Somatic vs Germline Genetic Testing

Understanding Genetic Testing in Cancer

The Simple Explainer

When someone is diagnosed with cancer, doctors may recommend genetic testing to learn more about why the cancer developed, inform the risk of another cancer, and which treatments are most likely to work. There are two main types of genetic testing:

1. **Somatic Genetic Testing** tests the DNA of the tumour
2. **Germline Genetic Testing** tests the DNA from all of the person's cells

Both types of genetic testing provide important information. **Somatic DNA changes** occur only in the cancer cells and are not present in the rest of the body. These changes develop over a person's lifetime and are not passed on to children. In contrast, **germline DNA changes** are present in every cell of the body from birth. These changes may sometimes help explain why a person developed cancer and may have implications for family members' health. Together, both kinds of genetic testing may give a fuller picture of what's happening in a cancer diagnosis.

Somatic vs Germline: A Basic Comparison

1. Somatic (Tumour) Testing

- Looks at the genes inside the tumour itself.
- May help identify factors contributing to cancer
- Can guide doctors regarding best treatment options.
- Most gene changes found in tumours arise sporadically.
  - Sometimes changes found in somatic testing mean that germline testing is indicated.

2. Germline Testing

- Looks at the genes you were born with.
- It helps understand whether a gene change runs in the family and what steps may help manage cancer risk.
- Most germline changes are inherited, but some can occur for the first time in a person.

What happens if a somatic (tumour) test finds a gene change?

Most genetic changes in tumours are acquired and not inherited. However, if a change is known to be associated with inherited cancer (for example, in *BRCA1*, *BRCA2* or MMR genes), doctors may recommend a germline test to see if it is present in all cells and could have implications for family members.

Accessing Somatic or Germline Testing in Australia

In Australia, many somatic (tumour) tests are now eligible for Medicare rebates when they guide treatment decisions. Germline tests can also be Medicare-funded when certain clinical criteria are met.

Germline vs Somatic Testing: A Detailed Comparison

	Germline Genetic Testing	Somatic (Tumour) Genetic Testing
What it is looking for	• Gene changes present from birth in every cell, via samples such as blood or saliva. • These changes may have contributed to the development of cancer and can increase lifetime cancer risk. • Most are inherited, although some can occur for the first time in a person.	• Gene changes that develop only in the tumour. These changes are not inherited and cannot be passed on to children. • These gene changes develop over time and can drive cancer growth.
Why it is done	• To understand whether a gene change present from birth contributed to the development of cancer, and whether there may be an inherited cancer risk.	• To understand the genetic changes within a tumour that may be driving cancer growth and behaviour.
What it may inform	• Screening, prevention, or early detection options • Whether there may be increased risk of a second primary or other cancers • Risk assessment and risk management for family members • Treatment decisions in the context of some cancers	• Treatment choices • Whether certain targeted therapies may be effective • Predictions regarding how the cancer may behave

	Germline Genetic Testing	Somatic (Tumour) Genetic Testing
Limitations	• Not all gene changes are tested in every panel. • Results may be uncertain or unclear. • A gene change does not mean cancer will definitely develop. • Results can have emotional or family impacts.	• Only reflects the tumour tested. • Some gene changes may be missed depending on the test used. • Results may change over time or differ between tumours • Does not explain inherited cancer risk.
Accessibility in Australia	• Medicare funding may be available if certain clinical criteria are met. • Referral is usually through a specialist or genetics service.	
When both might be considered	• In some situations, doctors may use both somatic and germline testing, depending on the cancer type and the questions being asked. • For example, in some cancers, tumour testing may help guide treatment, while germline testing can help determine whether a gene change present from birth contributed to the cancer and whether this information may be relevant for family members or future cancer screening. • Your care team will decide which tests are appropriate based on individual circumstances.	
If a somatic change is found	• Most tumour changes are not inherited and only affect that cancer. But sometimes a tumour result does raise the suspicion of a germline gene change. • If the same change is known to be associated with inherited cancer risk, a genetics referral is recommended to check if it is present in the germline. • If the change is found to also be germline, relatives may also be offered testing, screening, or preventive strategies.	