BRCA1 Gene Variant Support Guide

Your guide for clarity, support, and next steps.

How This Guide Can Help

This guide is here to inform and support you. Below is a quick overview of what's included, so you can find the information that's most helpful to you right now.

- **Understanding Inherited Cancers**
  - A simple explanation of genes and genetic changes, and how they relate to cancer.
- **About the *BRCA1* Gene Variant**
  - Helpful information about *BRCA1* and what it might mean for your and your family's health.
- **What You Can Do Next**
  - Options to consider, including screening, lifestyle changes, and medical care.
- **Support for Family and Future Planning**
  - Overview on cascade testing, family conversations, and reproductive options.
- **Where to Find Support**
  - People and services that can help, including healthcare professionals and ICA programs and resources.

What are inherited cancers?

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 caused by shared genes, lifestyle, or environmental factors, but without a single identifiable inherited genetic cause.

Only about 5–10% of all cancers are inherited. Inherited cancers occur when a person is born with a pathogenic gene variant. These variants can be passed from parent to child, which may significantly increase the chance of developing certain types of cancer.

Key Genetics Terms Made Simple

- **Genes:** Instructions inside our cells that tell the body how to grow and function.
- **DNA:** The code that makes up our genes.
- **Variant:** A change in a gene. Some are harmless, but others can raise the chance of disease.
- **Pathogenic Variant (formerly called "mutation"):** A gene change known to increase the chance of disease, including cancer.

Your genes are part of your DNA

Genes carry instructions for your body

Variants are differences in DNA from birth

Pathogenic variants disrupt genes which can lead to disease

How do cancer genes work?

Our bodies contain a large number of special genes that help protect us from cancer. Examples include *BRCA1*, *BRCA2*, *PALB2, ATM* and *TP53*. These genes fall into different categories, each playing a unique role:

- **Tumour Suppressor Genes**: Act like brakes, slowing down cell growth or fixing damaged DNA.
- **DNA Repair Genes**: Help fix mistakes in our genetic code.
- **Oncogenes**: Normally help cells grow, but when altered, they can cause cells to grow out of control.

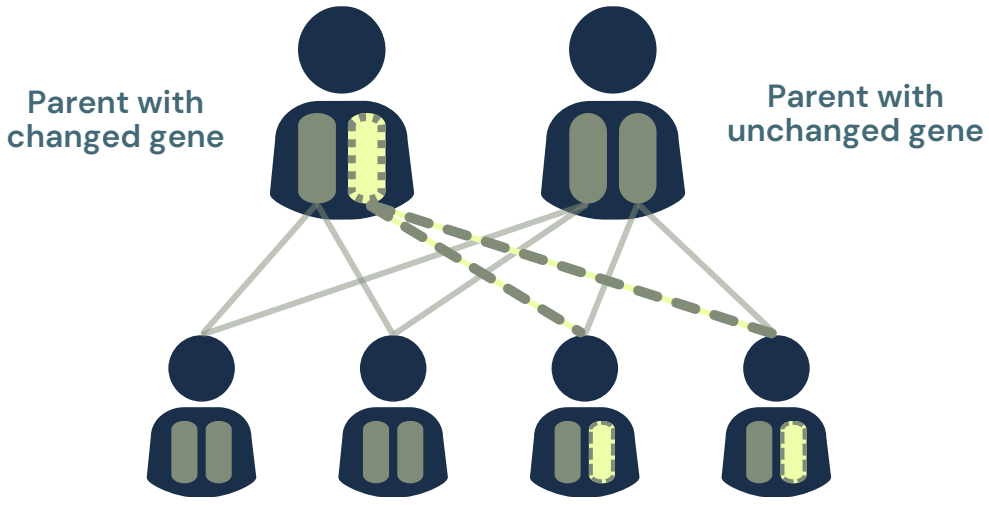
What happens when these genes are disrupted?

If a person inherits a **pathogenic variant in one of these protective genes**, 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 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.

- **A person with an inherited variant doesn't always develop cancer**, but their chance is higher.
- These changes 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**.
- Each child of a parent who has a cancer variant has a **50% chance of inheriting it**.



Each child has a 50% chance of inheriting the variant

How common is it to be *BRCA1* positive?

- Around 1 in 500 people has a *BRCA1* gene variant [1, 2]
- Around 1 in 40 people of Ashkenazi Jewish heritage has a *BRCA1* or *BRCA2* gene variant[3], making it more common in this group than in the general population.

References:

1. Hereditary Cancer Syndromes: A Comprehensive Review with a Visual Tool
2. eviQ: Cancer predisposition genes: population carrier frequency
3. The risk of cancer associated with specific mutations of BRCA1 and BRCA2 among Ashkenazi Jews

What does having a *BRCA1* gene variant mean for me?

Having a *BRCA1* gene variant increases the chance of developing certain cancers.

For people assigned female at birth, this includes a higher chance of **breast** and **ovarian cancer**, and possibly a small increase in the chance of **pancreatic** or **uterine cancer**.

For people assigned male at birth, it increases the chance of male **breast** and **prostate cancer**, and may also slightly raise the chance of **pancreatic cancer**.

This doesn't mean you will definitely get cancer. It means you now have information that can help you plan ahead, like doing regular screening or learning about ways to reduce your chance of developing cancer. Support is available to help you take it one step at a time.

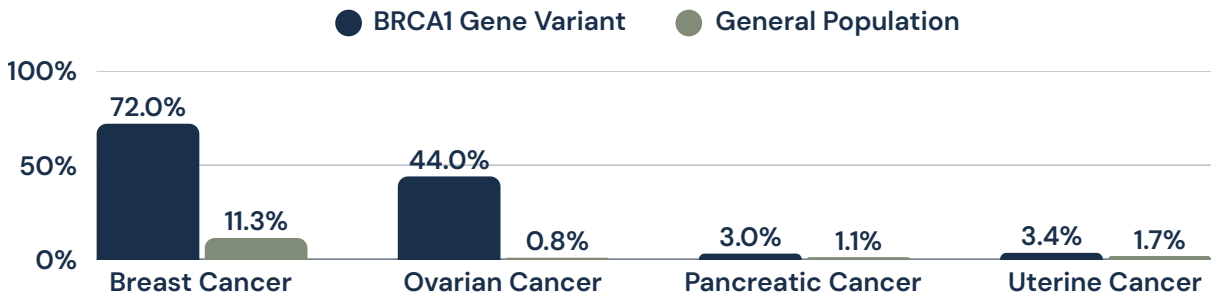
Understanding your chance of developing cancer is a powerful first step. Support is available to help you move forward at your own pace.

Find Support

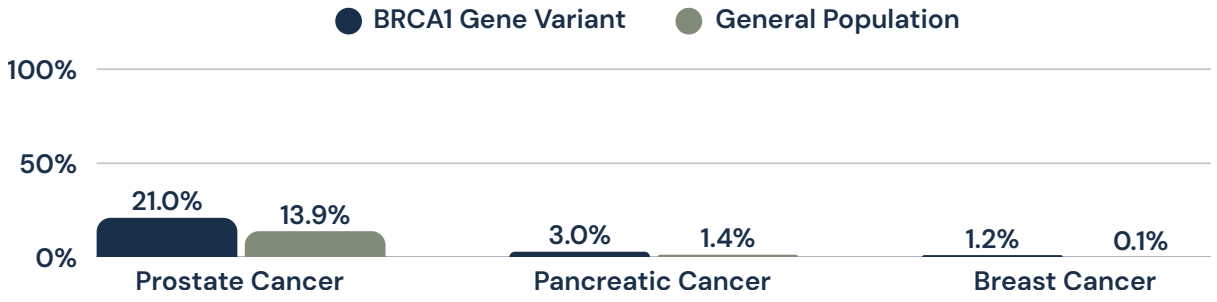
What is the lifetime chance of developing cancer if I have a *BRCA1* gene variant?

The graphs below show the **estimated lifetime chance by cancer type** for people with a *BRCA1* gene variant compared to the general population. These numbers are based on research and are here to **help you understand your chance of developing cancer** and **start thinking about next steps with your doctor or healthcare team**.

♀ For People Assigned Female at Birth



♂ For People Assigned Male at Birth



Remember, an increased chance doesn't mean you will definitely get cancer. Understanding your chance of developing cancer is an important first step. On the next page, you'll find general guidance on what to discuss with your GP or specialist to help you plan your next steps.

Our resource hub includes tips, checklists, and appointment questions to guide your next steps, and to help you support others in your family.

Browse Resources

♀ For People Assigned Female at Birth

If you were assigned female at birth and have a *BRCA1* gene variant, discuss the following risk management options with your GP or specialist:

- **Breast cancer**
  - High-risk breast screening, starting from age 25–30
    - Annual breast MRI (If MRI is not available, contrast-enhanced mammography (CEM) may be considered as an alternative.)
    - Mammogram every year starting from age 40
  - Risk-reducing mastectomy
  - Risk-reducing medication
- **Ovarian/fallopian tube cancer**
  - Risk-reducing bilateral salpingo oophorectomy (RRSO, removal of both ovaries and fallopian tubes) is recommended between ages 35–40, after having children if that's part of your plan.
  - Screening with ultrasound or blood tests (CA125) is not effective.
- **Pancreatic cancer**
  - There's limited but emerging evidence that surveillance might improve outcomes from age 50. If available, surveillance should ideally be done as part of a clinical trial.
  - In Australia, **clinical trials like APRISE** may be available for people with a *BRCA1* gene variant who also have a first- or second-degree relative with pancreatic cancer.
  - These trials may involve regular surveillance using imaging tests such as MRI/MRCP or endoscopic ultrasound (EUS).
- **Uterine cancer**
  - A hysterectomy is not routinely recommended.
  - Some people may consider having a hysterectomy at the same time as RRSO to allow the use of oestrogen-only menopausal hormone therapy (MHT), which may help lower breast cancer risk.
- **General health**
  - Maintaining a healthy lifestyle (avoiding smoking, limiting alcohol, staying a healthy weight) may help reduce risk.

You can explore the **full clinical guidelines at eviQ** for the latest recommendations on cancer risk management.

♂ For People Assigned Male at Birth

If you were assigned male at birth and have a *BRCA1* gene variant, discuss the following risk management options with your GP or specialist:

- **Prostate cancer**
  - Consider annual PSA blood tests (± digital rectal exam) from around age 40.
- **Male breast cancer**
  - No routine screening, but be chest aware and report any changes to your GP.
- **Pancreatic cancer**
  - There's limited but emerging evidence that surveillance might improve outcomes from age 50. If available, surveillance should ideally be done as part of a clinical trial.
  - In Australia, clinical trials like APRISE may be available for people with a *BRCA1* gene variant who also have a first- or second-degree relative with pancreatic cancer.
  - These trials may involve regular surveillance using imaging tests such as MRI/MRCP or endoscopic ultrasound (EUS).
- **General health**
  - Maintaining a healthy lifestyle (avoiding smoking, limiting alcohol, staying a healthy weight) may help reduce risk

You can explore the **full clinical guidelines at eviQ** for the latest recommendations on cancer risk management.

Visit our resource hub for questions to ask at appointments, checklists, tips, and more to help you prepare for these conversations.

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What does this mean for my family?

A gene variant can be passed down through generations, which means your genetic testing result may also have important meaning for your family.

Sharing your result with your family can give them the chance to understand their own risk. If they choose to, they can take steps early, including genetic testing, to manage it, as inherited cancer can affect anyone, regardless of gender or age.

This process is known as **predictive** or **cascade genetic testing**: where close family members may be offered genetic testing to see if they also have the same variant.

Predictive / Cascade Genetic Testing

- It's called *cascade* testing because it **creates a flow of genetic information through the family**.
- One person's genetic result can open the door for others to understand their own risk and take action early, if they choose to.
- Cascade genetic testing **may be Medicare-funded** for eligible relatives.

How do I navigate these family conversations?

Conversations about cascade 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.

Our conversation guides and templates can support you in sharing information with your family, in a way and at a time that feels right.

Browse Resources

Support for Reproductive Decision-Making

If you're considering or planning to have children, support is available to help you understand how a gene variant may relate to family planning and reproductive decisions.

- A **genetic counsellor** can talk through what having a gene variant means for your potential future children and help you explore your options.
- Genetic counsellor can discuss reproductive options with you, which might include **prenatal diagnostic testing of a naturally conceived pregnancy**, or **preimplantation genetic testing (PGT) of embryos created through IVF**.
- Fertility specialists can provide more information about options such as **IVF** or **egg preservation**, and whether **PGT** is possible through their clinic.

Explore our fertility resources to learn about the options available and find support for decisions that feel right for you.

Browse Resources

Who might be involved in my care?

Understanding and managing an inherited gene variant often involves various health professionals across different stages. This list is here to show the range of support available and may help you recognise what kind of care could be helpful as your needs evolve. Your GP is usually the first step and can guide referrals based on your needs. Visit our **Resource Centre** for helpful questions to ask at your next appointment.

Genetic & Specialist Care

- Genetic counsellor
- Clinical geneticist
- Fertility specialist
- Menopause specialist / clinic

Surgery & Screening

- High-risk screening clinic
- Breast surgeon
- Plastic surgeon
- Gynaecological surgeon
- Breast care nurse

Mental Health Support

- GP (for referrals & care planning)
- Psychologist / counsellor
- Psycho-oncologist
- Relationship or family counsellor

Wellbeing Support

- Physiotherapist
- Exercise physiologist
- Dietitian
- Social worker

How can Inherited Cancers Australia help?

Discovering a *BRCA1* gene variant can bring up a lot of thoughts and questions. You might be figuring out what it means for your health, your family, or your next steps. You don't have to work through it alone.

Inherited Cancers Australia offers personalised support grounded in lived experience. Whether you're looking for information, guidance, connection, or a community that understands, we're here to support you in the way that feels right for you.

Inherited Cancer Support Service

Talk to our genetic counsellor who can help answer questions and guide your options.

Get Support

National Online Support Groups

Join a safe space to connect with people who are navigating a similar experience.

Join Community

Peer Support Program

Connect with a trained community member for support, understanding, and reassurance from someone who's been there.

Connect Now

Info & Support Events

Join nationwide events that bring together local experts and community voices to help you stay informed, connected, and supported.

Upcoming Events

Online Resource Centre

Explore evidence-based, expert-reviewed resources to help you feel more prepared and informed about your next steps.

Browse Resources

Psychosocial Support

Access helpful tools and resources that support your emotional and mental wellbeing while living with an inherited gene variant.

Find Support Tools

Visit our website for more information: **inheritedcancers.org.au**