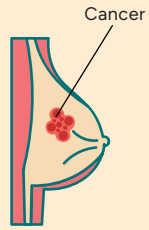
GERMLINE GENETIC TESTING FOR HEREDITARY BREAST AND OVARIAN CANCERS

ACE agency for care effectiveness

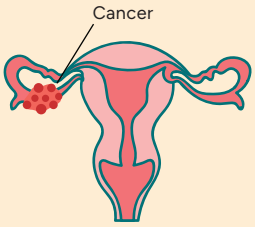
WHAT ARE HEREDITARY BREAST AND OVARIAN CANCERS?

Hereditary breast and ovarian cancers account for around **1 in 10** breast cancers and **1 in 7** ovarian cancers.[1] They are cancers that **run in families** due to changes in certain genes that are passed down through generations (**inherited**). These inherited gene changes can increase a person's chance of developing breast or ovarian cancer.



Breast cancer

Breast cancer occurs when abnormal cells grow uncontrollably in the ducts or lobules of the breast and spread into the surrounding breast tissue. It is the **most common** cancer diagnosed in females in Singapore.[2]



Ovarian cancer

Ovarian cancer occurs when abnormal cells grow uncontrollably in the ovaries or fallopian tubes and spread into the surrounding tissue. It is the **sixth most common** cancer diagnosed in females in Singapore.[2]

Certain genes in our bodies are responsible for controlling cell growth and repairing damage to keep cells healthy. When these genes develop harmful changes (also known as **genetic mutations** or **pathogenic variants**), the body may be less able to protect against cancer.



Mutations in the *BRCA1* and *BRCA2* genes are the **most common** cause of hereditary breast and ovarian cancers. However, mutations in other genes can also increase risk including *ATM*, *CHEK2*, *PALB2*, *RAD51C* and *RAD51D*, among others.

DID YOU KNOW?[3,4]

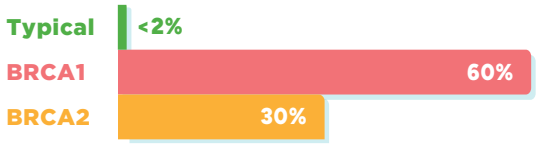
Breast cancer risk

Women with a *BRCA1* or *BRCA2* mutation have a more than **60% lifetime** risk of developing breast cancer, compared with about 13% in the general population. Men with these mutations are also at a higher risk of developing breast cancer.



Ovarian cancer risk

Women with a *BRCA1* mutation have up to a **60% lifetime** risk of developing ovarian cancer, while women with a *BRCA2* mutation have up to a **30% lifetime** risk. This is much higher than the general population risk of less than 2%.



HOW CAN YOU KNOW IF YOU ARE AT RISK?

Germline genetic testing[4,5]

A blood sample is used to look for gene mutations that you inherited (germline genetic testing). It is usually a **one-time** test. The genes tested depend on your personal and family history of cancer. Your doctor or genetic counsellor will advise which test is appropriate for you.

Genetic counselling[6]

Before having germline genetic testing, you will receive **genetic counselling**.
A healthcare professional will explain what the test involves, what the possible results could mean, and how the results may affect you and your family. Genetic counselling can help you understand your options and make an informed decision about testing, treatment and family planning.



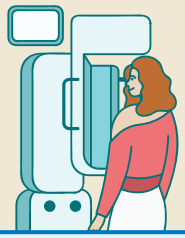
How testing can help[6]

Knowing your genetic status can help explain a personal or family history of cancer and identify family members who may also be at risk. If you have cancer, your result may help guide treatment and surgery choices, and inform you of other cancers you may be at risk of developing. If you do not have cancer, your result may help guide screening, risk-reducing options, and reproductive choices if you are planning to have children.

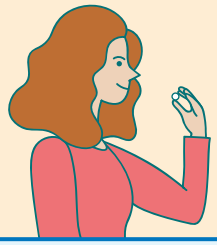
MANAGING THE RISK OF HEREDITARY BREAST AND OVARIAN CANCERS

Knowing your risk early can help you take steps to reduce your chance of developing cancer or detect cancer at an earlier stage, when treatment may be more effective. Regular medical follow-up is also **important** so you can review your risk and monitor your condition over time. If you are found to be at increased risk, there are several ways to help reduce or manage your risk of developing these cancers:

EARLY DETECTION	RISK-REDUCING OPTIONS		LIFESTYLE CHANGES
Closer monitoring through regular screening tests (**Early and frequent surveillance**)	**Risk-reducing surgery**	Medicines to lower risk (**chemoprevention**)	Exercising regularly Eating healthily Quitting smoking (if you smoke) Drinking less alcohol







More about risk-reducing surgery

Risk-reducing surgery is an operation that can **significantly** reduce the risk of cancer developing. It does not remove the risk completely, but it may be an effective option for some people at high risk of hereditary breast or ovarian cancer.

Removing **breast** tissue from one or both breasts (**risk-reducing mastectomy**) may be done with or without breast reconstruction. This can reduce the risk of developing breast cancer by up to **95%**.[7]

Removing both **ovaries and fallopian tubes** (**risk-reducing bilateral salpingo-oophorectomy**) can reduce the risk of developing ovarian cancer by up to **95%**.[8]

Surgery is a big decision and may not be suitable for everyone. Your doctor will consider your gene mutation, personal and family history, age, general health and plans for having a family. You may also need to consider other issues, such as breast reconstruction, early menopause, fertility, body image, and the possible need for additional surgeries, treatment or follow-up care.

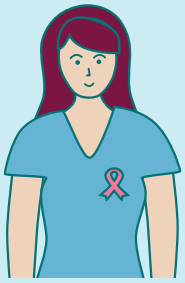
Speak with your **doctor** and **genetic counsellor** so you can understand the benefits, risks and timing of each option before making a decision.



SUBSIDY INFORMATION[4]

Germline genetic testing and some risk-reducing interventions are subsidised for eligible people. You may be eligible depending on your clinical assessment, and your personal and family history.

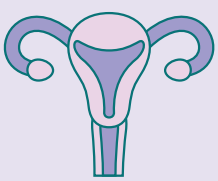
Subsidised germline genetic testing is available for certain people who belong to any of the following groups. You can speak with your doctor about whether testing is suitable for you or your family.



1 Group 1

You have been diagnosed with breast cancer

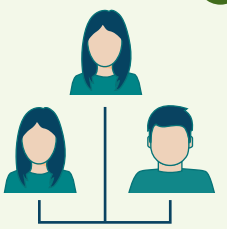
- Diagnosed at age 50 or younger
- Have certain types of breast cancer
- Have had more than one breast cancer
- Relevant family history (such as a close biological relative with breast, ovarian, pancreatic, or certain prostate cancers)
- Your doctor has identified relevant tumour test results, or has assessed that you have a high likelihood of carrying a *BRCA1* or *BRCA2* pathogenic variant.



2 Group 2

You have been diagnosed with ovarian, fallopian tube, or peritoneal cancer

Your doctor has identified relevant tumour test results, or has assessed that you have a high likelihood of carrying a relevant pathogenic variant.



3 Group 3

You have not been diagnosed with cancer but have a significant family history

You are a first-degree relative of someone with breast or ovarian cancer who is unavailable for genetic testing and your doctor has assessed that you have a high likelihood of carrying a relevant pathogenic variant based on your personal and family history.



4 Group 4

A pathogenic variant has already been identified in your family

You may be eligible for targeted testing for that specific variant if you are a first-degree relative of someone whose genetic test found a pathogenic variant that causes hereditary breast and ovarian cancers.

First-degree relatives share about 50% of your genes and include your **biological** parents, siblings (brothers and sisters) and children.

Subsidised risk-reducing surgeries

Subsidised risk-reducing surgeries are available for **eligible** people with a confirmed pathogenic or likely pathogenic variant in a relevant gene.

Risk-reducing mastectomy with or without breast reconstruction may be considered for people aged 25 years and older who carry an eligible pathogenic or likely pathogenic variant. To qualify, you must have **received genetic counselling**, and **pre-operative counselling** about the psychological, social, and sexual consequences of the surgery, and are assessed to be in reasonably good health by your doctor.

Risk-reducing bilateral salpingo-oophorectomy may be considered for women aged 35 years and older, or earlier if there is a family history of earlier-onset ovarian cancer, who carry an eligible pathogenic or likely pathogenic variant. To qualify, you must have **completed** family planning, **received genetic counselling**, and **pre-operative counselling** about the psychological, social, and sexual consequences of the surgery, and are assessed to be in reasonably good health by your doctor.

Both surgeries should only be considered after your doctor has assessed your personal risk, family history and individual circumstances.



KEY MESSAGES

Identifying hereditary breast and ovarian cancer early is crucial to **prevent** cancer or **catch it at a curable stage**. There are subsidised options available to help you manage your risk. Discuss with your doctor if testing is suitable for you or your family, and to find out which management or treatment options are most suitable for your situation.

Sources

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