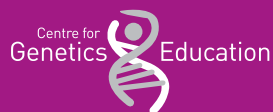




**Understanding genetic tests for
breast and ovarian cancer that runs in the family**
Information and decision aid



Health



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**Understanding genetic tests for
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Who is this booklet for?

This booklet contains information for women who are considering genetic testing for breast and ovarian cancer that runs in their family.

This information is not a replacement for discussing genetic testing with your doctor or family cancer services.

Choose the sections that are relevant to you, coupled with information from your health professionals, to make an informed decision that is right for you.

Genetic testing might be suitable if your family includes one or more of the following:

- A genetics specialist has suggested genetic testing might be helpful in your situation
- One of your blood relatives has received a genetic test that shows they have a faulty cancer protection gene
- Three relatives on the same side of the family in at least two generations have been diagnosed with breast or ovarian cancer
- Two relatives on the same side of the family in two or more generations have breast or ovarian cancer, one of whom has been diagnosed with at least one of the following:
 - breast cancer before 40 years of age
 - breast cancer in both breasts
 - both breast and ovarian cancer
 - breast cancer in a male relative
- The family has Ashkenazi Jewish ancestry.

These checkboxes are only a guide. It is recommended that you discuss your family's suitability for genetic testing with a genetics specialist.

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About breast and ovarian cancer that runs in the family

Cancer is common in Australia with many families having one or more members who have had breast or ovarian cancer.

In some families, these cancers occur in a number of generations. This is called a family history of cancer.

Why do I have a family history of cancer?

There are three reasons why you might have a family history of cancer:

Just by chance Breast cancer is a common disease. This means that many people have several people in their family who have developed breast cancer by coincidence.

Environmental factors Many families live in similar environments, so they often share similar diets and lifestyles. These non-inherited factors may influence the chance of developing cancer.

Inherited factors An inherited factor is the genetic information we receive from our parents and pass on to our children. In some families inherited genes play a role in the development of breast and ovarian cancer.

Cancer and our genes

Our genetic information provides instructions for growth and development and is in the form of many thousands of different genes including cancer protection genes.

Some cancers occur when cells grow out of control in a particular part of the body and a lump might be the first sign of cancer.

Cancer protection genes stop cells from growing out of control.

Variations in genes are common and can:

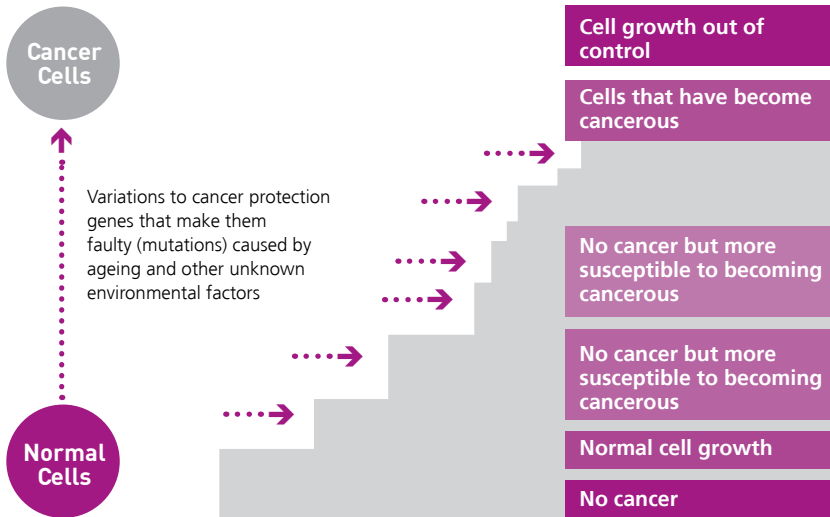
- occur when genes are copied for the growing of new cells and tissues
- be caused by environmental factors
- happen as we age
- be inherited from our parents.

Most variations do not cause problems for growth and development.

However, occasionally, variations in cancer protection genes cause faults that could lead to cancer. Faults in genes are called mutations.

As we age the variations in our genes increase. This is why most cancers develop later in life.


In the diagram below, each arrow is a new variation marking the cancer protection gene which becomes faulty over a period of time. The cell becomes cancerous when too many of the cancer protection genes become faulty.




It takes a long time for variations to build up in different cancer protection genes and for cancer to develop. In fact, variations may never build up enough for cancer to develop.

Breast cancer that is caused by faulty cancer protection genes

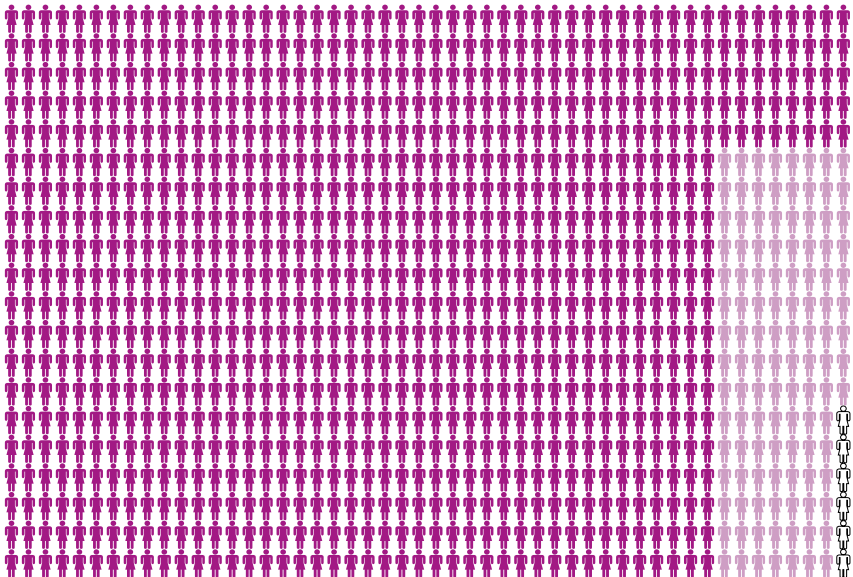
The diagram below shows that of 1000 Australian women, about 120 (12%) will develop breast cancer some time in their life. Most of these women will develop cancer after turning 60 years old.¹

 About 6 of the 120 women (5%) will have a strong family health history of breast cancer, which suggests that there is a faulty cancer protection gene specific to the family.

Key:


 880 will not develop breast cancer at any time

 120 will develop breast cancer sometime in their lifetime





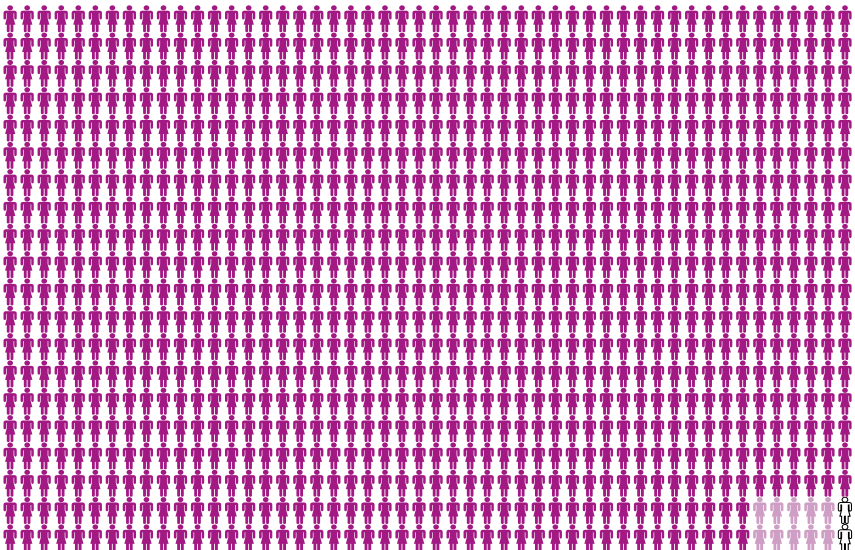
Ovarian cancer that is caused by faulty cancer protection genes

Ovarian cancer is less common among Australian women than breast cancer. The diagram below shows that of 1000 Australian women, about 12 (1.2%) will develop ovarian cancer some time in their life.

 About 2 of these 12 women (17%) will have developed the cancer mostly because they have inherited a faulty cancer protection gene. The average age that most women develop ovarian cancer is around the age 63 years.¹

Key:

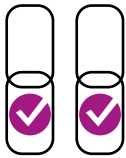
-  988 will not develop ovarian cancer at any time
-  12 will develop ovarian cancer sometime in their lifetime



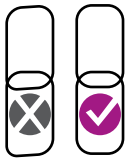
Inheriting faulty breast and ovarian cancer protection genes

Currently we know of a number of cancer protection genes that, when working properly, prevent the development of breast, ovarian, and other types of cancer.

Two important protection genes for breast and ovarian cancer are called BRCA1 (BReast CAncer 1) and BRCA2 (BReast CAncer 2). Everyone is born with two copies of a BRCA1 protection gene and two copies of a BRCA2 protection gene. A small number of people are born with a faulty copy of one of these genes.



Most people start life with **two working copies** of their BRCA1 and BRCA2 cancer protection genes in each cell of their body. One copy is inherited from their mother and one from their father.



Rarely, people might inherit **one faulty copy** of a BRCA1 or BRCA2 cancer protection gene from either their mother or father. The working copy provides cancer protection even though the faulty gene does not work properly.

Key



working copy of a BRCA1 or BRCA2 cancer protection gene



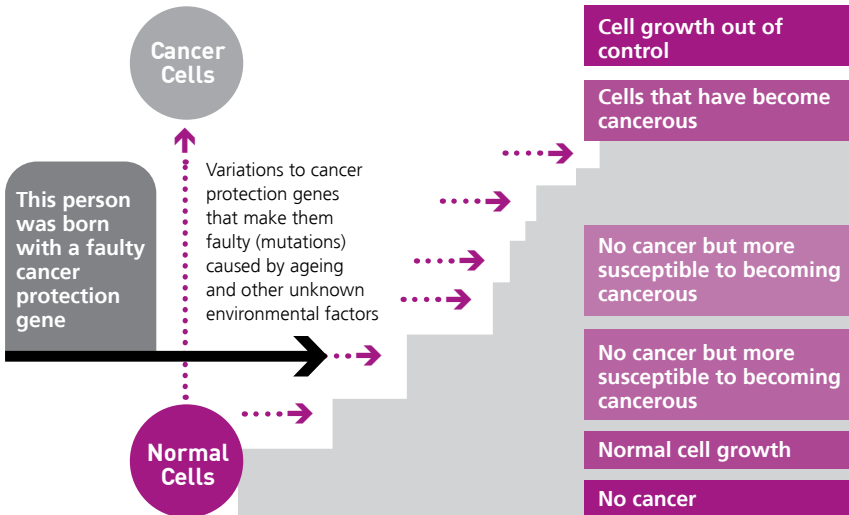
faulty copy of a BRCA1 or BRCA2 cancer protection gene

Men and women can inherit a faulty copy of the cancer protection gene. They then have a 50% chance (1 in 2) of passing the faulty copy on to each of their children.

When a woman has inherited a faulty BRCA1 or BRCA2 cancer protection gene

As shown in the diagram below, a woman who inherits one working copy and one faulty copy of the BRCA1 or BRCA2 genes will sit where the black arrow is.

She is already on the path that might lead to a cell becoming cancerous, meaning she has an increased chance of developing breast and/or ovarian cancer, as well as some other cancers.

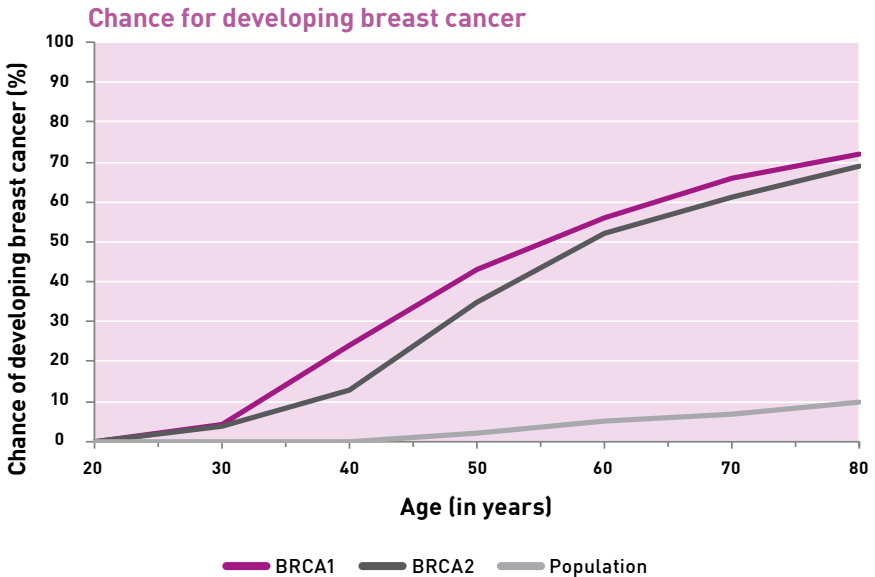


However, because variations take time to build up, this woman might not ever develop breast or ovarian cancer. If, however, she does develop breast and/or ovarian cancer, it could occur at a younger age than women who have not inherited a faulty gene.

Chance for developing breast cancer

The graph below provides the current estimate for a woman with a faulty BRCA1 or BRCA2 gene developing breast cancer in her lifetime.²

Risk of cancer will vary depending on a woman's age, family history and personal risk factors.

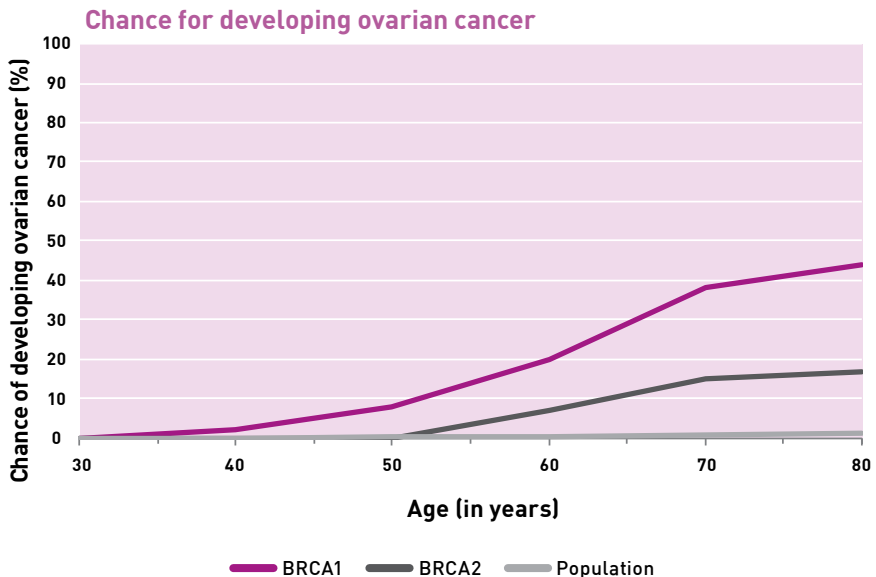


- For women who have inherited a faulty BRCA1 gene, the average age when breast cancer develops is around 44 years old.
- For women who have inherited a faulty BRCA2 gene, the average age when breast cancer develops is around 48 years old.

Chance for developing ovarian cancer

The graph below provides the current estimate for a woman with a faulty BRCA1 or BRCA2 gene developing ovarian cancer in her lifetime.²

Risk of cancer will vary depending on a woman's age, family history and personal risk factors.



- For women who have inherited a faulty BRCA1 gene, the average age when ovarian cancer develops is about 54 years old.
- For women who have inherited a faulty BRCA2 gene, the average age when ovarian cancer develops is about 60 years old.

What does it mean for a man who has inherited a faulty BRCA1 or BRCA2 gene?

Men who inherit a faulty copy of a cancer protection gene have a slightly increased chance of developing prostate, breast, and certain other types of cancer.

Most men with a faulty BRCA1 or BRCA2 gene will not develop cancer despite the increased risk.

The sons and daughters of men who carry a faulty copy of one of the BRCA1 or BRCA2 genes have a 50% chance (1 in 2) of inheriting the faulty copy of the gene.

More information for men considering BRCA1/2 testing can be found in the booklet "Understanding genetic tests for men who have a family history of breast and ovarian cancer"
(<http://www.genetics.edu.au>)

Genetic tests for an inherited faulty BRCA1 or BRCA2 gene

Two types of tests are currently available:

Mutation search genetic test.

A mutation search genetic test is a blood test that searches for variations in the BRCA1 and BRCA2 genes that make the gene faulty.

This type of genetic test is usually for a family member who has already had breast and/or ovarian cancer.

In some cases, testing may also include other genes associated with these cancers. This will depend on the family history.

Predictive genetic test.

If a mutation search identifies the faulty gene running in the family, other family members who might or might not have had breast and/or ovarian cancer can consider a predictive genetic test.

A predictive genetic test is a blood test which looks for the family specific fault in the BRCA1 or BRCA2 genes.

Mutation search: A genetic test for a faulty **BRCA1** or **BRCA2** gene

Your doctor or family cancer service will be able to tell you if a mutation search is available to you.

It is usually appropriate for:

- women who have already had breast and/or ovarian cancer and have a strong family history of the cancer
- someone who has not had breast or ovarian cancer but who has a parent and a child who has. This is not a common occurrence.

Why might I consider this type of test?

If you have already developed breast and/or ovarian cancer, a genetic test result that shows that you have inherited a faulty cancer protection gene could:

- give you more accurate information about your future chance of developing another cancer
- influence your decisions about cancer screening, prevention and management
- also help clarify who in your family has a higher chance of developing these cancers
- make it possible to offer a predictive test to your blood relatives who have not developed cancer. A predictive test investigates if they have inherited the faulty gene that runs in your family. More information about a predictive test can be found on page 31.

Possible test results for a mutation search:

1) Inconclusive (a faulty gene was NOT found)

Many families receive this result. However, there may still be a faulty gene causing cancer in the family. If you receive this result:

- It was not possible to find a variation in your BRCA1 or BRCA2 genes that makes them faulty
- Because of your family history of cancer, you may still have an increased chance of developing cancer.

Why might you receive an 'inconclusive' result?

- Your family could carry a variation in a gene that hasn't been discovered yet.
- Current technology cannot find all of the variations that make the BRCA1 or BRCA2 genes faulty.
- Your family history of cancer could be due to a combination of many factors (both genetic and environmental).

If you receive an inconclusive result, other members of your family cannot be offered a predictive test.

2) A faulty gene was found

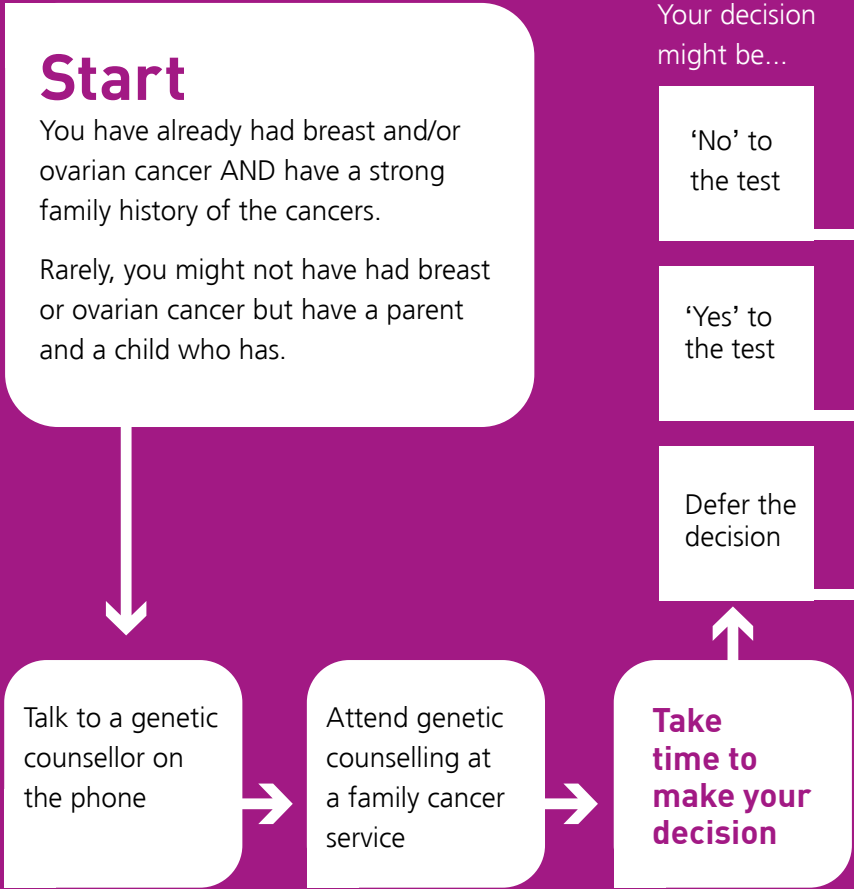
If you receive this result you have inherited a faulty cancer protection gene.

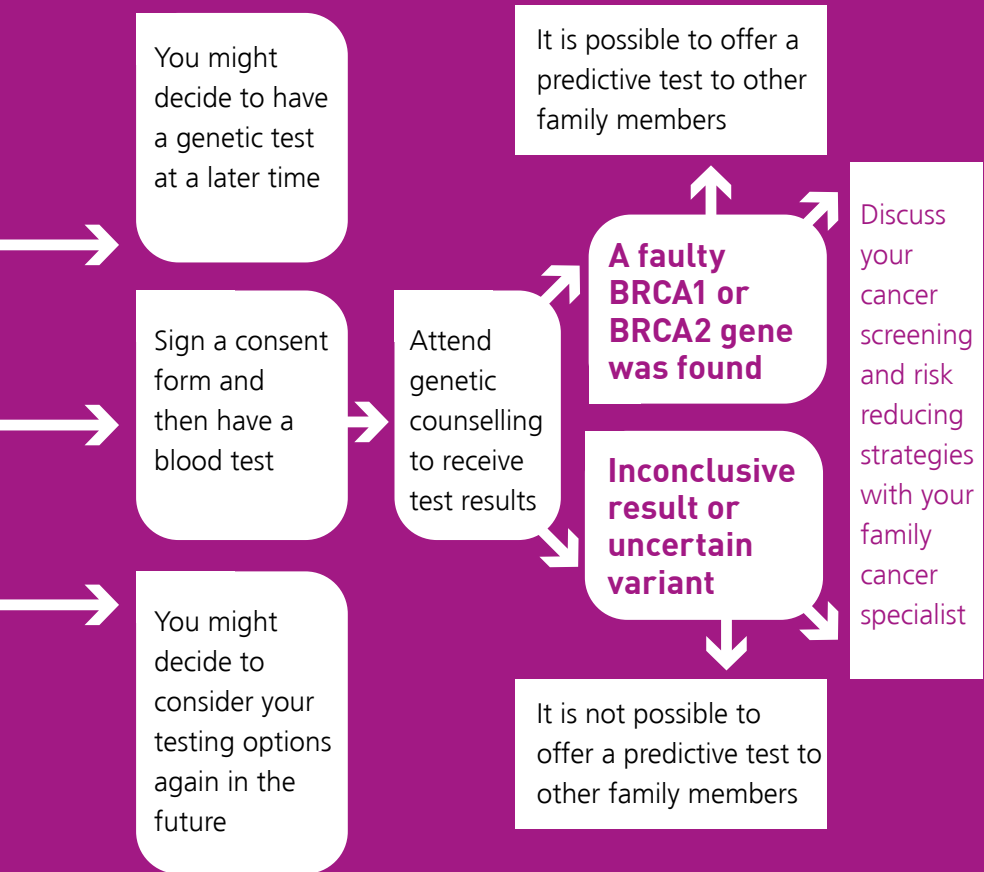
- You have an increased chance of developing more than one cancer
- Other blood relatives who might or who might not have had cancer can have a predictive test. They will be able to find out whether they have inherited the same faulty gene. See page 31.

3) Uncertain variant

If you receive this result, it means the mutation search has found a variation in the information in the BRCA1 or BRCA2 gene, but it is not clear currently if the variation is harmless, or if it is a variation that is making the gene faulty.

The mutation search process





How could a mutation search test affect me?

Waiting time

A mutation search may take several weeks. You might feel frustrated or anxious while awaiting results.

An inconclusive or uncertain variant

It can be frustrating to receive this result, generally because you do not know what has caused your family history of cancer.

You might feel some relief, even though you might still have an increased chance of developing further cancer.

It also means other family members cannot have a predictive test.

A faulty gene was found

This result could cause you to worry or feel anxious about cancer. Research has found that for women who have had genetic testing, anxiety usually lessens over about 12 months to the level that is normal for them.³ However people often continue to feel anxiety leading up to regular screening appointments.

You may feel less anxious after this result because you now understand what is causing an increased chance of cancer in your family and may be glad other family members now have the option of a predictive test.

Some questions to think about:

- How would I feel about waiting for my result?
- How might I react to an 'inconclusive' or 'uncertain variant' result?
- How might I react to a 'faulty gene was found' result?

How might my result affect my family?

Sometimes the process of having a genetic test can bring families closer together. Sometimes it can lead to disagreements.

It is important to remember that your test result might give you more information as to who in your family could have a higher chance of developing these cancers. Your genetic specialist can help you decide how best to share the information within your family.

Different people, different choices

Each individual within a family has the right to make their own decision about genetic testing. However, it can be difficult for other family members to accept these decisions. This is especially so if you choose not to have the mutation search, as this will probably mean that other relatives cannot have a predictive test.

Different people, different feelings

There is no right way to feel about genetic testing. Partners of people considering a test might also find the process difficult. They may feel left out of the decision making because they are not blood relatives, yet the results might be important for their children and future planning.

What can I do to avoid family disagreements?

It might help to think about any potential disagreements in your family ahead of time so that you can plan the best ways to handle them before deciding about a test.

You might like to consider who, when and how to tell other people about your test decision. It might help you to have a support person to share your thoughts and feelings with.

How will it affect my children?

If your children are adults:

You might like to think about what information you want to share with them about your family history of cancer and genetic testing.

You might suggest they visit a genetic counsellor for information regarding the condition. They can then decide whether or not they want to have a predictive test.

If your children are not adults:

Children are not usually offered a predictive test.

This is because even if they have inherited a faulty cancer protection gene, they do not have an increased chance of developing cancer until they are adults.

Future children:

It might be helpful to think about how your decision about genetic testing and the possible results might influence your plans for children in the future.

If you have inherited a faulty BRCA1 or BRCA2 gene there is a 50% chance (1 in 2) that each of your children will inherit the faulty gene.

Some couples may consider family planning options to prevent the faulty gene being passed on to their children. This may involve using a technique called pre-implantation genetic diagnosis (PGD) where an embryo, created through IVF, is tested before it is implanted in the womb.

How would my genetic test result affect my health management?

In some cases, genetic test results may help you and your doctors make decisions about your cancer treatment or surgery.

The results might also change the way you manage your chance of developing a new breast and/or ovarian cancer in the future.

An inconclusive or uncertain variant

You need to continue your cancer screening plan as if you haven't had the genetic test. You might still have an increased chance of developing another breast and/or ovarian cancer because of your family history.

If a faulty gene was found

If you have already had breast cancer there might be an increased chance of a cancer developing in the other breast. You might also have an increased chance of developing ovarian cancer.

Your doctor will be able to discuss health management options with you. Recommended screening options might include:

- Increased breast awareness and the reporting of any changes to your doctor
- Regular breast examination by your doctor or breast specialist
- Mammogram with or without a breast ultrasound
- Magnetic Resonance Imaging (MRI) of the breast for younger women.

Some women might consider surgery, medication, and/or lifestyle changes to reduce their chance of developing cancer. Surgical options include:

- Removal of breast tissue (risk-reducing mastectomy)
- Removal of the ovaries along with the fallopian tubes (risk-reducing salpingo-oophorectomy) after they have finished having children.

How could my test result affect my finances?

Cost of genetic testing

Patients are not usually charged for genetic testing provided by a family cancer service at a public hospital. Private laboratories usually charge for genetic testing.

Health insurance

Your genetic testing decision will not affect your health insurance.

More information about managing your chance of developing cancer and research trials is available from your family cancer service or your doctor.

Life insurance including trauma/disability and income protection policies

If you decide to have a genetic test and find you have inherited a faulty cancer protection gene you will only have to tell the insurance company your results if you take out a new policy or change your existing policy.

Your results will not affect any life insurance you have already secured.

The Financial Services Council (FSC) have also agreed that its insurance companies will:

- consider the potential beneficial effects of cancer screening or other preventive strategies
- not make someone have a genetic test if they do not want to
- not use one family member's results to assess other family members' policies when applying to the same company
- ensure confidentiality of your results.

FSC (02) 9299 3022

Information is also available from the Centre for Genetics Education:
www.genetics.edu.au or (02) 9462 9599.

Predictive test: A genetic test for the family specific faulty BRCA1 or BRCA2 gene

A predictive genetic test is a blood test available for an unaffected or affected family member for the specific faulty BRCA1 or BRCA2 gene which has previously been found to cause the cancer in your family.

Your doctor or family cancer service will be able to tell you if a predictive test is available to you.

A predictive genetic test is a blood test that looks for the specific faulty BRCA1 or BRCA2 gene which has been found to cause the cancer in your family.

Your doctor or family cancer service will be able to tell you if a predictive test is available to you.

Why might I consider a predictive test?

If you learn that you have inherited the faulty BRCA1 or BRCA2 gene that runs in your family, you can make more informed decisions about managing your future chance of developing breast and/or ovarian cancer.

If you learn that you have not inherited the faulty BRCA1 or BRCA2 gene that runs in your family, you do not have an increased chance of developing cancer. You and your children can avoid the extra cancer screening and anxiety that would be appropriate if you were at an increased chance of developing cancer.

Possible test results for a predictive test:

1) The faulty gene was not found

- You have not inherited the faulty BRCA1 or BRCA2 gene that has been found to cause the cancer in your family.
- You do not have an increased chance of developing the breast and or ovarian cancer that has affected your relative(s).

- You might still develop cancer at some stage in your life because your chance of developing cancer is the same as that of other people in the general community who do not have a family history of cancer.
- You cannot pass on the faulty gene found in other family members to your children. This means that your children do not have an increased chance of developing this type of cancer.

2) The faulty gene was found

- Your results will tell you whether it is the faulty copy of the BRCA1 or the BRCA2 gene which you have inherited. Variations in the two genes have different effects. (See pages 14 and 15).
- If you are a woman, you have an increased chance of developing breast or ovarian cancer. Remember, you might not ever develop cancer.
- Each of your children has a 50% chance (1 in 2) of inheriting your faulty BRCA1 or BRCA2 gene copy. If this occurs, you will then be at an increased chance of developing these cancers and passing the faulty gene on to your children.

You also have a 50% chance (1 in 2) that they will not inherit your faulty gene.

The predictive test process

Start
You have a family history of breast and or ovarian cancer.
Testing of a close blood relative has identified the specific fault in a cancer protection gene that is causing the cancer.

Talk to a genetic counsellor on the phone



Attend genetic counselling at a family cancer service



Take time to make your decision

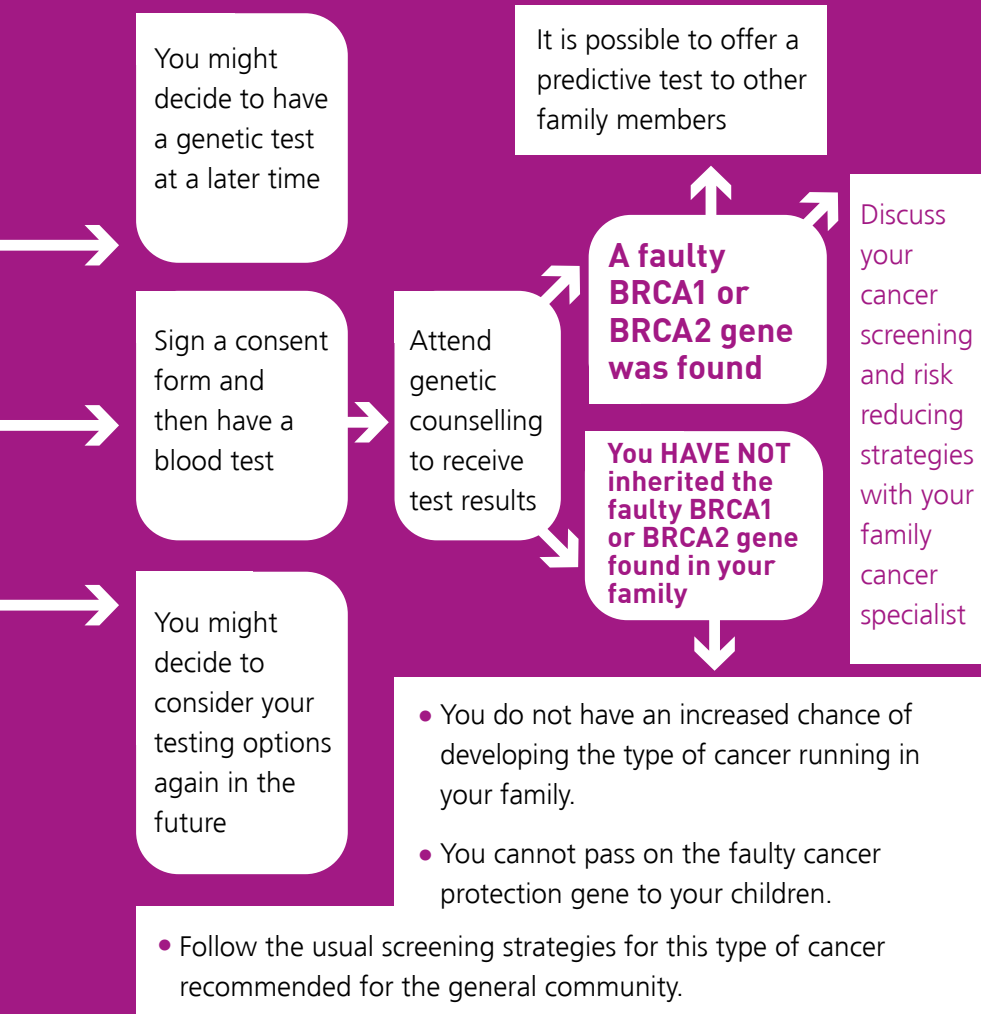
Your decision might be...

'No' to the test

'Yes' to the test

Defer the decision





How could a predictive test affect me?

Waiting time

It takes about four to six weeks for the predictive test result to become available. You might feel worried or anxious while waiting for your test results.

Your family's faulty gene was not found in you

If you receive this result you might feel relieved that you and your children do not have an increased chance of developing breast and/or ovarian cancer.

If you do not receive the same result as other family members, you might find it difficult because you cannot share their experiences.

Your family's faulty gene was found in you

This result could cause an increase in anxiety about cancer. Research has found that for women who have had genetic testing, anxiety usually lessens over about 12 months to the level that is normal for them.³ However people often continue to feel anxiety leading up to regular screening appointments.

You may feel less anxious after you receive this result because you have a better idea of your chance of developing cancer and can plan future health management.

How might my result affect my family?

Sometimes the process of having a genetic test can bring families closer together. Sometimes it can lead to disagreements.

Your genetics specialist can help you decide on how best to talk to your family about this information.

Different people, different choices

Every person in the family has the right to make their own decisions. It can be difficult for other family members to accept these decisions.

Different people, different feelings

There are no specific feelings that everyone considering a predictive test experiences.

Partners of people considering testing may find the process difficult too. They may feel left out because they are not blood relatives, and yet the results are important for their children and future planning.

Different people, different results

Sometimes family members who receive the same result will feel closer to each other. Family members who receive different results may feel uncomfortable with each other and find it harder to talk to each other. This is less likely to happen if you talk about it ahead of time.

What can I do to avoid family disagreements?

It might help to think about any potential disagreements in your family ahead of time. Then you can plan the best ways to handle this before deciding about a genetic test.

You might like to consider who, when, and how to tell other people about your predictive testing decision. You might want to choose a support person to share your thoughts and feelings with.

How will knowing about a faulty gene affect my children?

If your children are already adults

Think about what information you want to share with them regarding your family history of cancer and predictive testing.

You might suggest they visit a genetic counsellor for information regarding the condition. They can then decide whether or not they want to have a predictive test if you are found to carry a faulty copy of a BRCA1/2 gene.

If your children are not adults

Children are not usually offered predictive testing.

This is because even if they have inherited a faulty cancer protection gene, they do not have an increased chance of developing cancer until they are adults.

Future children

It might be helpful to think about how your decision about genetic testing and the possible results might influence your plans for children in the future.

If you have inherited a faulty BRCA1 or BRCA2 gene there is a 50% chance (1 in 2) that each of your children will inherit the faulty gene.

Some couples consider family planning options to prevent the faulty gene being passed on to their children, this may involve using a technique called pre-implantation genetic diagnosis (PGD) where an embryo, created through IVF, is tested before it is implanted in the womb.

How will my predictive test result affect my health management?

The results of your predictive test may change the way you manage your future chance of developing cancer.

You have not inherited the faulty gene

You have the same chance of developing breast and/or ovarian cancer as any woman in the general population. Therefore you only need the cancer screening suggested for any woman your age.

You have inherited the faulty gene

You have an increased chance of developing breast and/or ovarian cancer. However, you may never develop this cancer. Recommended screening options include:

- Increased breast awareness and the reporting of any changes to your doctor
- Regular breast examination by your doctor or breast specialist
- Mammogram with or without a breast ultrasound
- Magnetic Resonance Imaging (MRI) of the breast for younger women.

Some women might consider surgery or medications to reduce their chance of developing breast and/or ovarian cancer. Surgical options include:

- Removal of breast tissue (risk-reducing mastectomy)
- Removal of the ovaries along with the fallopian tubes (risk-reducing salpingo-oophorectomy) after they have finished having children.

More information about managing your chance of developing cancer and research trials is available from your family cancer service or your doctor.

How could my result affect my finances?

Cost of genetic testing

Patients are not usually charged for genetic testing at family cancer services at public hospitals. Private laboratories usually charge for genetic testing. The cost of testing may vary with time.

Health insurance

Your genetic testing decision will not affect your health insurance.

Life insurance including trauma/disability and income protection policies

In Australia, life insurance includes disability and income protection insurance.

If you find you have inherited the faulty BRCA1 or BRCA2 gene you will only have to tell the insurance company your results if you take out a new policy or a variation of your existing policy.

Your results will not affect any life insurance you have already secured.

If you have not inherited the faulty BRCA1 or BRCA2 gene, it may be beneficial in terms of your insurance premiums. It may be helpful to discuss your situation with your financial advisor.

The Financial Services Council (FSC) have also agreed that its insurance companies will:

- consider the potential beneficial effects of cancer screening or other preventive strategies
- not make someone have a genetic test if they do not want to
- not use one family member's results to assess other family members' policies when applying to the same company
- ensure confidentiality of your results.

FSC (02) 9299 3022

Information is also available from the Centre for Genetics Education:
www.genetics.edu.au or ph (02) 9462 9599.

My worksheet: An aid for decision making

There might be lots of reasons why you would or wouldn't have genetic testing. Part of making an appropriate decision is to consider all the pros and cons at the same time.

The worksheets on the following pages are designed to assist you with putting all the facts about genetic testing together and determining what is important to you.

What's important to me?

There are two examples of worksheets filled out by other people considering genetic testing on pages 46 and 48.

Step 1: Clarify the decision

What is the decision I face?

"I am deciding whether or not to have a mutation search (or a predictive genetic test) for a faulty BRCA1 or BRCA2 gene."

What is your reason for making this decision?

Step 2: Weighing the options

a. What I know

In the table on page 50, please list the pros and cons (positives and negatives) for you that are associated with genetic testing for breast and or ovarian cancer.

b. What is important for me?

Show how important each pro and con is to you by placing one star (*) or up to five stars (*****) in the columns labelled 'Personal importance'. More stars show more importance to you. If a pro or a con is not important to you at all, then give it zero stars.

c. How sure do I feel?

See which column has the most stars in it. The side with the most stars is probably the right option for you. Using the scale, tick the box that most closely reflects how you're feeling about a genetic test at the moment.

For a tough decision like this, people rarely feel completely sure. With careful decision making, many people feel more comfortable with their choices when they have made a written record of what mattered most to them at the time.

Step 3: Plan the next steps

In the space below, list what you need to do before you make your decision about a genetic test. There is a list of helpful contacts at the end of this booklet.

Step 4: Questions to ask your doctor

If you have any questions, you might want to write them down here and take this list to your consultation.

The design of this worksheet is based on the Ottawa Personal Decision Guide, developed by O'Connor, Jacobsen and Stacey, 2002.

Other people's experiences...

Sophie has a family history of breast cancer and has had breast cancer herself. She completed this worksheet to help her decide whether or not she would have a **mutation search** test. She listed all the pros and cons she could think of.

Personal importance	Possible pros of having a mutation search
***	"I can better manage my chance of developing another cancer."
***	"By learning if I have a faulty gene or not I will be able to clarify chances for other members of my family."
*	"I prefer to know things no matter how bad, I don't like mystery or suspense."
**	"Regardless of the result, I will have greater awareness of appropriate screening."
*	"I have to be prepared and not live with 'what ifs.'"
**	"I may be able to help my children make choices regarding their chances."
***	"So I can know if I have possibly passed it on to my children."
TOTAL 15	

*Actual quotes from people considering genetic testing for cancer risk during an Australian research project in 2006/07.⁴

This list does not include all the possible pros and cons of a **mutation search** for you. You might think of others that might be more important to you.

Possible cons of having a mutation search	Personal importance
"I might be constantly worried if I knew I had an increased chance of developing another cancer."	****
"I will be more concerned about my family's future health."	***
"I may worry about the effect of my genetic testing decision on my children in the future."	***
"It may be worrying for some family members."	**
"The testing may cause family disagreements."	**
"If I have inherited a faulty gene, it will force me to look at strategies such as surgery to lower my chances of another cancer."	**
"I just don't want to know."	**
	TOTAL 19

At this point in time are you leaning towards wanting to have a mutation search for breast and ovarian cancer or not? (Tick the box that is closest to how you feel)

I am leaning towards having a mutation search

I am not sure yet

I am leaning towards not having a mutation search

Other people's experiences...

Julie has a family history of breast cancer but has not had cancer herself. A faulty gene causing the cancer has been identified in one of her relatives. She completed this worksheet to help her decide whether or not to have a **predictive test**.

Personal importance	Possible pros of having a predictive test
*****	"I can better manage my chance of developing another cancer."
****	"By learning if I have inherited the faulty gene or not I will be able to clarify chance for other members of my family."
***	"I prefer to know things no matter how bad, I don't like mystery or suspense."
***	"Regardless of the result, I will have greater awareness of appropriate screening."
**	"I have to be prepared and not live with 'what ifs.'"
**	"I may be able to help my children make choices regarding their chance."
**	"So I can know if I have possibly passed it on to my children."
TOTAL 21	

*Actual quotes from people considering genetic testing for cancer risk during an Australian research project in 2006/07.⁴

She listed all the pros and cons she could think of. This list does not include all the possible pros and cons of **predictive testing** for you. You might think of others that might be more important to you.

Possible cons of having a predictive test	Personal importance
“I might be constantly worried if I knew I had an increased chance of developing cancer.”	***
“I will be more concerned about my family’s future health.”	**
“I may worry about the effect of my genetic test decision on my children in the future.”	**
“It may affect my, or my family’s, financial situation.”	**
“The testing may cause family disagreements.”	*
“If I have inherited a faulty gene, it will force me to look at strategies such as surgery to lower my chance.”	**
“I just don’t want to know.”	*
	TOTAL 13

At this point in time are you leaning towards wanting to have a predictive search for breast and ovarian cancer or not? (Tick the box that is closest to how you feel)



I am leaning towards having a predictive search

I am not sure yet

I am leaning towards not having a predictive search

My worksheet...

You might want to use this worksheet to help you decide if you want to have a mutation search or a predictive test.

Personal importance	My list of PROS of having a genetic test
TOTAL	

List all the pros and cons for you that you can think of and then determine how important they are to you.

My list of CONS of having a genetic test	Personal importance
	TOTAL

At this point in time are you leaning towards wanting to have a genetic test for breast and ovarian cancer or not? (Tick the box that is closest to how you feel)

I am leaning towards having a genetic test

I am not sure yet

I am leaning towards not having a genetic test

Glossary

A blood relative is someone in your family with whom you share a common ancestor but is not related by marriage or adoption. A close blood relative is a blood relative in your immediate family such as a mother, father, child or sibling.

Cancer protection genes describe genes that, when they are working correctly, help to prevent cancer from developing.

Family history of cancer means having one or more blood relatives on the same side of the family who have had cancer. These relatives could be on either your mother's or your father's side of the family.

Genes are in every cell in our body. Genes contain the information that guides our growth, health and development.

Mutations are variations in genes that make the gene faulty. This booklet discusses mutations in cancer protection genes that make them less able to prevent cancer developing.

Faulty genes are genes that are not working properly due to a mutation and so do not give the cell the correct instructions for growth and development.

Family cancer team might include:
A specialist doctor (clinical geneticist or oncologist) who has completed specialist training in genetics or cancer medicine after their general medical training
A genetic counsellor who has specialist training in counselling and in genetics

Genetic testing is the process of searching genes for specific gene faults or other variations.

Mutation search genetic testing is usually conducted first on a person who has already had breast and/or ovarian cancer. This test tries to find the specific gene variation that is causing the cancer to run in a family.

Predictive genetic testing is usually only offered to people who have a relative who has been found to have a specific fault in a cancer protection gene. A predictive genetic test looks only for the specific gene variation that has already been found in the family.

Where to from here?

Family cancer services

You may have already visited a family cancer service. If not, you may wish to contact one of the family cancer services in your area.

AUSTRALIAN CAPITAL TERRITORY

ACT Genetics Service
The Canberra Hospital
GARREN ACT 2605
Phone: (02) 5124 7630

NEW SOUTH WALES

Camperdown

Department of Cancer Genetics
Royal Prince Alfred Hospital
CAMPERDOWN NSW 2050
Phone: (02) 9515 8780

Kogarah

Hereditary Cancer Clinic
Cancer Care Centre
St George Hospital
KOGARAH NSW 2217
Phone: (02) 9113 3815

St Leonards

Family Cancer Service
Royal North Shore Hospital
ST LEONARDS NSW 2065
Phone: (02) 9463 1554

Wollongong

Wollongong Hereditary Cancer Clinic
Illawarra Cancer Care Centre
Wollongong Hospital
Private Mail Bag 8808
South Coast Mail Centre NSW 2521
Phone: (02) 4222 5576

**Newcastle and rural
outreach services**

Hunter Family Cancer Service
Hunter Genetics
PO Box 84
WARATAH NSW 2298
Phone: (02) 4985 3132

Randwick

Bright Alliance
Level 2 Prince of Wales Hospital
High St RANDWICK NSW 2031
Phone: (02) 9382 5107

Darlinghurst

Family Cancer Clinic
St Vincents Hospital
DARLINGHURST NSW 2011
Phone: (02) 9355 5647

Westmead

Familial Cancer Service
Westmead Hospital
WESTMEAD NSW 2145
Phone: (02) 8890 6947

Liverpool

Department of Cancer Genetics
Liverpool Hospital
Locked Mail Bag 7103
Liverpool BC NSW 1871
Phone: (02) 8738 9746

Other locations in NSW

For contact details of genetic counselling services in other areas of NSW that might also provide cancer genetics services, phone the Centre for Genetics Education NSW on (02) 9462 9599

NORTHERN TERRITORY

Northern Territory Clinical Genetic Service
Royal Darwin Hospital
TIWI NT 0810
Phone: (08) 8944 8731

QUEENSLAND

Brisbane

Genetic Health Qld
Royal Brisbane and Women's Hospital
HERSTON QLD 4029
Phone: (07) 3646 1686

SOUTH AUSTRALIA

Familial Cancer Unit
SA Clinical Genetics Service
SA Pathology at Royal Adelaide
Hospital
NORTH ADELAIDE SA 5006.
Phone: (08) 7074 2697

TASMANIA

Clinics held regularly at Burnie,
Launceston and Hobart.
Tasmanian Clinical Genetics Service
Royal Hobart Hospital
PO Box 1061
HOBART TAS 7001
Phone: (03) 6166 8296

VICTORIA

Clayton

Monash Medical Centre
Cancer Centre
Special Medicine Building
246 Clayton Rd
CLAYTON VIC 3168
Phone: (03) 9594 2009

East Melbourne

Peter MacCallum Familial Cancer
Centre
305 Grattan Street
MELBOURNE VIC 3000
Phone: (03) 8559 5322

Parkville

Familial Cancer Centre
Royal Melbourne Hospital
Grattan St
PARKVILLE VIC 3050
Phone: (03) 9342 7151

Heidelberg West

Austin Health Genetics Service
PO Box 5555
HEIDELBERG VIC 3084
Phone: (03) 9496 3027

WESTERN AUSTRALIA

Perth

Perth Breast Clinic
Mount Hospital, Suite 44
146 Mounts Bay Road,
PERTH WA 6000
Phone: (08) 9483 4621

Subiaco

Familial Cancer Program
Genetic Services of Western Australia
King Edward Memorial Hospital
Level 3, Agnes Walsh House
374 Bagot Rd
SUBIACO WA 6008
Phone: (08) 6458 1603

Perth

Breast Assessment Clinic
Royal Perth Hospital
PERTH WA 6000
Phone: (08) 9224 2723

My support people include (family, friends etc):

Other helpful organisations

These organisations can provide you with support and helpful books and articles about topics that concern you. They also have very informative websites.

Cancer Helpline This is a free and confidential telephone service provided by each state and territory cancer organisation.	13 11 20
Centre for Genetics Education Royal North Shore Hospital, St Leonards NSW 2065	Ph: (02) 9462 9599 Fax (02) 9906 7529 www.genetics.edu.au
The Cancer Council NSW 153 Dowling Street, Woolloomooloo, NSW 2011	Ph: (02) 9334 1900 Fax (02) 8302 3570 www.cancercouncil.com.au
The Cancer Council Victoria 1 Rathdowne Street, Carlton, VIC 3053	Ph: (03) 9514 6100 Fax (03) 9514 6800 www.cancervic.org.au
The Cancer Council Australia You can also access the other state Cancer Council websites from this page.	www.cancer.org.au
Cancer Australia Cancer Australia, amalgamated with the National Breast and Ovarian Cancer Centre (NBOCC), is Australia's national authority and information source on breast and ovarian cancer.	http://canceraustralia.gov.au
Breast Cancer Network Australia	Ph: 1800 500 258 www.bcna.org.au

Science studies

1. Australian Institute of Health and Welfare 2017. Cancer in Australia: 2017. Cancer series no.101. Cat no. CAN100. Canberra: AIHW.
2. Kuchenbaecker K, Hopper J, Barnes D, Phillips K, Mooij T, et al 2017. Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. *JAMA* 317 (23): 2402-2416.
3. Meiser B, Butow P, Friedlander M, Schnieden V, Watson M, Brown J, Tucker K. 2002. Psychological affect of genetic testing for women for breast cancer susceptibility. *European Journal of Cancer*. 38: 2025-31.
4. Wakefield C, Kasparian N, Meiser B, Homewood J, Kirk J, Tucker K. 2007. Attitudes toward genetic testing for cancer risk after genetic counseling and decision support: A qualitative comparison between hereditary cancer types. *Genetic testing*. 11 (4): 401-411.

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The design of this booklet is based on the Ottawa Personal Decision Guide (OPDG), developed by O'Connor, Jacobsen and Stacey, 2002. The OPDG and more information about decision guides are available at www.ohri.ca/decisionaid.

The information in this booklet is correct at the time of publication. However, as research is ongoing, the booklet will be updated every two years.



Notes



Further copies are available from:

Centre for Genetics Education, NSW Health

RNS Community Health Centre
Level 5, 2c Herbert Street
St Leonards NSW 2065
Phone: (02) 9462 9599 Fax: (02) 9906 7529
Email: contact@genetics.edu.au
www.genetics.edu.au

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Important

This information was correct at date of production.
Please check with your doctor or genetic counselling service for any new information.