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 and to help you think of any other questions you might have. You need to give your consent before the testing starts. 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. Will it cost anything to have testing?
6. What might it mean for my family if I or my child gets the test?
7. What support is available for me during testing and after receiving the results?
8. Will I need to go to a clinical genetic service?
9. Will the test change the way I or my child receive health care?
10. How will the test be performed?

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

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?

choosingwisely.org.au

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 (**variants**) in their genes. Most **variants** are harmless and do not impact how the gene works. However, some people may have **variants** that do affect how a gene works, which 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. A **genetic** test may be suggested by your health professional to look for variants that change how a gene works if you have unexplained health issues.

HOW IS THE TEST DONE?

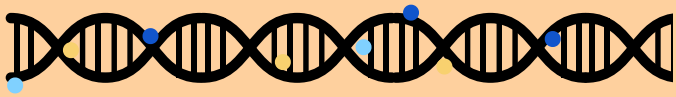
Both genetic and genomic tests require a sample of DNA. **Blood** and mouth swabs are the most common samples used. You do not need to fast for the blood test.

WHAT IS THE DIFFERENCE BETWEEN A GENETIC AND 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.

Genome sequencing looks at most or all of 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 condition.

WHAT IS THE PURPOSE OF THE TEST?

The purpose of having genetic or genomic testing may include:

- diagnosing a genetic condition (**diagnostic**)
- finding out if you have a variant that has caused a genetic condition in your family. This may impact your health in the future (**predictive**)
- seeing if you carry a variant that could be passed onto your children, but which is unlikely to affect your health (**carrier**)
- diagnosing a genetic condition in your pregnancy (**prenatal**)
- confirming a clinical diagnosis you have previously received (**confirmation**), or
- understanding how a variant that has been found previously, has been passed down in your family (**segregation analysis**).

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 relatives know the chance of developing a condition.
- To provide information about the chance of having a child with the same condition.

WHAT ARE THE POSSIBLE RESULTS?

The result of your test 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.

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CONSENT FORM:

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 in our DNA. The test only looks for genetic changes related to your condition. This means that it does not detect all genetic changes 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').	A genomic test may show something that is not related to the reason for testing such as an unrelated health risk or an unexpected family relationship. 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 family.
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.
The sample or results may be re-examined in the future using new knowledge of testing methods.	If no variant is found, it may be possible to find one in the future if testing technology improves or understanding about the genetic cause of the condition changes.
Results may have health implications for 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.
Results may show unexpected family relationships.	This only happens in some circumstances.
Results may affect the ability to obtain some types of insurance.	In Australia, genetic and genomic testing will not stop you from getting health insurance or impact your health insurance premiums. However, it could affect how easy it is for you or other family members to get some types of insurance in the future. These include income protection, travel insurance and life insurance. Testing could impact the price of your premium for new products or different cover in the future but will not impact existing cover you may have. Until 2024, Australians can purchase new life insurance policies without the results of previous genetic tests being taken into account. However, an existing clinical diagnosis may affect your ability to obtain these kinds of insurance. Industry regulation prevents insurers from asking 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 by searching for 'Moratorium' at fsc.org.au/resources. Understanding how genetic and genomic testing can impact insurance can be confusing. Ask your health professional if you have any questions or concerns.

CLAUSE	EXPLANATION
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 legal storage period of the sample has expired, you may ask to have the sample destroyed. 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 scientific knowledge to improve the chance of a diagnosis for you or others like you in the future. This includes sharing test results and clinical information with large secure databases. This may help improve our understanding by comparing your results to those of other people. 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. Ways this could occur include if someone has your genomic data and matches it to those found in the database, or you have an extremely rare condition, and your data is included in a journal article. In this case, someone may be able to identify you from the article.
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 choose not to be told the results if I change my mind, but the report will remain in medical records.	The testing process can be stopped at any time, and you can 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. Results cannot be removed if they have already been entered into your personal records. If you choose not to receive your results, it will not impact your relationship with your health professional.

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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 with the health professionals in 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 may be replaced with a unique code so that information can be returned to me in some situations.	You may wish to provide consent to share data for research that is investigating the same or a related health condition. In this case, names may be replaced with a unique code so that samples remain de-identified. If there are findings from this research that have implications for your clinical care, it may be possible to be re-identified, so that your results can be returned to you in some situations. Not all research projects return their findings to their participants. 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 may not 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.
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 of the information on this document and the Consent Form before you provide your consent for testing. All of your questions may not be answered in this Fact Sheet. Therefore, ask your health professional if you require further explanation.
Consent for Biological Parents Undergoing Duo/Trio Genomic Analysis	In the case of trio testing where both parents and their child are tested, 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.

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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 in the process. Check out the support groups listed 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

SWAN Australia
swanaus.org.au

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

NSW

Centre for Genetics Education
genetics.edu.au

QLD

Support Groups Queensland
supportgroups.org.au

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

Mental Health Support
Lifeline Australia
24/7 Crisis Support
13 11 14

Beyond Blue
beyondblue.org.au
1300 224 636

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 jus 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.

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.

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.

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