

Understanding Genetic Testing for Cancer

A guide for people considering
diagnostic genetic testing

Table of contents

PAGE

01

Introduction

PAGE

02

Genetic testing
and cancer

PAGE

04

Why Is Genetic Testing
Recommended?

PAGE

06

The genetics
of cancer

PAGE

09

Genetic testing
for breast cancer:
Multi-gene panels

PAGE

13

How Genetic Testing
Can Help Your Treatment
and Ongoing Care

PAGE

18

Practical considerations
regarding genetic testing

PAGE

21

Why Genetic Testing
Matters for Your Family

PAGE

23

Genetic test results
and reproductive
decision making

PAGE

24

Support
and Resources

PAGE

26

Glossary

PAGE

30

Notes



Being told you or your loved one may benefit from genetic testing can feel overwhelming, especially when you are already navigating a cancer diagnosis or supporting someone who is.

This booklet has been created to help you understand what genetic testing is, why it may be recommended, and what the results could mean for you and your family.

Genetic testing can provide important information about the cause of your cancer diagnosis, your cancer treatment, your future health, and whether relatives may also be at increased risk. Many people find that having clear, reliable information helps them feel more prepared and more in control of their healthcare decisions.

In this booklet, you'll find an explanation of the typical genetic testing process, and what to expect before and after receiving your results. We also outline management options for people found to have an inherited cancer risk, and talk about the emotional, practical, and family considerations that often arise.

We know that real stories can be powerful. Included throughout this booklet, you will find quotes from two community members who have been through inherited cancer testing and want to share what helped them along the way.

Finally, this booklet connects you with evidence-based national resources and supportive care services that offer peer support, online groups, access to information days, and specialised support.

Our goal is to ensure you feel informed, supported, and empowered at every step of your genetic testing journey.

The information in this booklet is designed to support, not replace, conversations with your doctor, genetic counsellor, or familial cancer service.

Genetic testing and cancer

When someone is diagnosed with cancer, doctors may recommend genetic testing to better understand why the cancer developed, assess the risk of future cancers, and help determine which treatments are most likely to be effective. This type of genetic testing is often referred to as diagnostic genetic testing.

The outcome of diagnostic genetic testing also provides valuable information to family members regarding their own risk of inherited cancer and their risk management options.





Genetic information was powerful.
It changed my treatment. It likely
saved me from future cancer.”

Samira

BRCA2 pathogenic variant

Diagnosed with breast cancer at 25



Genetic Testing Is Your Choice

For some people, genetic test results can help doctors tailor care more specifically.

This may include identifying treatment options that work particularly well for certain inherited gene changes, guiding eligibility for targeted therapies or clinical trials, or informing screening and prevention strategies for future health.

For others, testing may not change current treatment but could still provide useful information for future care or for family members.

At the same time, genetic testing can raise complex or unexpected information, and not everyone feels ready to have this information straight away. Deciding whether to have testing is a personal choice that depends on your values, preferences, and what feels right for you at this time.

It is important to know that genetic testing is optional. While there can be important benefits, it should not be viewed as routine or compulsory.

Choosing not to have genetic testing will not prevent you from receiving appropriate cancer treatment, care, and support based on the clinical information available.

PEOPLE CAN CHOOSE TO:

- proceed with genetic testing now
- defer genetic testing to a later time
- decline genetic testing altogether

Your healthcare team can support you, whichever option you choose.

Why Is Genetic Testing Recommended?

Genetic testing may be recommended when the result could help guide your cancer treatment, inform your future health care, or provide important information for your family.

In Australia, in addition to diagnostic genetic testing being available through clinical genetics services, it is increasingly offered directly by cancer specialists (such as oncologists or surgeons) as part of routine care for some cancers. Your doctor may offer testing based on your type of cancer, your age at diagnosis, your personal or family history of cancer, or because the result may influence treatment options. Recent changes to Medicare funding have also made testing more widely available for people with breast, ovarian, prostate and pancreatic cancers.

YOU MAY BE OFFERED TESTING IF:

- You have been diagnosed with breast, ovarian, prostate, or pancreatic cancer
- You were diagnosed with cancer at a younger age than expected
- Genetic test results may help guide targeted therapies or treatment pathways
- You have multiple relatives with related cancers
- There is a known inherited gene variant in your family that is associated with an increased risk of cancer (in which case targeted testing may be considered, often with genetic counselling)

Your doctor will explain why testing is recommended in your specific situation.

Types of genetic testing

Germline genetic testing

Germline genetic testing looks for inherited gene variants that are present in every cell of your body from birth. These variants can sometimes help explain why a person developed cancer and may have implications for family members, as they can be passed on to children. This is the type of testing discussed in this booklet.

Tumour (somatic) testing

Tumour (or somatic) testing looks at the DNA changes found only in the cancer cells. These changes develop over a person's lifetime, are not inherited, and cannot be passed on to children. However, they can provide important information to help guide cancer treatment.

Both types of genetic testing can be valuable, and together they may provide a more complete picture of a cancer diagnosis. Sometimes, one or both type of testing is offered. Your doctor will tell you which tests are relevant for you.

The genetics of cancer

What is genetic testing looking for?

To understand what genetic testing is looking for, it helps to have a basic understanding of cancer genetics.

Cancer is unfortunately very common, but the reasons it develops vary. Most cancers are sporadic, meaning they occur by chance and are not linked to inherited factors.

Sometimes, cancer appears to “run in families”, these are called ‘familial’ cancers. Familial cancers may be influenced by shared genes, lifestyle, or environmental factors, but there is often no single identifiable inherited genetic cause.

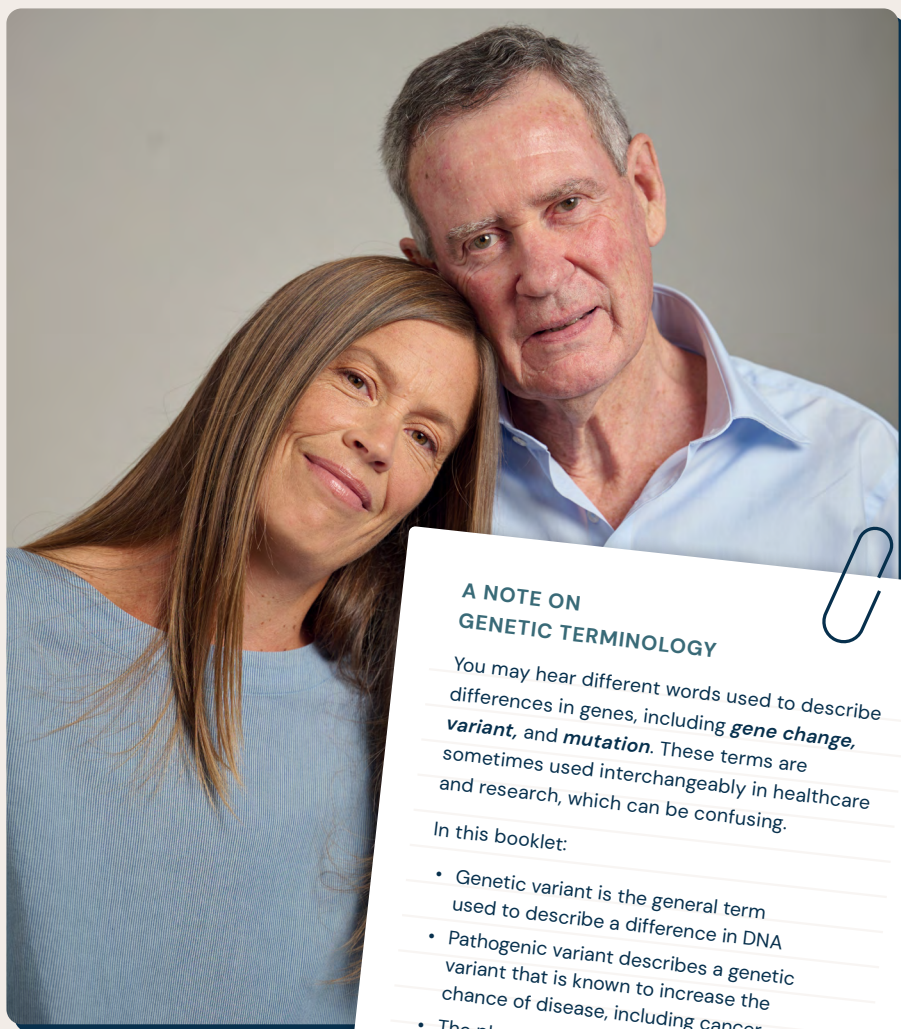
Only about 5–10% of all cancers are hereditary. Hereditary cancers occur when a person is born with a gene change that increases their chance of developing certain types of cancer. These variants can be passed from parent to child and may significantly increase cancer risk across generations.

Genetic testing looks for these inherited pathogenic gene variants that may explain why cancer developed and help inform care for both the individual and their family.

Like many young women, cancer wasn’t something I imagined facing so early in life. She knew there had been cancer in her family, a grandmother and an aunt but it wasn’t something that was openly discussed.”

Samira
BRCA2 pathogenic variant
Diagnosed with
breast cancer at 25





A NOTE ON GENETIC TERMINOLOGY

You may hear different words used to describe differences in genes, including **gene change**, **variant**, and **mutation**. These terms are sometimes used interchangeably in healthcare and research, which can be confusing.

In this booklet:

- Genetic variant is the general term used to describe a difference in DNA
- Pathogenic variant describes a genetic variant that is known to increase the chance of disease, including cancer
- The phrase gene change is sometimes used as a more everyday way of describing a genetic variant

The term **mutation** has historically been used to describe disease-causing genetic changes. However, it can sound alarming and is less precise than current clinical terminology, so we avoid using it here.



How do cancer genes work?

Our bodies have genes that help protect us from cancer by controlling how cells grow and repairing damage in our DNA. When changes occur in these genes, cells can grow or divide in an uncontrolled way, which can increase the chance of cancer developing.

What happens when these genes are disrupted?

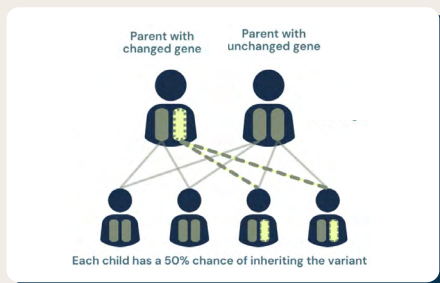
We inherit our genes from our parents, which means that if a gene carries a change in its DNA sequence that is associated with a health condition, we can see that health condition running in families. If a person inherits a pathogenic variant in one of the cancer protective genes mentioned above, the body may lose the ability to control cell growth, increasing the chance of cancer.

How is cancer predisposition inherited?

We inherit two copies of most of our genes: one from each parent. If one copy carries a pathogenic variant, a person may have an increased chance of developing cancer, even though they still have one unchanged gene.

It is important to note the following facts about inherited cancer predisposition:

- A person with an inherited variant doesn't always develop cancer, but their chance is higher.
- Genetic variants can be passed down from either the mother or the father, to both sons and daughters.
- Most inherited cancer syndromes follow an autosomal dominant pattern of inheritance. This means that having just one altered copy of the gene is enough to increase the chance of getting cancer.
- If a parent carries a cancer-related genetic variant, each of their children has a 50% chance of inheriting it.



Genetic testing for breast cancer: Multi-gene panels

For most people diagnosed with breast cancer, genetic testing is usually done using a multi-gene panel.

This means several genes are tested at the same time, rather than looking at just one gene. Germline testing focuses on genes where changes are known to increase the risk of developing cancer, and where the results may help guide treatment, screening, or family risk assessment.

COMMON GENES INCLUDED IN BREAST CANCER PANEL TESTING ARE:

- **BRCA1 and BRCA2:**
associated with breast, ovarian, uterine and prostate cancers
- **PALB2:**
linked with breast and pancreatic cancers
- **ATM, CHEK2, RAD51C/RAD51D:**
moderate-risk breast cancer genes
- Other genes, which may be included depending on a person's own cancer diagnosis or their family history.

Genetic testing for breast cancer: the process



INFORMED CONSENT AND PRE-TEST COUNSELLING

Before testing, you will have a discussion with a healthcare professional (e.g. oncologist, breast surgeon, genetic counsellor) who will explain the test, including the possible results, and what they may mean for you and your family. This helps you decide whether testing is right for you and ensures you can give informed consent.

SAMPLE COLLECTION

A small sample is collected, most commonly a blood sample, cheek swab or sometimes saliva. This sample contains DNA, which is the genetic material being tested.

LABORATORY TESTING

In the laboratory, scientists use advanced sequencing technology to examine many genes at the same time, looking for changes in the DNA that may increase cancer risk. You won't notice this step—it all happens behind the scenes.

TURNAROUND TIME, RESULTS AND POST-TEST COUNSELLING

Results are usually available within 2 to 8 weeks, depending on the type of test and the laboratory. Typically, genetic testing that is facilitated through private providers and laboratories has a faster turnaround time.

The results are then explained to you by a healthcare professional, who discusses what they mean for you and your family. This may involve referral to a clinical geneticist or genetic counsellor if you have not already met with one.

What are the different types of results from diagnostic genetic testing?

Genetic test results are usually reported in one of three main categories:

POSITIVE

A pathogenic variant was found that increases the risk of certain cancers. This result may help explain the cause of your cancer and can be used to guide your care, including treatment options, follow-up, and inform risk management options for family members.

**NO
ABNORMALITY
DETECTED
(NAD)**

No variants considered to explain or cause the condition being tested were identified. This does not always rule out a genetic cause, as not all genetic changes can currently be detected or fully understood, and other factors may still contribute to risk.

**VARIANT OF
UNCERTAIN
SIGNIFICANCE
(VUS)**

A genetic variant was found, but it is not currently known whether it affects health or increases risk. A VUS is typically not used to make medical decisions on its own and may be reclassified in the future as more evidence becomes available.

Your healthcare provider or genetic counsellor will explain what your result means for you and, if relevant, for your family.

What can diagnostic genetic testing tell you?

Diagnostic genetic testing can tell you:

- Whether an inherited genetic change explains your cancer diagnosis
- Whether you carry an inherited gene variant that increases the risk of you developing other types of cancer
- Whether certain treatments may be more effective for you
- Whether your family members may also be at increased risk
- Whether extra screening or preventive steps may be recommended

Diagnostic genetic testing cannot tell you:

- Whether you will develop another cancer in the future
- Exactly when a cancer might occur
- That cancer was caused only by genetics—many cancers have multiple causes
- About genetic changes outside the genes included in the test

How Genetic Testing Can Help Your Treatment and Ongoing Care

Understanding what the test can and cannot tell you can help you make informed decisions and prepare for the next steps. Genetic testing can provide important information that helps guide cancer treatment, future screening, and risk-reduction options. Your care team will use your results to support decisions that are right for you.



Genetic testing shaped every major decision I made."

Samira

BRCA2 pathogenic variant

Diagnosed with breast cancer at 25

Cancer Treatment Options

If you are diagnosed with cancer, treatment decisions can be influenced by inherited pathogenic variants in genes such as **BRCA1** or **BRCA2**.

GENETIC RESULTS CAN GUIDE:

- Targeted therapies (e.g. PARP inhibitors), which may be more effective for some people. PARP inhibitors work by blocking a backup repair system that cancer cells with a BRCA pathogenic variant rely on. This makes it harder for the cancer to survive, so these drugs can be more effective for people with BRCA changes.
- Surgical planning, including the type and timing of breast or ovarian cancer surgery. If a pathogenic gene variant (such as **BRCA1** or **BRCA2**) is identified, this may influence decisions about surgery for your current cancer and help address the risk of developing future breast or ovarian cancers. In some cases, this may lead your care team to discuss more extensive surgery, such as removing both breasts and ovaries, to reduce future cancer risk.
- Eligibility for clinical trials that require genetic information. Many research studies focus on treatments for people with BRCA or other inherited gene changes. A positive result may give you access to trials testing promising new therapies.

Your oncology team will explain whether your result is relevant to your current or future treatment.

Screening & Early Detection Options

If testing shows an inherited risk of breast and/or ovarian cancer, you may be offered:

- Earlier and more frequent breast screening than the general population
- Breast MRI, mammography at younger ages, and/or targeted breast ultrasound
- Coordinated follow-up through a familial cancer clinic or specialist team

These strategies aim to detect cancer as early as possible, when treatment is most effective.



I was told that having a *BRCA2* variant increased my risk of developing cancer again, and that treatment could become an ongoing reality. A double mastectomy was discussed as a longer-term option one that could reduce future risk and limit the need for repeated treatment”

Samira

BRCA2 pathogenic variant

Diagnosed with breast cancer at 25

Risk-Reduction Options

Depending on your level of risk and personal circumstances, options may include:

PREVENTIVE MEDICATIONS

- Some people may choose medications (often hormone-based treatments) that can lower the chance of developing breast cancer. Your doctor will discuss whether these are suitable for you based on your age, medical history, and type of risk.

RISK-REDUCING SURGERY, SUCH AS:

- Risk-reducing breast surgery (also called prophylactic or preventive mastectomy) – This surgery involves removing most breast tissue to significantly reduce the chance of developing breast cancer. It is called risk-reducing because the goal is to lower risk, not to treat an existing cancer. Breast reconstruction is often offered as an additional option and can be discussed with your surgical team.
- A genetic test result can sometimes help guide decisions about:
 - Whether breast-conserving surgery (lumpectomy) or mastectomy is more appropriate if breast cancer has already been diagnosed.
 - Whether surgery on one breast (unilateral) or both breasts (bilateral) may be worth considering, particularly if there is a higher risk of developing a new cancer in the other (contralateral) breast in the future.
 - Removal of the ovaries and fallopian tubes (bilateral salpingo oophorectomy) – This surgery reduces the risk of ovarian cancer and, in some situations, may also reduce breast cancer risk. It is usually considered after childbearing is complete and has important effects such as inducing menopause, so careful discussion with your doctor is essential.

These options are discussed carefully, considering your age, personal and family history, fertility considerations, and personal preferences.

WHAT THIS MEANS FOR YOU

- Care is individualised—not everyone will need or want the same options
- Decisions are made with you, supported by specialist advice
- The goal is to reduce cancer risk where possible and support early detection

Your healthcare provider or genetic counsellor can help you understand which options are relevant for you and guide next steps.

I did however feel like there was now a 'reason' for why I got cancer and it was nothing to do with anything I had done."

Rebecca

BRCA1 pathogenic variant

Diagnosed with triple negative breast cancer

Possible challenges associated with genetic testing

- Unexpected results (e.g. learning about family risk) – Genetic testing can sometimes reveal information about inherited cancer risk that affects other family members, which may come as a surprise or feel difficult to share.
- Anxiety or worry while waiting for results – The time between having the test and receiving results can be stressful, particularly when you are already coping with a cancer diagnosis or treatment decisions.
- Uncertainty (especially with VUS) – Sometimes results identify a variant of uncertain significance, meaning it is unclear whether the change is linked to cancer risk, which can feel frustrating or confusing.
- Impact on family relationships – Learning about shared genetic risk can raise emotional, practical, or communication challenges within families, as not everyone may feel the same about testing or sharing information.
- Information overload during cancer treatment – Receiving complex genetic information at the same time as a cancer diagnosis and treatment planning can feel overwhelming and difficult to process.

Practical considerations regarding genetic testing

Cost of genetic testing

From 1 January 2025, Medicare expanded eligibility for some genetic testing items (such as 73295 and 73296), meaning more people can now access genetic testing without out-of-pocket costs, particularly when results are needed to guide cancer treatment or clinical care.

Genetic testing may also be funded through public familial cancer clinics, where eligibility is based on personal and family history and assessment by a specialist team. Testing may also sometimes be offered as part of a research study, at no cost.

However, not all genetic tests are Medicare-funded, and in some situations, testing may not meet funding criteria. In this context, private genetic counselling consultation and testing is available but usually involves an out-of-pocket cost, which can vary depending on the clinic, test and laboratory.

Your healthcare provider or genetic counsellor can explain which testing options are appropriate for you, whether your test is Medicare-funded, provided through a public clinic, privately funded, or available through a research program, and help you understand any potential costs before you decide to proceed.

Life insurance

Current protections for people undergoing genetic testing.

Recent changes to Australian law are strengthening protections against the use of predictive genetic test results in life insurance underwriting. Once these changes are fully in effect, life insurers will not be permitted to use the results of predictive genetic testing when making decisions about new life insurance policies.

These changes are intended to remove insurance-related barriers to genetic testing and enable people to make decisions about testing based on their healthcare needs. Insurers may still ask about your personal medical history, diagnosed health conditions and other relevant risk factors, but not your predictive genetic test results.

As insurance arrangements can change over time, your genetic counsellor or genetics service can provide you with the most up-to-date information relevant to your circumstances before you decide whether to proceed with genetic testing.



Without my surgeon's insistence for testing, I wouldn't have known about the *BRCA1* pathogenic variant that ultimately changed the course of my treatment."

Rebecca
BRCA1 pathogenic variant
Diagnosed with triple negative breast cancer

My Health Record

The government's Better and Faster Access Program is changing how test results, including certain genetic test results appear in My Health Record.

Once fully rolled out in 2026, new genetic test results may be available in My Health Record for both patients and the healthcare professionals who order them, often sooner than before. These changes will not affect genetic tests already completed or past results.

Genetic results can be complex and sometimes unexpected, and may bring feelings of relief, worry or uncertainty, or raise questions for you and your family. Because genetic information can also have implications for your relatives, privacy, consent and appropriate support are important when results are shared digitally.

If you or your family are considering genetic testing, you can choose whether results are uploaded to My Health Record, based on what feels right for you. A genetic counsellor can help you understand these changes and support you in making an informed decision.

YOU HAVE THE RIGHT TO:

- Check and adjust your My Health Record privacy settings
- Ask your provider if results may be uploaded, and if so, when this will occur



Genetic testing gave me the data to make an informed decision and ultimately it also helped save other members of my family having to go through what I did."

Rebecca

BRCA1 pathogenic variant

Diagnosed with triple negative breast cancer

Why Genetic Testing Matters for Your Family

Some gene variants that increase the risk of cancer can be inherited, meaning they may be passed from parent to child.

If you are found to carry an inherited pathogenic variant, your biological relatives (such as parents, siblings, children, and sometimes extended family members) may also be at increased risk—even if they have never had cancer themselves.

In this situation, predictive genetic testing may be offered to family members to see whether they have inherited the same variant. Predictive testing can help relatives who carry the variant:

- Access appropriate screening earlier or more frequently than the general population
- Consider risk-reduction options where appropriate
- Make informed decisions about their health and future planning

Relatives who do not carry the variant can often be reassured and may avoid unnecessary screening or interventions.

In Australia, Medicare-funded genetic testing items are available for eligible family members when a known familial variant has been identified, meaning testing is often available without out-of-pocket cost. A genetic counsellor or specialist can explain who in the family may be eligible for testing and how this is arranged.

Learning about inherited risk can feel empowering for some families, while others may need time, support, or guidance to process the information and decide what to do next. Both responses are completely normal, and support is available to help individuals and families navigate these decisions.

Navigating family conversations

Conversations about predictive testing can be complex, but you don't have to do it all at once, or on your own:

- You can take time to consider what feels right to share, when to share it, and who you want to share it with.
- Genetic counsellors can support you in these conversations by helping you plan what to say and offering resources you can pass on to family members.
- If you're sharing with children or young adults, it doesn't have to be a single or complete explanation. These conversations can be gradual and ongoing, with support from genetic counsellors to help you find the right timing and language.



Telling my family was hard and trying to get them to understand the gravity of what this meant for them was challenging"

Rebecca

BRCA1 pathogenic variant

Diagnosed with triple negative breast cancer

Genetic test results and reproductive decision making

If you have a pathogenic variant associated with inherited cancer, and are planning or considering having children, support is available to help you understand your options and to make decisions.

A genetic counsellor can explain what your result means for your future children. They can talk through the chances of passing on the gene change and help you explore a range of reproductive options, which may include:

- Prenatal diagnostic testing: Testing during pregnancy to see whether the gene change has been inherited.
- Preimplantation genetic testing (PGT): Screening embryos created through IVF so only embryos without the gene change are used.
- Use of donor eggs or sperm: Using a donor who does not carry the gene change.
- Adoption or non-biological parenting options
- Choosing not to have biological children

A fertility specialist can provide more details about treatments such as IVF, egg or sperm preservation, and whether PGT is possible through their clinic. They can also discuss timing and any medical considerations related to cancer treatment and fertility.

You do not have to make these decisions alone. Your care team can connect you with genetic counsellors and fertility specialists who will support you in choosing what feels right for you and your family.

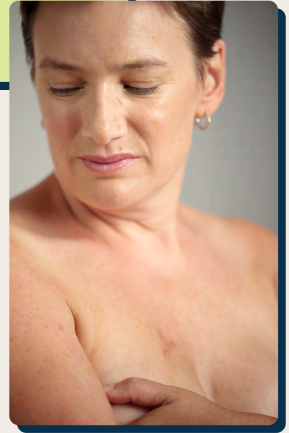


I believe everyone should be provided with the information to make their own informed choices."

Rebecca

BRCA1 pathogenic variant

Diagnosed with triple negative breast cancer



Support and Resources

Inherited Cancers Australia (ICA)

ICA offers a range of supportive care services to help individuals and families.

These include:

- A peer support program where you can connect with others who share similar experiences
- Genetic counselling free of charge, available via phone, Zoom or email.
- Private online support groups to ask questions, learn from others, and stay connected with the inherited cancers community.
- Online resource library for reliable information
- In-person community catch-ups and information and support days.
- Information and support navigating referral pathways ensure you can easily reach the right services when needed.

02 8084 2288

info@inheritedcancers.org.au

www.inheritedcancers.org.au

 **inherited**
CANCERS AUSTRALIA

Other National Resources

13YARN (13 92 76)

24/7 support line where Aboriginal or Torres Strait Islander Crisis Supporters are ready to listen and walk alongside you.

EVIQ PATIENT INFORMATION

contactus@eviq.org.au
<https://www.eviq.org.au/>

BEYOND BLUE

1300 22 46 36
24/7 Support

LIFELINE

13 11 14
24/7 crisis support

BREAST CANCER NETWORK AUSTRALIA

1800 500 258
<https://www.bcna.org.au/>

OVARIAN CANCER AUSTRALIA

1300 660 334
<https://www.ovariancancer.net.au/>

CANCER COUNCIL AUSTRALIA

13 11 20
<https://www.cancer.org.au/>

PANCARE FOUNDATION

1300 881 698
<https://www.pancare.org.au/>

CENTRE FOR GENETICS EDUCATION

0459 930 589
<https://www.genetics.edu.au/>

PROSTATE CANCER FOUNDATION OF AUSTRALIA

1800 22 00 99
<https://www.pcfa.org.au/>

Glossary

Core genetics terms

DNA

The chemical code inside our cells that carries genetic information.

GENE

A section of DNA that provides instructions for how the body grows and functions.

INHERITED (GERMLINE)

A genetic change present from birth and found in every cell of the body.

PATHOGENIC VARIANT

A genetic variant known to increase the chance of disease (including cancer).
Formerly called a mutation.

SOMATIC (TUMOUR)

Genetic changes found only in cancer cells & not passed on to children.

VARIANT

A change in a gene. Many variants are harmless.

Cancer and risk concepts

AUTOSOMAL DOMINANT INHERITANCE

A pattern of inheritance in which inheriting one altered copy of a gene is enough to increase cancer risk. Each child of a parent with an inherited cancer gene has a one-in-two or 50% chance of inheriting it.

CANCER PREDISPOSITION

An increased chance of developing cancer due to inherited genetic factors.

FAMILIAL CANCER

Cancer that occurs more often in a family, likely due to shared genes and/or environment, without a single inherited gene cause.

HEREDITARY CANCER

Cancer caused by an inherited gene change that increases cancer risk.

SPORADIC CANCER

Cancer that occurs by chance and is not inherited.

Genetic testing terms

DIAGNOSTIC GENETIC TESTING

Testing done after a cancer diagnosis to help guide treatment and future care.

GENETIC TESTING

A test that looks for changes in genes that may affect health or cancer risk.

INFORMED CONSENT

Deciding about testing after understanding the benefits, risks, and limitations.

MULTI-GENE PANEL

A test that looks at several genes at the same time.

PREDICTIVE GENETIC TESTING

Testing for family members to see if they carry a known inherited gene change.

Genetic test results

NO ABNORMALITY DETECTED (NAD)

No relevant gene changes were found, though this does not remove all cancer risk.

POSITIVE RESULT

A gene change was found that increases cancer risk.

VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)

A gene change where it is not yet known whether it affects cancer risk

Glossary

Management and care

BILATERAL MASTECTOMY

Surgery to remove both breasts. This may be done to treat cancer in both breasts, or as a risk-reducing (preventive) option for people at high inherited risk of developing breast cancer. Some people choose to have breast reconstruction at the same time or later.

BILATERAL SALPINGO OOPHRECTOMY

Surgery to remove both ovaries and both fallopian tubes. This is done to reduce the risk of ovarian cancer and, in some situations, may also lower breast cancer risk. It causes menopause if it has not already occurred

FAMILIAL CANCER CLINIC

A specialist service that supports people and families with inherited cancer risk.

LUMPECTOMY

Surgery to remove a breast cancer along with a small amount of surrounding healthy tissue, while keeping most of the breast. This is often followed by radiation therapy. It is also called breast-conserving surgery.

MASTECTOMY

Surgery to remove breast tissue. It may be done to treat breast cancer or to greatly reduce the chance of developing breast cancer in the future (risk-reducing or prophylactic mastectomy).

RISK-REDUCING SURGERY

Surgery to lower the chance of developing cancer.

SURVEILLANCE

Regular screening to detect cancer early.

TARGETED THERAPY

Treatment designed to work against cancers with specific genetic features (e.g. PARP inhibitors).

UNILATERAL MASTECTOMY

Surgery to remove one breast. This may be done when cancer is present in one breast or, less commonly, as a risk-reducing option for one side.

Reproductive and family planning

GAMETE DONOR

Use of donated sperm or eggs from someone who does not carry the gene change.

PREIMPLANTATION GENETIC TESTING (PGT)

Testing embryos created through IVF to select embryos without a specific gene change.

PRENATAL TESTING

Testing during pregnancy to see if a genetic condition has been inherited.

Other

GENETIC COUNSELLOR

A health professional with expertise in genetics and counselling, who explains genetic information, discusses the benefits and limitations of testing, and helps you understand what results may mean for your health and your family. They also provide emotional support, help you communicate information with relatives if needed, and work with your healthcare team to support you in making informed decisions that feel right for you.

MY HEALTH RECORD

Australia's secure digital health record system.

This booklet was produced with support from AstraZeneca.
The content was independently developed by Inherited Cancers
Australia without influence from AstraZeneca

Notes

Notes

Notes

Notes



02 8084 2288

info@inheritedcancers.org.au

inheritedcancers.org.au