Cancer Council Victoria

FactSheet

Understanding genetic testing for breast cancer

Genetic testing is offered to families at high risk of breast and/or ovarian cancer. These families have a family history of cancer that may be due to an inherited gene change.

This type of genetic testing begins with taking a blood sample from the family member who has had cancer, as they are the most likely person in the family to carry a gene change if it is present.

In Victoria, genetic testing is only offered through specialist Family Cancer Centres.

What are genes?

Genes are tiny and complex pieces of information that occur in each cell. They tell our bodies how to grow, function and develop. Each person has approximately 20,000 to 30,000 genes, which are in pairs, as we inherit one copy from each parent.

What genes can we test for?

Two **genes** are known to predispose to breast and ovarian cancer in some families. They are **BRCA1** and **BRCA2**, which stand for BReast CAncer gene 1 and BReast CAncer gene 2.

BRCA1 and BRCA2 are tumour suppressor genes. This means that they play a part in preventing cancer, particularly in the breast and ovaries.

What is genetic testing?

Genetic testing looks for changes (also called 'mutations') in a gene, which may stop the gene from working properly.

Looking for a gene change for the first time in a family is known as 'mutation detection'. It's a bit like looking through a huge book for a single spelling error. Results can therefore take some months.

What are the outcomes of genetic testing?

There are three possible outcomes:

Gene change (mutation) found

- This confirms that for the person tested there is an inherited or genetic cause for their cancer. It confirms that some family members will be at **high risk** of certain cancers; in particular breast and ovarian cancer for women and breast and sometimes prostate cancer for men.
- It shows that the person tested may be at increased risk of developing a new primary cancer and they may need to think about strategies that will manage their risk.
- Testing now becomes available to other family members who may not have developed cancer (predictive testing).

Gene change (mutation) not found

- This test result is considered uninformative.
- The result may be due to the limitations of current testing methods, which are unable to identify all possible gene changes. Or, there may be other genetic causes that are not yet known.
- The family is still considered to be at high risk for cancer. However, testing is not available to family members as it is not known what to test for.

Variant found

- This result is considered uninformative. A gene change has been found, but its relation to increased cancer risk is not established. In this situation, it is not possible to offer testing to other family members.

As more is discovered about cancer genetics, further tests may become available for families where a mutation is not found or a variant is found.

What is predictive testing?

When a gene change is found in a family member who has had cancer, it is possible to offer genetic testing to other family members, whether or not they have had cancer. This can show if they are at an increased risk of cancer or are simply at **average risk**. This is known as 'predictive testing'.

The result of the test is always conclusive. Results take between four to eight weeks.

There are two possible outcomes of predictive testing:

- A *positive* result indicates that the change in the gene has been inherited, and that the person tested is at high risk of breast, ovarian and associated cancers. They can start to think about strategies that will manage their risk. Not everyone with a positive result will develop cancer. It means that person is at higher risk.
- A negative result indicates that they have not inherited the change in the gene known of in the family. This means they have an average risk of the cancers associated with the gene change.

Genetic testing is usually only offered once someone is of adult age as there is no risk of childhood cancers associated with BRCA1 or BRCA2.

There are lots of factors to consider when making a decision about genetic testing.

One such factor is that of income protection and life insurance applications, which genetic testing may influence.

Ask your **Family Cancer Centre** for more information.

What that word means

associated with This expression is used in science to mean that there is scientific evidence to show that in the presence of one factor (for example, a change in a certain gene), another factor (for example, breast cancer) is more likely to occur.

average risk This means that someone's chance of developing a disease is no higher or lower than average. Also known as population risk.

BRCA1 & BRCA2 Genes that appear to protect against breast and ovarian cancer. If there is a change in one of these genes, breast or ovarian cancer is more likely to occur.

Family Cancer Centres Centres where people can get information about a family cancer history. Their services include genetic counselling, testing, medical advice, psychological support and information about research.

family history A careful assessment by a Family Cancer Centre of cancer occurrences in a family.

gene change A change somewhere in a gene. A change may be inherited or be caused by an error while a cell is reproducing itself, by factors such as some chemicals or viruses, or by events that science is yet to discover. A change in a gene may lead to disease such as cancer. However, people with a change in a gene that may predispose to cancer don't always get cancer. Also known as a gene error or gene mutation.

genetic testing Testing for gene changes which may explain why a disease has occurred or whether a disease is more likely to occur.

high risk This means that someone's chance of developing a disease in the future is higher than average, due to a family history of the disease and/or a change in a gene known to predispose to that disease. People assessed as at high risk are advised to consider strategies that could reduce their risk.

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