not clear what they mean. Testing of other genetic relatives may help to find out if the result is the likely cause of the genetic condition. Further testing/analysis to understand the result may be done in the future.
- **An uninformative result:** No gene fault is found that could explain the genetic condition. This may be because:
    - A gene fault is present but could not be found using current technology;
    - A fault is present in another gene that was not tested;
    - Not all gene faults that cause the genetic condition have yet been identified. Testing in the future may happen as knowledge increases. The timeframe for this is unknown.
- **Benign result:** One or more changes in the DNA are found which are known to be harmless and not cause genetic conditions.
- **An incidental finding** - also called additional or secondary findings. Some of the findings will be considered to be clinically significant by your doctor. These relate to gene faults causing or leading to increased risk for conditions that can be treated or prevented or where knowledge can result in other health benefits.

What are the clinically significant incidental findings that might be found?

There are two types of these results that occur unexpectedly and rarely:

- You have a gene fault that causes or can lead to an increased risk of a serious health problem in you or your genetic relatives, which is found by chance when looking for the cause of the genetic condition being tested for. Some of the gene faults will be in regard to conditions that may be able to be treated or prevented. Others will be in regard to conditions that will not be able to be treated or prevented or where knowledge can result in other health benefits.
- You are carrying a gene fault that will usually not cause any health problems in you. This is called a genetic carrier result. All our genes come in pairs and genetic carriers have one faulty copy and one working copy of the gene. Some genetic conditions are only expressed when the faulty gene has been inherited from both the mother and father so that the child inherits both faulty copies and has no working gene copy. Women who are carriers of a faulty gene located on the X chromosome (one of the sex chromosomes) will usually be unaffected as they will have a working copy on their other X chromosome. Their sons could be affected with the condition if they inherit the faulty gene as males usually have only one X chromosome and a Y chromosome.

Which incidental findings can I choose to be informed about?

At the time of consent, you may choose whether or not you wish to be informed about an incidental finding. You will be asked to decide if you wish to know about a gene fault found unexpectedly that causes or leads to an increased risk for:

- Any genetic condition; OR
- Only those conditions that can be treated or prevented or where knowledge can result in other health benefits; OR
- None at all, regardless of whether the genetic condition is potentially treatable/preventable;
- In addition you will be asked if you wish to know about genetic carrier results.

Making these decisions can be difficult, and over time you may have differing thoughts or your circumstances may change. Your doctor may ask you later, when the results are available, if you have the same or differing views on any unexpected findings unrelated to the condition for which testing was being done.

While you can withdraw or change the consent at any time, results of any gene faults found before you changed the consent will be reported to your doctor and will be in your medical record.

Which incidental findings can I choose to be informed about when I am consenting as a parent /guardian of a patient less than 16 years?

A clinically significant incidental finding that is currently treatable or preventable in childhood will be reported to you.

However, at the time of consent, you may choose whether or not you wish to be informed about an incidental finding that does not onset until adulthood.

You will be asked to decide if you wish to know about a gene fault found unexpectedly that causes or leads to an increased risk for:

- Any genetic condition that does not onset until adulthood; OR
- Only those conditions that do not onset until adulthood that can be treated or prevented or where knowledge can result in other health benefits;
- In addition you will be asked if you wish to know about genetic carrier results.

Are there any risks in having genetic or genomic testing?

The results may affect the ability to obtain some types of insurance such as life or income insurance.

Our DNA is inherited from our parents and passed onto our children. This means that:

- The test result can change the chance that other genetic relatives may/will develop the condition;
- The results may show that your mother and/or your father are not your biological parents (non-paternity or non-maternity).

What if I am consenting on behalf of a minor?

If you are a parent or guardian consenting on behalf of a minor to genetic testing you should ensure that you fully understand the benefits and risks to your child prior to consenting to the testing. Genetic Alliance Australia recommends that parents consider having their children tested for conditions or genetic carrier status only when the result is likely to directly benefit the child's health during childhood.

What if I am providing substitute consent for a person 16 years or over who does not have the capacity to consent for themselves?

If you are a person responsible under the Guardianship Act you have a right and a responsibility to know about the proposed genetic tests and to ensure that you fully understand the benefits and risks to the patient prior to consenting to the testing.

Section B – Storage

What will happen to your blood sample and DNA?

When the testing is completed, your DNA will remain the property of the laboratory where it is stored for a period of time determined by laboratory practice or regulation after which it will be destroyed. There is no guarantee it will be suitable for future use or testing.

Section C – Confidentiality

What will your DNA or test result be used for after testing?

Your DNA will only be used for the purpose for which you have given consent except where information is lawfully required or authorised under law.

- Your **identified DNA or blood/tissue sample** will not be used for any other purpose without your written consent except in situations where disclosure is lawfully required or authorised under law;
  e.g. In accordance with the Health Records and Information Privacy Act 2002 (NSW), genetic information can be used and disclosed without consent in order to lessen or prevent a serious risk to the life, health or safety of a genetic relative no further removed than 3rd degree; and, only where the disclosure is made in accordance with the guidelines issued by the Information and Privacy Commission NSW www.health.nsw.gov.au/policies/ib/2014/IB2014_065.html
- Your **identified test result and the fact that you have had a genetic test** will not be revealed to any other person or organisation not involved in your care without your written consent except in situations where disclosure is lawfully required or authorised by law.
- Your **non-identifiable DNA sample/ clinical information/test result** may be lawfully used without your consent for:
  - Research purposes (see the Privacy Leaflet for Patients of NSW Health Genetics Services).
  - Quality activities and education where use is allowed under national pathology accreditation requirements.

Section D – Research

Why is it important for your DNA and testing results to be used for research?

DNA testing gives information for you and your genetic relatives and also helps doctors, scientists and genetic counsellors learn more about gene changes and the causes of health problems.

The meaning of gene changes identified in the DNA is still developing. Being able to study DNA from many different people affected with different genetic conditions or with a variety of health problems will help that understanding.

How will the test results be stored for research purposes?

It is important that researchers from all over the world share their findings through databanks to increase understanding. Qualified researchers from the government, academic, commercial or other organisations can then use the information to look at links between the test results and genetic conditions.

What about your privacy and confidentiality?

It is unlikely that there will be risks to your privacy and confidentiality by sharing your results with these databanks. There are many safeguards in place to protect your privacy.

- Results are non-identifiable and sent to researchers with a code number attached.
- Only information that is non-identifiable will be given to databanks.

What are some of the concerns about research?

While every effort is made to protect the privacy of your information, there is a very small chance that your test results could be shared with others by genuine mistake. These include the public, employers, the law or other family members which could impact on family relationships.

DNA test results could identify you if someone already has an **identified DNA** sample from you. They could compare the information from that DNA sample to the non-identifiable information held in the databank.

Information that a research study provides to you may affect your chance to get or renew some forms of insurance (such as life or income insurance).

Will I be contacted with any findings from research?

Depending on the research protocol you may not receive any findings from research studies unless they are clinically significant findings. There is usually no direct benefit from taking part in research, however studying your DNA test results will add to science and medical knowledge.

Can I change my mind about using my DNA in research?

You can withdraw your consent at any time if you do not wish your results to be stored in databanks. There will be no consequences from withdrawing your consent. However, results that have already been sent to researchers cannot be taken back from those researchers.

Section E – Consent

What if I have any other questions?

If you have any concerns or questions, please contact your doctor or genetic counsellor.

Contact us

If you have questions regarding this information please speak to your NSW Health genetic health professional.

If you have questions or a complaint about the privacy of your personal information, please contact the Privacy Contact Officer in your local health district, health organisation or specialty health network.

www.health.nsw.gov.au/patients/privacy/Pages/privacy-contact.aspx

Further information

If you would like further information please visit:

Centre for Genetics Education
www.genetics.edu.au

Genetic Alliance Australia
www.geneticalliance.org.au

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