



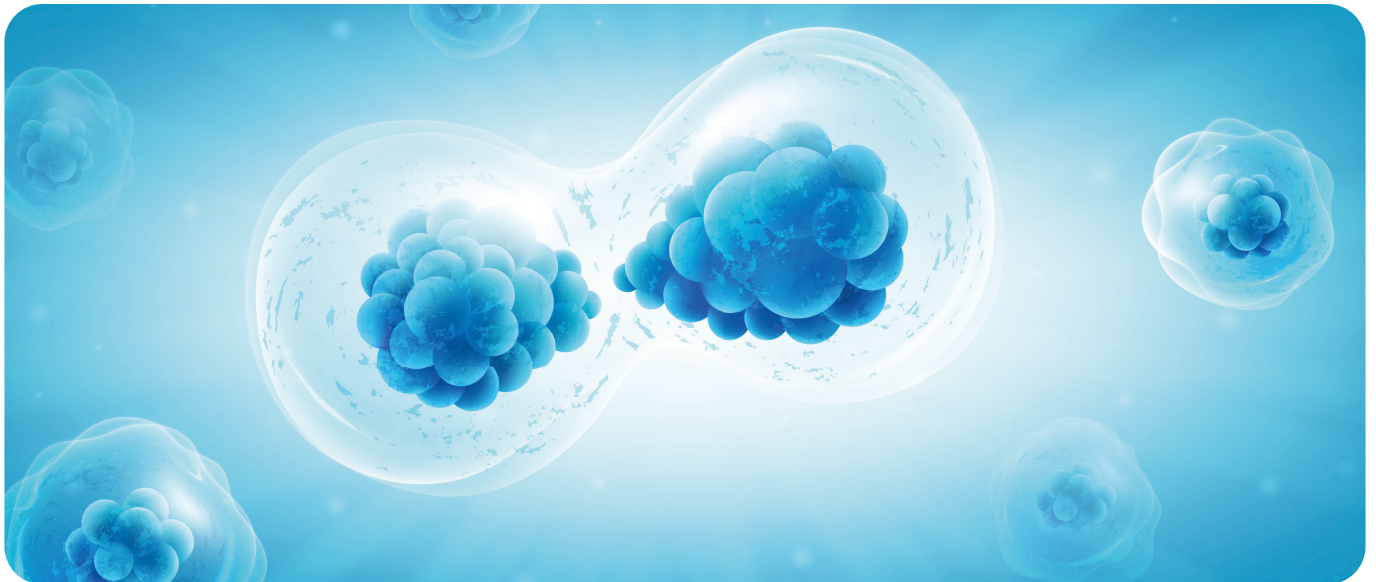
# PATHOLOGY TESTS EXPLAINED

Information about pathology tests to help everyone take control of their health and make the right decisions about their care.

## WHAT YOU SHOULD KNOW ABOUT **SOMATIC TUMOUR TESTING**

If you have cancer your surgeon or oncologist will send a sample of the tumour to a pathologist for investigation under a microscope. The pathologist describes their findings in a report containing details about your diagnosis and this is sent to your doctor.

Sometimes a microscopic assessment of the tumour cells does not provide all the information needed and your tumour sample is passed on to a genetic pathologist for somatic genetic testing. This looks for specific gene mutations in the cancer cells and can give important information about what sort of cancer you have, how it can be expected to behave and show if you will benefit from one of the newer targeted treatments or immunotherapy drugs.



### What is a somatic mutation?

Genetic mutations are classified as either somatic or germline. While germline mutations are changes to your DNA that you inherit from your parents, somatic mutations are changes to your DNA that happen during your lifetime. Most genetic mutations that cause cancer are somatic. This means you were not born with them. Only tumour cells carry the DNA mutations.

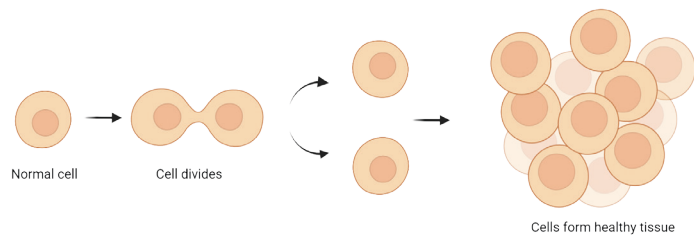
### Why do somatic gene mutations matter in cancer?

Our genes control the way our body works by sending instructions to make proteins – our body's building blocks. If a gene has a mutation, it can affect the structure, function and amount of the proteins being made.

Gene mutations occur during the normal course of life. Our cells have a lifespan – they get old and die off. Before they die, they copy themselves and this can lead to mistakes being made when genes are copied. Normally, our immune system gets rid of these copying mistakes but as we age it gets less good at this.

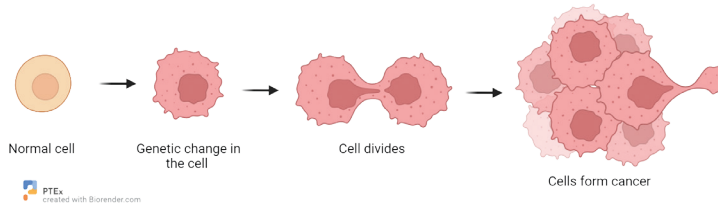
Genes can also mutate because of environmental factors like smoking and ultraviolet radiation – think of the sun causing skin cancer. Also, a virus or bacterial infection can cause inflammation that eventually leads to cancer.

## Normal cells



vs

## cancer cells



## What somatic tumour testing can do

Many different kinds of mutations are found in cancer cells. Often these are specific to the type of cancer. Targeted therapies have been designed to interfere with the action of these mutated genes. Somatic genetic testing can identify the genes to show which treatments will work and which won't.

For example, BRCA1 and BRCA2 gene mutations in breast cancer can be treated with a drug called Lynparza.

When it comes to skin cancer, melanoma is the most dangerous type of skin cancer and there are several genes involved in its development. The most common of these are BRAF and MEK. The BRAF gene mutation can be

treated with a BRAF inhibitor, Dabrafenib and the MEK gene mutations can be treated with a MEK inhibitor, Trametinib. Using a combination of these two drugs has been found to be more effective at shrinking melanoma tumours than using either type of drug on its own.

Some testing focuses on a specific group of cancers. This includes testing a panel of genes most relevant to your type of tumour.



## What can your results tell you?

A pathology report will be prepared and sent directly to your surgeon and/or oncologist.

A positive test result means a genetic mutation has been identified in one or more of the genes tested. The mutations are termed pathogenic or disease-causing variants.

The variants of strong or potential clinical significance are highlighted and the results will help your doctors select the most appropriate treatment for the mutations identified.

Repeat testing can be used to monitor the effectiveness of your treatment and potentially direct a change in ongoing therapy.



## Questions to ask your doctor

Why does this test need to be done?

How could it change the course of my care?

What will happen next, after the test?

For more detailed information on these and many other tests go to [pathologytestsexplained.org.au](http://pathologytestsexplained.org.au)



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EXPLAINED

[www.pathologytestsexplained.org.au](http://www.pathologytestsexplained.org.au)

Pathology Tests Explained is the primary national source of consumer information on pathology testing. Information is written and edited by practising pathologists and scientists, including leading experts. This ensures integrity and accuracy.

Pathology Tests Explained is managed by a consortium of medical and scientific organisations representing pathology practice in Australia. More details at: [www.pathologytestsexplained.org.au/about](http://www.pathologytestsexplained.org.au/about)



Please use this QR code to access more information

## My Health Record

You'll find a direct link to the Pathology Tests Explained website embedded in the pathology results pages of your My Health Record.

Click on the link to find information about what your tests are investigating or measuring and what your results can tell your doctor.