PATIENT FACT SHEET

TISSUE-TARGETED (SOMATIC) GENOMIC TESTING

Australian Genomics

This fact sheet is to help you understand the consent form and the possible results of tissue-targeted (somatic) genomic testing. You (or the person in your care) can share this Patient Fact Sheet with health professionals, family, and friends to help decide if you want to have the test. It may also help you think of questions you might want to ask. Before testing can start you need to agree to have the test (give your consent). You can change your mind and stop the process any time before you get your results. **Having the test is your choice.**

GENES AND GENETICS

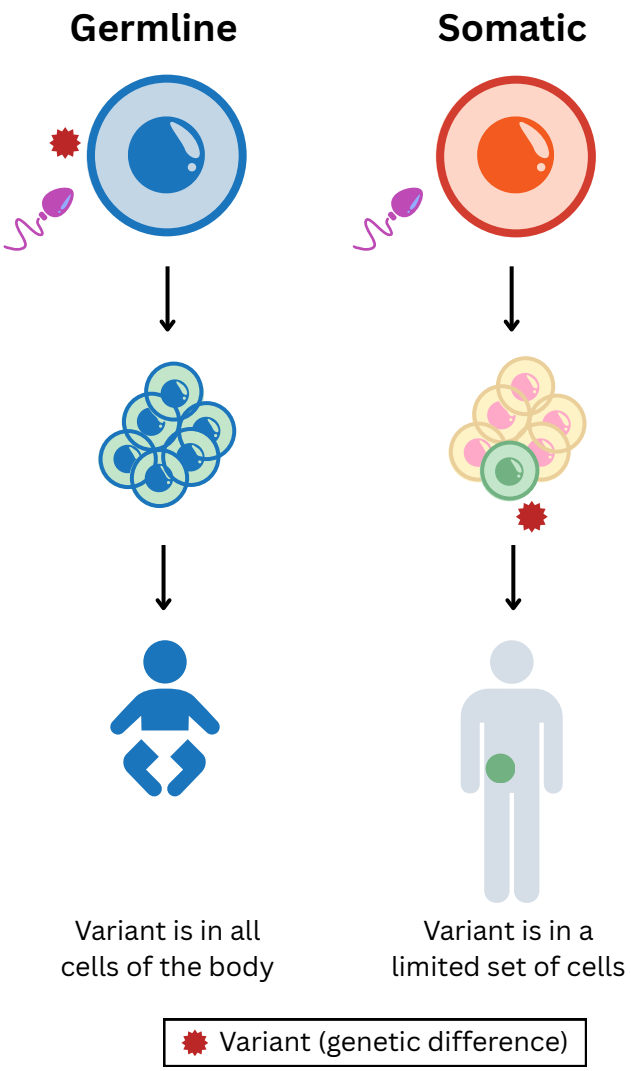
Genes are instructions that tell our bodies how to grow and develop. Genes are made up of DNA. All of a person's genetic information in their body is called their **genome**.

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.

There are two types of variants:

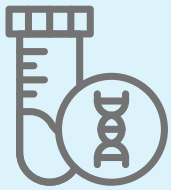
- **Germline** (or hereditary) variants are genetic differences that a person is born with and are found in all cells in the body. They may have been passed down from a parent or may have happened for the first time in the person. These variants may be passed on to children.
- **Somatic** (or acquired) variants are genetic differences that occur during a person's life, in a limited set of cells or tissues. Unlike germline variants, they are usually not passed down from a parent and cannot be passed on to children. Tissue-targeted genomic testing looks for these types of variants.

Many tumours/cancers/lesions happen because of **somatic** variants that occur in a cell. These variants can happen by chance or may be caused by things such as radiation (e.g. from the sun), harmful chemicals, tobacco use and ageing. However, up to ten percent of tumours/cancers/lesions are caused by a **germline** variant, which is a variant someone is born with.



HOW IS THE TEST DONE?

Tissue-targeted (somatic) genomic testing needs a sample of DNA or other genetic material from the tumour/cancer/lesion. This may be done on a sample that is already stored or a new sample may need to be taken *(Consent form: clause 4)*.



WHAT IS THE PURPOSE OF THE TEST?

The purpose of tissue-targeted genomic testing is to look for somatic variants in tumour/cancer/lesion cells *(Consent form: clause 1)*.

Tissue-targeted (somatic) genomic testing can provide information related to *(Consent form: clause 2)*:

- **Diagnosis:** results can help confirm or improve the accuracy of a diagnosis.
- **Treatment options:** results may help doctors choose treatments better suited to the tumour/cancer/lesion type or figure out if you can join a clinical trial.
- **Prognosis or recurrence risk:** results may help predict how a health condition might develop or progress (prognosis) or can tell us whether a tumour/cancer/lesion might come back (recurrence risk).

WHAT ARE THE POSSIBLE RESULTS?

The result of testing may be:

- **A variant is found** that may be relevant to the diagnosis, prognosis or treatment of your health condition.
- **No relevant variant/s** are found:
  - no variants are found that are relevant to the diagnosis, prognosis or treatment of your health condition, or
  - a variant of uncertain significance is found. This means that currently, it is not known whether the variant is linked to your health condition.

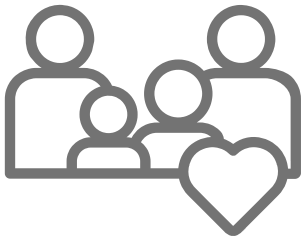
A VARIANT OF UNCERTAIN SIGNIFICANCE

Right now, there is still a lot we don't know about how variants affect health. As research continues, scientists learn more about how different variants are linked to specific health conditions. In the future, a variant that is unclear today may be reclassified as either relevant or not relevant to the condition.

The health service may keep your information on file. They might contact you (or your doctor) in the future if new information becomes available that changes the meaning of your test results. If you have questions or want updates in the future, you can also ask your doctor.

WHAT OTHER ISSUES SHOULD I THINK ABOUT BEFORE TESTING?

- Although unintended, tissue-targeted genomic testing may find a **germline** variant. This means the variant is not just in the tissue tested, but likely something you were born with. This germline variant may have health outcomes linked and/or not linked to your health condition. It could also be relevant to the health of your blood relatives. If this happens, more testing might be needed, and you may be referred to a doctor who specialises in genetics. Your doctor will ask your permission to share this information with health professionals to help with the testing of blood relatives *(Consent form: clause 3 and 8)*.



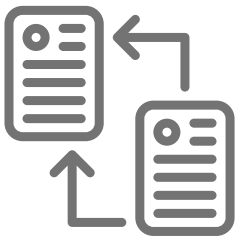
- Testing may unexpectedly find a **somatic** variant that is not related to the reason for testing. This could point to an unrelated health risk that was not being looked for. This is called an incidental finding, and it does not happen very often *(Consent form: clause 3).*

WITHDRAWAL FROM TESTING

It is your choice whether or not you have this test. If you change your mind, you can decide not to go ahead with the test or receive the results. If the test has already been done, the results might still be kept in your medical record, but you do not need to find out what the results are. Please talk to your health professional if you would like to withdraw from testing *(Consent form: clause 7).*

WHAT HAPPENS TO MY DATA AND SAMPLE?

Sharing with clinical databases: The health professionals helping you might also order more tests or share your genomic or health information with each other to better understand your results. Sharing your genomic and health information may also help doctors learn more about the health condition, lead to better treatments, improve our understanding of what to expect for you and others or might improve the chance of getting a diagnosis. This sharing of information may include sharing genomic test results and medical information with secure clinical databases.



All identifying information will be removed when sharing with these databases. Other than what is stated here in the fact sheet and consent form, the results, genomic data and identified sample will not be provided to third parties outside of your care, without your consent, unless required or allowed by law *(Consent form: clauses 5 and 6)*.

Review of your data or sample in the future: The test results or sample may be looked at again in the future. This might be done on the results/sample already collected, or a new sample might be needed. This re-examination might provide useful information as testing techniques improve over time and as we learn more about how different gene variants affect the health condition.

Your doctor may contact you if new information becomes available that changes the meaning of your test results. If you have questions or want updates in the future, you can also ask your doctor *(Consent form: clause 4).*

GLOSSARY

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

Genetic information - The set of instructions inside your body. It helps determine how your body looks, works and grows.

Genome - All of the genetic information in a person's body.

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

Germline variant - a genetic variant that can be found in all cells of the body and has been present since birth. These variants may have been inherited or may have happened for the first time in the person. These variants may be passed on to children.

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

Somatic variant– a genetic variant that has been acquired after birth in a limited set of cells or tissues. These variants usually cannot be passed on to children.

Variants - Differences in a person's genetic information. These differences may affect how the body works. Not all variants cause health conditions – many have no health effects at all.

Variants of Uncertain Significance - A variant has been found, but it is not known at this point in time whether or not it is linked to the health condition.