

INFORMATION BOOKLET

DNA testing for serious adult onset neurogenetic conditions

Clinical Genetics Network



This information booklet is to accompany the consent forms for **DNA testing for serious adult onset neurogenetic conditions**.

This information booklet provides further information so an informed consent can be provided if you are consenting for; yourself 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 or a panel of a number of genes rather than all of your 20,000 genes.

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 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:** You have inherited the gene fault. This is called a pathogenic variant or a mutation. You are at high risk of developing/will develop the condition and your children have a chance of inheriting this gene fault. Your siblings may also have been born with the gene fault. It is not possible to accurately predict the age of onset of the condition.

- **A negative result:** You have not inherited the gene fault. You are not at high risk of developing/will not develop the condition. You cannot pass the gene fault on to your children.
- **An intermediate result:** A gene fault is found but it is not clear whether you may or may not develop the condition. In some instances this may have implications for your siblings and children and their descendants.

Are there any risks in having genetic 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).

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 www1.health.nsw.gov.au/policies/lib/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).

Section E – Consent

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.

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



AGENCY FOR CLINICAL INNOVATION

Level 4, Sage Building

67 Albert Avenue, Chatswood NSW 2067

PO Box 699 Chatswood NSW 2057

T +61 2 9464 4666 | F +61 2 9464 4728

E aci-info@health.nsw.gov.au | www.aci.health.nsw.gov.au

Produced by: Clinical Genetics Network

Further copies of this publication can be obtained from the Agency for Clinical Innovation website at www.aci.health.nsw.gov.au

© Agency for Clinical Innovation 2017