PATIENT FACT SHEET

TISSUE-TARGETED (SOMATIC) GENOMIC TESTING

Australian Genomics

This fact sheet aims to help you understand the consent form and the possible **results** of tissue-targeted (somatic) 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 your results are returned. Testing 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 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:

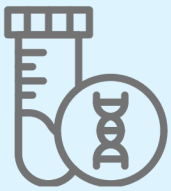
- Acquired (arising during a person's lifetime; also called **somatic** variants), and
- Hereditary (inherited from a parent or new in the individual; also called **germline** variants).

Many tumours/lesions are the result of acquired variants that occur in a cell. These variants are often caused by factors such as radiation (eg. from the sun), chemical exposure, tobacco use and aging or can occur by chance. However, up to 10% of tumours/lesions are caused by a **germline** variant.

Tissue-targeted genomic testing looks for **somatic** variants in tumour/lesion cells.

HOW IS THE TEST DONE?

Tissue-targeted (somatic) genomic testing requires a sample of DNA or genetic material from the tumour/lesion. This may be performed on an existing stored sample or may require a new sample.



WHAT IS THE PURPOSE OF THE TEST?

Tissue-targeted (somatic) genomic testing can provide information related to:

- **Diagnosis**: Genetic information may lead to an accurate diagnosis
- **Targeted treatments**: Enable treatments better suited to the tumour/lesion type or potential access to clinical trials
- **Recurrence risk or prognosis**: Predicts the outlook (prognosis) or the likelihood of recurrence of the condition.

WHAT ARE THE POSSIBLE RESULTS?

The result of testing may be:

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

WHAT ARE OTHER ISSUES TO THINK ABOUT BEFORE TESTING?

- Although unintended, testing may identify a **germline** variant which may be relevant to the health of blood relatives.
- Testing may unexpectedly identify a **somatic** variant that is not related to the reason for testing, such as an unrelated health risk. This is uncommon.

GLOSSARY

Acquired or somatic variant – A genetic variant that has been acquired after conception in a set of cells or tissues. Somatic variants cannot be passed on to children.

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.

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

Genome – All of a person's genetic information.

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 conception. These variants can be inherited or new in the individual from the point of conception. Germline variants may be passed on to children.

Health information – This may include samples, test results, hospital entries or primary care data.

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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