PATIENT FACT SHEET
GENETIC AND GENOMIC TESTING

Australian Genomics

This fact sheet aims to help you understand the consent form and the possible **results** of genetic and genomic testing. You can show this Patient Fact Sheet to health professionals, family, and friends to help you decide whether to have testing. It will also help you think of any other questions you may wish to ask. You need to give your consent before the testing starts. You are free to withdraw your consent at any time before testing. Testing is **your choice**.

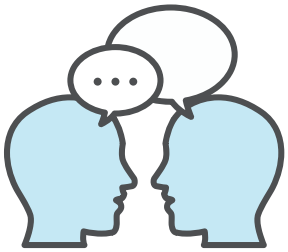
QUESTIONS TO THINK ABOUT AND ASK YOUR HEALTH PROFESSIONAL:

1. Where will my test be done?
2. How long will it take to get the results back?
3. How will I receive the results?
4. What is the chance of the test finding an answer?
5. What happens if the test does not find an answer?
6. Will it cost me anything to have testing?
7. What might it mean for my family if I or my child gets the test?
8. What support is available for me during testing and after receiving the results?
9. Will I need to go to a clinical genetic service?
10. Will the test change the way I or my child receive health care?
11. How will the test be performed?
12. What if I change my mind and wish to withdraw my consent?
13. Will my General Practitioner (GP) be told my test results?

Choosing Wisely Australia suggests asking your health professional these questions before having any health-related testing:

Scan me!

1. Do I really need this test?
2. What are the risks?
3. Are there simpler, safer options?
4. What happens if I don't do anything?
5. What are the costs?



Your **health professional** may be a clinical geneticist, a genetic counsellor or another type of specialist like a cardiologist.

CLINICAL GENETICISTS & GENETIC COUNSELLORS

The clinical staff at genetic services are **clinical geneticists** and **genetic counsellors**. They play different roles in the management of people with genetic conditions or suspected genetic conditions. Clinical geneticists are medical doctors specialising in the diagnosis and management of patients with or at risk of a genetic condition. Genetic counsellors are allied health practitioners with specialist knowledge in human genetics and counselling. Both of these health professionals provide emotional and practical support to help people adjust to living with, or being at risk for, a genetic condition. They work together to support patients, families, and clinical teams in the management of genetic conditions.

GENES AND GENETICS

Genes are instructions that tell our bodies how to grow and develop. Genes are made up of DNA. All of a person's genetic information is called their **genome**.

Each person has many differences (or **variants**) in their genes. Most **variants** are harmless and do not impact how the gene works. However, some people have variants that do affect how a gene works. These variants can cause or increase the chance of developing a **genetic condition**. Variants can be passed down from parents or can appear, at random, for the first time in a person.

If you have unexplained health issues your health professional may suggest doing a **genetic test**. A genetic test looks for variants that change how a gene works. This variant(s) may be the reason for your health issues.

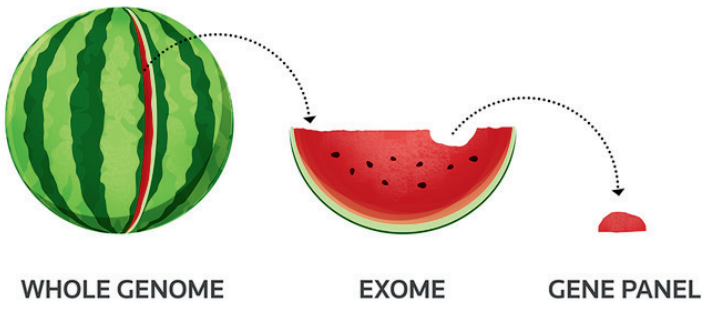
WHAT IS THE DIFFERENCE BETWEEN A GENETIC & A GENOMIC TEST?

A **genetic test** looks at a single gene or a small number of genes at a time. The test may look for a single variant or multiple variants.

A **genomic test** looks at many genes, sometimes all 20,000-25,000 genes at once.

The different types of genomic tests are:

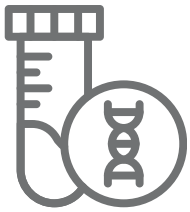
- **Genome sequencing** looks at most or all a person's genome to find variants.
- **Exome sequencing** looks at about 1-2% of a person's genome to find variants.
- A **gene panel** focusses on just the genes that could be related to a person's health condition.



Types of genomic tests

Your health professional will talk to you about what type of test they will order for you.

HOW IS THE TEST DONE?



Both genetic and genomic tests require a sample of DNA. Blood and mouth swabs are the most common samples used. You do not need to fast for the blood test.

WHAT IS THE PURPOSE OF THE TEST?

The purpose of having genetic or genomic testing may include:

- **Diagnostic testing:** trying to find the cause of a genetic condition,
- **Predictive testing:** finding out if you have a variant that has caused a genetic condition in your family. This may impact your health in the future,
- **Carrier testing:** seeing if you carry a variant that could be passed to your children, but which is unlikely to affect your health,
- **Prenatal testing:** diagnosing a genetic condition in your pregnancy,
- **Confirmation testing:** confirming a clinical diagnosis you have previously received, or
- **Segregation testing:** understanding how a variant that has been found, has been passed down through your family.

WHAT ARE THE POTENTIAL BENEFITS?

- To find out a cause of your or your child's condition.
- To support or guide medical care for you or your child.
- To improve understanding of a condition, access support, and plan for the future.
- To help you or your blood relatives know the chance of developing a condition.
- To provide information about the chance of having a child with the same condition as you.

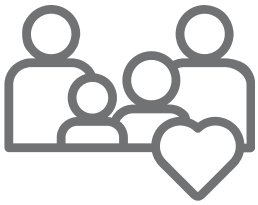
WHAT ARE THE POSSIBLE RESULTS?

The result of testing may be:

- A variant is found that is likely to cause your or your child's condition.
- No variants are found as the cause of your or your child's condition.
- A **variant of uncertain significance** has been found. This means that at this point in time it is not known whether it is the cause of your or your child's condition.

WHAT ARE OTHER ISSUES TO THINK ABOUT BEFORE TESTING?

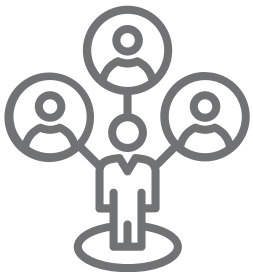
1. Testing can have impacts on your **blood relatives**. A **blood relative** can be anyone who is related to you by genetics. This could be your ancestors, your biological family, or someone who is not born yet. The results of your test may reveal information about your blood relatives' health because you share genetic information with them. This can cause some people to worry.



2. A genomic test may unexpectedly show something that is not related to the reason for testing such as an unrelated health risk. This is called an **incidental finding.** The incidental finding may be discussed with you if it is considered to be important for you or your blood relatives' health.

An example of an incidental finding: A person has a genomic test for kidney disease and the test finds they are a carrier of a gene variant that causes cystic fibrosis (CF). Although CF is a genetic condition it is not a kidney condition. It is not related to the reason for testing. Finding the person is a carrier for CF was an incidental finding.

3. Unexpected family relationships may be discovered in genetic and/or genomic testing for example when a person's parent is shown not to be their genetic parent. This only happens in some circumstances. You may wish to speak to your health professional if you think this is a possibility in your family.



4. INSURANCE:

Health insurance: In Australia, genetic and genomic testing will not stop you from getting health insurance or impact how much you will pay for your health insurance. However, a diagnosis of a condition may mean that health insurance companies may apply waiting periods for treatment of the condition.

Income protection, life insurance or travel insurance: Genomic test results do not affect existing insurance cover. If you already have signs or symptoms of a genetic condition, this may affect how easy it is and/or the cost to get new income protection, life insurance or travel insurance in the future. In some circumstances, the genomic test results may also impact how easy it is and/or the cost for you (and your genetic parents, if undergoing trio testing; defined in the table below) to get these types of insurances.

Under the current industry-led regulation, Australians can purchase new life insurance policies or increase their current level of cover without the results of previous genomic tests being taken into account, up to certain financial limits. For cover over specific limits, you can be required to disclose genomic test results that show increased risk. Industry regulation prevents insurers from asking blood relatives for your genetic test results and from requesting you to have testing.

Your health care provider will **not** provide your results to an insurance provider without your permission.

Further details can be found below by clicking on the links or scanning the QR codes:

Centre for Genetics Education - Life Insurance Fact Sheet

Financial Services Council - Moratorium on Genetic Tests in Life Insurance

Understanding how genetic and genomic testing can impact insurance can be confusing. Ask your health professional if you have any questions or concerns.

SUPPORT

If you need support, there is a large network of patient support groups that can help you through your experience. You can access these groups at any time. Check out the support groups listed below for organisations specific to your state. There are also many social media groups online that bring families together who have the same genetic condition. Ask your health professional for help choosing the right support group for you.

National

Rare Voices Australia
rarevoices.org.au
0497 003 104

SWAN Australia
swanaus.org.au
0404 280 441

Genetic Alliance Australia
geneticalliance.org.au
(02) 9295 8359

QLD

Support Groups Queensland
supportgroups.org.au
(07) 3344 6919

VIC

Genetic Support Network Victoria
gsnv.org.au
(03) 8341 6315

SA

Lived Experience Telephone Support Service
letss.org.au
1800 013 755

NT

Northern Territory Mental Health Line
1800 682 288

WA

Helping Minds
helpingminds.org.au
(08) 9427 7100

NSW

Genetic Alliance Australia
geneticalliance.org.au
(02) 9295 8359

National Mental Health Support Services

Lifeline Australia
24/7 Crisis Support
13 11 14

Beyond Blue
beyondblue.org.au
1300 224 636

Scan here for **Gene Equal's Easy Read Health Booklets** that can help you learn more about genetic healthcare and make choices about genetic and genomic testing.

GLOSSARY

Blood Relative - Anyone who is related to you by genetics. This could be your ancestors, your biological family, or someone who is not born yet.

Exome Sequencing - A genomic test that looks at about 1-2% of a person's genome to find variants.

Gene Panel - A genomic test that focusses on just the genes that could be related to a person's condition.

Genetic Condition - A health condition that is caused by a person's genetics and affects how their body works.

Genetic Test - A genetic test looks at a single or a small number of genes at a time. The test may look for a single variant or multiple variants.

Genome - All of a person's genetic information.

Genome Sequencing - A genomic test that looks at most or all of a person's genome to find variants.

Genomic Test - A genomic test looks at many genes, sometimes all 20,000-25,000 genes at once.

Health Information - This may include samples, test results, hospital entries or primary care data.

Incidental Finding - A result that shows something that is not related to the reason for testing, such as an unrelated health risk or an unexpected family relationship.

Misattributed Parentage - When the assumed parent of a child is not the biological parent.

Variants - Differences in a person's genes.

Variants of Uncertain Significance - A variant has been found, but it is not known at this point in time whether it is or is not the cause of your or your child's condition.

Australian Genomics

CONSENT FORM - EXPLANATIONS

See the table below for explanations about the clauses on the Consent Form. These are shown in the same order as shown on the Consent Form for your reference.

CLAUSE	EXPLANATION
The test does not detect all genetic changes or predict all possible health conditions.	We carry many genetic changes (**variants**) in our DNA. The test only looks for variants related to your condition. This means that it does not detect all variants that you may carry. Therefore, the test cannot predict all possible health conditions that you may develop in the future.
Genomic Testing Consent Form Only **The test may find a genetic change not related to the reason for testing ('incidental finding').**	An incidental finding may be discussed with you if it is considered to be important for you and your blood relatives' health. See above in **'What are other issues to think about?'** for more information.
The test may find a genetic change of uncertain significance.	If a **variant of uncertain significance** is found, more testing may be done to try to find out what this means.
More tests or analysis may be needed to understand the results. This may include testing blood relatives.	The test results/sample may be reanalysed in the future. This may be done on the data that has already been collected, or a new sample might be required. If further testing is carried out, it may or may not identify the cause of your condition. Your blood relatives may need to be tested to help your health professional to understand your genetic condition and/or find out how a variant is passed down through your family. Testing blood relatives can also help better understand variants of uncertain significance.
The sample or results may be re-examined in the future using new knowledge of testing methods.	If no variant is found as the cause of the health condition, it may be possible to find a variant in the future if testing technology improves or if understanding about the genetic cause of the condition changes.
Results may have health implications for blood relatives.	Testing may impact your blood relatives as it could raise concerns for their health. Sometimes, testing your blood relatives can help your health professional to understand your condition. See **'What are other issues to think about?'** for more information.
Results may show unexpected family relationships.	In some circumstances, unexpected family relationships may be discovered through genetic and/or genomic testing. See **'What are other issues to think about?'** for more information.

CLAUSE	EXPLANATION
Results may affect the ability to obtain some types of insurance.	Please see **'What are other issues to think about?'** for more detail. Understanding how genetic and genomic testing can impact insurance can be confusing. Ask your health professional if you have any questions or concerns.
The sample will be stored and may be shared with other laboratories to assist with genomic testing.	Genetic information from the test will be stored securely using systems that meet Australian and international privacy and security standards and laboratory guidelines. Genetic results are confidential, will be stored in medical records and will only be shared outside of the health system with your consent, unless required or permitted by law. Once the storage period of the sample has expired (as suggested by industry guidelines), you may ask to have the sample destroyed. Talk to your health professional if you wish to have your sample destroyed earlier. Your data may be used for the purpose of quality control, internal validation, training purposes and test improvement by the laboratory that organises your test.
Results and related health information may be shared with genomic and medical databases that are used for patient care. All identifying information will be removed.	Sharing genomic data and health information can advance medical knowledge to improve the chance of a diagnosis for you or others like you. This includes sharing test results and clinical information with large secure databases. Comparing your results with others like you may help to improve our understanding of your condition. When data are shared there are safeguards in place to help protect your privacy, such as: • Personal identifiers such as names and address are removed (de-identified) • Security measures preventing unauthorised access or misuse. There is a very small chance that you or your child might be re-identified. One way this could occur is if someone has your genomic data and matches it to those found in the database.
Results are confidential and will only be shared with consent, or as required or permitted by law.	The health professionals involved in your care may also order further testing or share your genomic data with each other to help work out what your test results mean. The results, genomic data and identified sample will not be used or disclosed outside of your or your child's care without your consent, unless required or allowed by law.
I can change my mind about testing and choose not to be told the results, but if testing has started, a report will remain in medical records.	If testing has not begun, you may withdraw your consent. If testing has begun, you can change your consent and choose not to be told the results of the test. However, the report may remain in medical records depending on at what stage you make this decision. Sometimes the laboratory will have already started analysing the DNA sample. If you decide not to be told the results at this stage, the results of the laboratory's analysis will be entered into their records but not disclosed to you. Reports cannot be removed if they have already been entered into your medical record. If you choose not to receive your results, it will not impact your relationship with your health professional.

OPTIONAL CONSENT FORM CLAUSES

CLAUSE	EXPLANATION
I consent to share the results and related information with health professionals to help with the genetic testing of blood relatives. I understand that identifying information will not be disclosed to the relative wherever possible.	Your test results may help your blood relatives with their health care. Identifying information such as your name and address will be removed before your results are shared. Your results will not be shared with your blood relatives directly, but with the health professionals providing their care.
I consent to share the sample, genomic data, and related health information for ethically approved research into the same or related conditions. I understand identifying information will be removed and will usually be replaced with a unique code so that information can be returned to me in some situations.	You may provide consent to share data for research that is investigating the same or a related health condition. In this case, names will usually be replaced with a unique code so that samples are de-identified. It is unlikely that you will receive any personal results from sharing your data for research. The results of most data sharing research projects do not have implications for a specific person's clinical care, so the findings are not returned to participants. In rare situations, there may be findings that are important for your clinical care, and it may be possible to be re-identified, so that your results can be returned to you. All researchers are bound by the law and ethical guidelines. Research will only happen for projects approved by a Human Research Ethics Committee. You may be recontacted in the future and asked to participate in other research projects. It is your choice whether you want to take part in research. Although participating in research is unlikely to directly benefit you, your data could help others. By comparing patient data, scientists can achieve a deeper understanding of the human body from a genetic perspective and learn how to prevent and treat genetic conditions. Please let your health professional know if you would prefer to talk about research at a later time, instead of in your first appointment.
• **I consent to genomic testing.** • **I understand the reason for testing and the potential benefits, consequences, and limitations.** • **I have been able to discuss the information with a health professional, ask questions and have any concerns addressed.** • **I am satisfied with the explanations and answers to my questions.**	It is important that you understand all the information in this document and the **Consent Form** before you provide your consent for testing. Some of your questions may not be answered in this Fact Sheet. Therefore, do not hesitate to ask your health professional if you have other questions.
Consent for Parents Undergoing Duo/Trio Genomic Analysis	Sometimes parents need to be tested to better understand their child's results. This is called **trio testing**. In the case of trio testing, the report may be put in the child's records only, and the parents may not get a separate report. Ask your health professional about receiving these results.