OVARIAN CANCER AUSTRALIA

Genetic Testing and Hereditary Ovarian Cancer

A guide for women with ovarian cancer and their families

OVARIAN CANCER AUSTRALIA

IMPORTANT NOTE

The information in this booklet has been developed with the help of women who either have, or are at risk of, hereditary ovarian cancer and have been through many of the experiences you may be facing.

All information has been reviewed by appropriately qualified medical professionals and, to the best of Ovarian Cancer Australia's knowledge, reflects the state of knowledge at the time of printing. However, as new information about ovarian cancer and genetics develops and becomes available constantly, Ovarian Cancer Australia cannot guarantee and does not assume legal responsibility for the accuracy, currency or completeness of the information in this booklet. This booklet is not a substitute for medical advice from a qualified medical or other health professional.

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Introduction

Finding out you have ovarian cancer can come as a huge shock. It can bring up a range of intense emotions and difficult questions. Many women wonder what caused their cancer. They also worry that it may be a hereditary cancer that other family members may be at risk of getting.

This guide is for women with ovarian cancer and families who would like to know more about hereditary ovarian cancer. It discusses:

- what a family history means
- whether your family members are at risk
- whether you and your family should have a genetic test
- what it means if you have hereditary ovarian cancer
- where to seek further support.

We hope the information will help you and your family make an informed decision about whether you should have a genetic test. It explains what the results mean and where you can get support if your test is positive.

If you would like more detailed information about ovarian cancer and its diagnosis and treatment, including where to access support, call Ovarian Cancer Australia on **1300 660 334** and ask for a copy of our Resilience Kit or download the kit: www.ovariancancer.net.au

CONTACT US

Please contact us on **1300 660 334** during business hours if you would like further support and resources or email the support team support@ovariancancer.net.au.

Introduction

Some definitions

This table defines some important terms used in this booklet. Please also see the glossary at the end of the booklet.

TERM	DEFINITION
Cancer 	A disease where cells in the body behave and grow abnormally and form a malignant (cancerous) growth called a 'tumour'. Malignant tumours can spread into other parts (tissues) of the body.
Cells 	The 'building blocks' of the body. A human is made up of billions of cells, which are adapted for different functions. Cells can reproduce themselves exactly, unless they are abnormal or damaged, as are cancer cells.
DNA* 	An extremely long chain of particles (molecules) containing all the information necessary for the life functions of a cell. It is our genetic coding.
Gene 	Small pieces of DNA: the material that acts as a 'master blueprint' for all the cells in your body. Your genes determine such things as what colour hair and eyes you have and how tall you are. If you inherit specific gene faults (mutations), it may mean you have an increased risk of developing certain cancers.
Genetic 	Relating to genes. A genetic condition is caused by a fault (mutation) in one or more genes and may have been inherited.

Introduction

TERM	DEFINITION
Faulty gene**	A change in the structure of a gene. The gene fault (mutation) may be passed from parent to child during conception. In this way, it can be passed from generation to generation. In this booklet, 'gene mutation' and 'faulty gene' mean the same thing.
Familial cancer	When a gene fault causes higher rates of particular cancer/s within a family. These cancers often affect family members at a young age.
Hereditary	Conditions, including diseases, capable of being passed from a parent to child through their genes during conception.
Hereditary cancer	When a gene fault linked to a cancer/s is passed from parent to child during conception. These are called 'inherited cancer genes'. You may develop a hereditary cancer from these genes. All cancer is triggered by faulty genes, but only 5–10% of cancers are hereditary.
BRCA1 and BRCA2 mutation	Faults in two genes, known as BRCA1 and BRCA2, which are associated with a higher risk of developing breast, ovarian and some other cancers. These faulty genes can be passed from parent to child.

*deoxyribonucleic acid. **also known as a gene 'mutation' or 'alteration'

Introduction

What is ovarian cancer?

‘Ovarian cancer’ is a general term used to describe a cancerous (malignant) tumour found in one or both ovaries.

The ovaries are made up of three main kinds of cells – epithelial cells, stromal cells and germ cells. Each of these cells can develop into a different type of tumour. The average age of women when they are diagnosed with ovarian cancer is 64. Ovarian cancer is most common in women over the age of 50; however, sometimes younger women get ovarian cancer. Ovarian cancer is the eighth most common cancer in Australia. About 1580 Australian women are diagnosed each year.

It is important to mention that some ovarian cancers start in the fallopian tubes, which link the ovaries to the uterus. Cancer can start in these tubes and then spread to the ovary. Cancer beginning in the fallopian tube is sometimes called ‘fallopian tube cancer’ (FTC). Whether the cancer starts in the fallopian tubes or ovaries, they are staged and treated in the same way.

Primary peritoneal cancer (PPC) is a relatively rare cancer that develops in a very similar way to epithelial ovarian cancer. We mention it as women diagnosed with this type of cancer should also be offered or encouraged to have genetic testing.

Meet Chrissy, who inherited a BRCA gene fault. She is one of many real-life women featured in this booklet

Hereditary ovarian cancer

What is hereditary ovarian cancer?

Cancer is a common illness, with one in three Australians developing cancer in their lifetime.

This means it is not unusual for members of the same family to be diagnosed with different types of cancer. Within a family, one person may have lung cancer, another have skin cancer and another have stomach cancer. While all of the cancers occur in the one family, the cancers are not necessarily inherited or related, unlike hereditary cancer.

'Hereditary cancer' is a cancer that develops in a person who has inherited a gene fault (mutation). The cancer that develops is specific to the faulty gene – such as breast cancer in a person who has inherited BRCA1.

WHAT ARE GENE FAULTS (MUTATIONS)?

Genes are made up of DNA. Genes act as 'chemical instructions' that tell our body's cells what to do. They control our body's growth, how we look and how our body functions. It is estimated humans have between 20 000 and 25 000 genes. We all inherit a set of genes from each of our parents.

Usually our genes allow our cells to function normally. But sometimes genes change. The change may stop that gene working as it should. Scientists call these changes 'faults', 'alterations' or 'mutations'.

People who inherit a faulty gene from a parent have a 50% chance of passing the faulty gene on to their children. If you inherit a faulty gene, it will mean you are at a greater risk of developing certain types of cancers. Approximately 20% of all ovarian cancers are caused by a faulty gene that has been passed from a biological parent.

Certain types of ovarian cancer, such as high-grade serous, are more likely to be caused by a faulty gene.

Ovarian cancer caused by inheriting a faulty gene is called 'hereditary ovarian cancer'. Another term you may hear is 'family' (or 'familial') cancer, which means cancer that occurs or tends to occur in families. This is sometimes called having a 'family history of ovarian cancer'.

Hereditary ovarian cancer

WHAT INCREASES MY RISK OF HEREDITARY OVARIAN CANCER?

Women who have a family history of ovarian cancer may be at greater risk of developing the disease than the general population, because of an inherited gene fault. You are considered to have a strong family history of ovarian cancer if:

- You have two or more close relatives on the same side of the family (father's or mother's relatives) who have or had ovarian cancer, and the cancers were diagnosed when the relatives were younger than 50.
- You have a family member who has had genetic testing confirming they have a faulty gene.
- You have Ashkenazi Jewish ancestors (who have a higher incidence of BRCA mutations than the general population).
- Your own cancer was diagnosed at an early age, or you have had more than one type of cancer.

If you or someone you know is worried about developing a hereditary cancer give us a call.

CALL

Ovarian Cancer Australia on 1300 660 334 or talk to your GP or cancer specialist, who can refer you to a family cancer centre.

Hereditary ovarian cancer

Genes that increase the risk

Mutated BRCA1 or BRCA2 genes (BReast CAncer genes 1 and 2) are involved in most cases of hereditary ovarian cancer. Inheriting one of these faulty genes increases a woman's risk but doesn't mean you will definitely develop ovarian cancer.

Some faulty genes are also linked to an increased risk of ovarian cancer. Scientists have not identified all the faulty genes linked with an increased risk of ovarian cancer.

BRCA1 AND BRCA2 GENES

The BRCA1 and BRCA2 genes were discovered in the mid-1990s after researchers identified some families who had a lot of cases of breast cancer. A fault in the BRCA1 or BRCA2 gene accounts for most cases (85–90%) of hereditary ovarian cancer. These faulty genes can also be associated with other cancers, including cancer of the fallopian tube, prostate, peritoneum, pancreas, breast (in men) and melanoma.

BRCA1 and BRCA2 genes normally help to prevent cancer, but when a woman inherits a faulty version of either gene, she is less protected against cancer.

- Women who inherit a faulty BRCA1 gene have about a 40% lifetime risk of developing ovarian cancer.
- Women who inherit a faulty BRCA2 gene have about an 18% lifetime risk of developing ovarian cancer. This compares to a less than 2% risk for the general population.

Many women who have a faulty BRCA1 or BRCA2 gene **do not** have a known family history of ovarian or breast cancer. This can happen for many reasons, including:

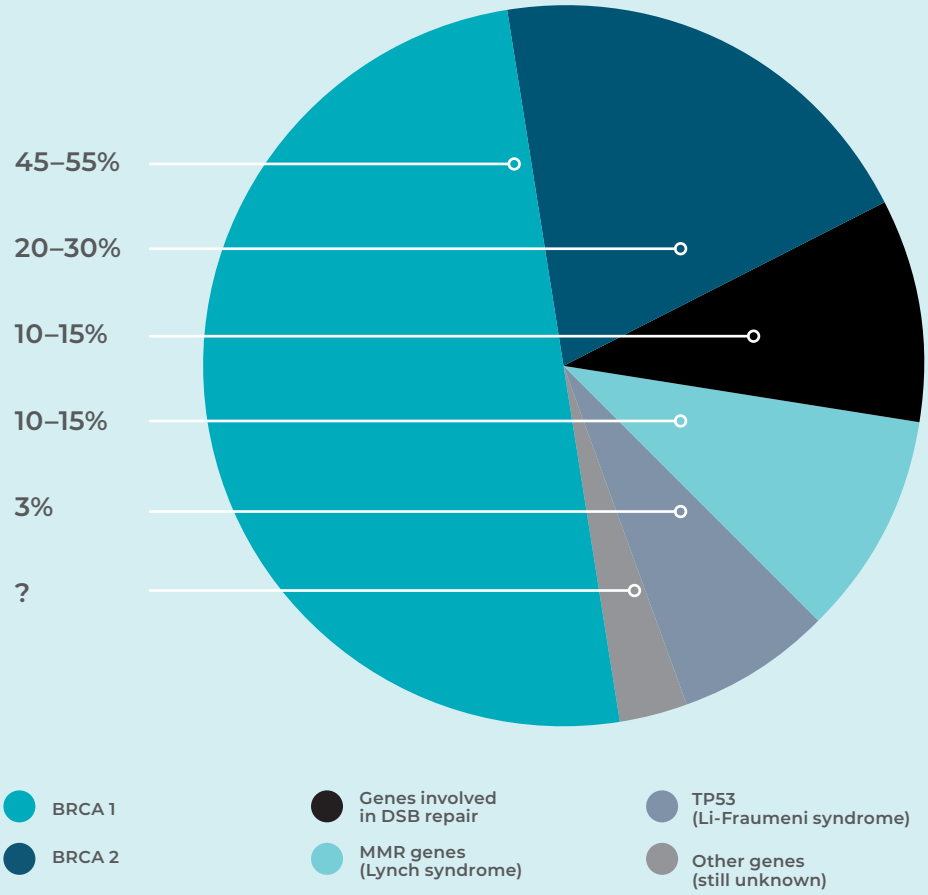
- other family members may have inherited a faulty gene but not have developed ovarian or breast cancer
- there may be few females in a family, making it difficult to see a pattern of family history
- there may be very little known about the genes of a woman's relatives.

Meet Krystyna, who has had breast and ovarian cancer and inherited the BRCA1 gene from her father

Hereditary ovarian cancer

OTHER GENE FAULTS (MUTATIONS)

Although rare, inheriting faulty genes other than BRCA1 and BRCA2 can also increase your risk of developing ovarian cancer. The pie chart shows some other faulty genes that increase a woman's risk of ovarian cancer.



SOURCE: Adapted from Toss A et al, 'Hereditary ovarian cancer: Not only BRCA 1 and 2 genes', Biomed Res Int 2015; 17 May, www.ncbi.nlm.nih.gov/pmc/articles/PMC4449870/.

Hereditary ovarian cancer

Not everyone who inherits a fault (mutation) in the BRCA1 or BRCA2 gene will develop cancer, but it does increase the risk.

Lynch syndrome (also known as hereditary nonpolyposis colorectal cancer syndrome (HNPCC) is a bowel cancer predisposition linked to faults in one of the genes **MLH1**, **MSH6** and **PMS2**. Women who inherit one of these genes have approximately a 9–12% lifetime risk of developing ovarian cancer.

RAD51C and **RAD51D** gene faults (very rare and increase the risk of developing ovarian cancer by the age of 80 by about 10%).

STK11 gene may also increase the risk of developing a type of ovarian cancer known as a sex cord-stromal tumour. Faults in this gene cause a very rare condition called Peutz-Jeghers syndrome, which causes an 18% risk of developing gynaecological cancers by the age of 70.

BRIP1 (FANCJ) gene increases the risk of ovarian cancer by about 2–6%. There is currently no routine test available to check for faults in the BRIP1 (FANCJ) gene.

Others – TP53, BARD1, CHEK2, RAD51, and PALB2

Inheriting any of the faulty genes mentioned in the pie chart may increase your risk of developing other types of cancers, not only ovarian cancer.

A clinical geneticist (a doctor who specialised in genetics) or a genetic counsellor will be able to look at the faulty genes you have inherited and give you an accurate and individual risk assessment. You can find a genetic counsellor or geneticist at a family cancer centre; for a list of centres, see Cancer Council Australia (www.cancer.org.au).

This booklet focuses on the BRCA1 and BRCA2 gene faults as these are currently the most commonly tested. Research into testing for other genes is in progress, along with research that may discover other unknown genetic links.

Hereditary ovarian cancer

Should I find out if my ovarian cancer is hereditary?

Thinking about whether your ovarian cancer is hereditary can be confronting, especially if you have only recently received the diagnosis. This is already a shock, so having to think about whether you carry a faulty gene can be a lot to handle.

There are several reasons why it is important to find out, including:

- it may influence the treatment you need for your ovarian cancer
- it could mean other family members will also have inherited the faulty gene putting them at an increased risk of ovarian or breast cancer
- some hereditary ovarian cancers also increase your risk of other cancers.

This booklet discusses each of these reasons and its possible outcomes.

"My mum was very reluctant at first to have a genetic test. She told me not to worry as I was more like my father and unlikely to inherit cancer from her. But I told her perhaps we do have this faulty gene – I had to be realistic. I just needed to know either way, so I could be more in control of my life." – Michelle, age 39

Michelle

Genetic testing

Who should have genetic testing?

All women diagnosed with invasive epithelial ovarian cancer, as well as those with fallopian tube or primary peritoneal cancer, whatever their age or family history, are encouraged to have genetic testing. They may carry a fault (mutation) in BRCA1 and/or BRCA2 even if they don't have any family history of ovarian cancer.

If you have not been offered genetic testing, we recommend you ask your specialist about this.

Cancer Australia recommends a woman with ovarian cancer should be offered genetic testing for a faulty BRCA1 or BRCA2 gene if:

- she has a Grade 2 or 3 invasive non-mucinous ovarian cancer diagnosed at 70 years or younger
- she has invasive non-mucinous ovarian cancer and a personal history of breast cancer (regardless of age)
- she has invasive non-mucinous ovarian cancer and a family history of breast or ovarian cancer (regardless of age)
- she is from a population known to have a higher risk of faulty BRCA1 or BRCA2 gene (such as Ashkenazi Jewish women)
- her ovarian cancer has come back after treatment with a platinum-based treatment and meets the Medicare Benefits Schedule (MBS) criteria for treatment with a poly ADP-ribose polymerase (PARP) inhibitor (see later section titled 'PARP inhibitors').

See the Cancer Australia website for more information (www.canceraustralia.gov.au).

Genetic testing

If you are found to have a faulty gene associated with a higher risk of ovarian cancer, your doctor may refer you to a genetic counsellor (a healthcare professional who is experienced in cancer genetics). They will suggest offering genetic testing to other family members. If other family members have inherited a faulty gene associated with an increased risk of ovarian cancer, a gynaecological oncologist can advise about ways to reduce their risk (see later in this booklet).

"Both my mum and her twin sister had breast cancer in their early 50s but they were treated, and everything was okay. My gran died of pancreatic cancer as did my uncle. It wasn't until Aunty developed ovarian cancer when she was in her 50s and the BRCA gene was picked up. Soon after this my mother was also diagnosed with ovarian cancer." – Lisa, age 45

Genetic testing

What does genetic testing involve?

If you are eligible for testing, your doctor may refer you to a genetic counsellor. You may also have genetic testing through your gynaecological oncology team.

The counsellor will be able to help you decide whether to have the test or not, and what this may mean for you and your family members.

During your first session with the genetic counsellor, they will take a detailed family history from you. They will also discuss:

- the specific tests you will have and the accuracy of these tests
- the benefits of genetic testing
- what a positive or a negative test result may mean in relation to your medical treatment
- how genetic test results can affect you emotionally and psychologically
- the risk of passing a gene fault to your children and how this may affect you.

Because of the possibility of other family members also inheriting the faulty gene, it can be very helpful to discuss genetic testing with your relatives at an early stage. Talk about this with your genetic counsellor.

The test itself is a simple blood test.

You will have a follow-up appointment with the genetic counsellor after the blood test to discuss your results. This may be done via a letter, phone call or in person. Thinking about having an ovarian cancer genetic test can bring up strong emotions. Some women say they feel an uncertainty about having the test and can become anxious about the possible impact the results of the testing will have on themselves, family and others close to them. Feelings of fear and guilt are common as women worry about passing the gene on to their family members. We discuss how you might feel later in this booklet.

Genetic testing

WHAT TESTING MEANS FOR YOU

Some women find it helpful to know why they developed ovarian cancer. But there will be several factors to consider if you are told you have a hereditary cancer. For example, if you have a BRCA1 or BRCA2 gene fault you will be at greater risk of developing breast cancer and some other types of cancer compared with the general population. It may also influence the type of treatment you receive, which your doctor will explain to you. You can find more information about this later in the booklet.

WHAT TESTING MEANS FOR YOUR FAMILY

Another consideration is the impact of your test results on your family. If you have the faulty gene, other family members may also have it. Depending on the side of the family affected by the faulty gene, this may include first-degree relatives such as your mother, father or siblings as well as your aunts, uncles and cousins.

Children who have a parent who has a BRCA1 or BRCA2 gene fault will have a 50% chance of inheriting this fault. This means they are at an increased risk of developing certain types of cancers.

We discuss talking to your family about your result and what it means for them later in the booklet.

"Finding out my sister and I both had the BRCA2 gene fault made sense. Finally, there was a reason why both mum and her sister had been diagnosed with and had passed with ovarian cancer. It is more important for us to know and take preventative steps than not to know at all!" – Karen, age 28

Genetic testing

When should I have genetic testing?

When you have a genetic test done is up to you. Everyone is different, and you need to approach this in a way that works for you. For some women, this may mean having it done immediately; for others, it might mean waiting a while. It is okay to allow yourself time to think about having the test and gather as much information as you feel you need to make an informed decision. Some women have DNA stored (from a blood sample) so that it is available for genetic testing at a later date.

We encourage women to consider testing at the time of diagnosis to help your treating team determine the best treatment for you.

"When I found out I could possibly carry the faulty gene, I put my hand up immediately to have the test." – Christine, age 52

"My pragmatic mum decided to get tested when she was first diagnosed. This enabled me to also get tested to assess my risk. I then had time to think about what I needed to do to reduce my risk and do it when I was ready." – Anne, age 39

Women who do not have ovarian cancer but are worried about their family history are encouraged to speak with a genetic counsellor about having genetic testing.

"Once I knew Mum had the BRCA gene I wanted to find out as soon as possible if I had it, even though I was afraid of what it could mean for me. But I needed to know so I could make informed decisions about how to decrease my risk." – Michelle, age 39

You may be given a choice of how you'd like to receive the results – this could be by phone or in writing, or sometimes face to face.

Genetic testing

"I took my results as a phone call and although I had a very strong feeling my result would be positive – actually hearing that was a very different thing. For a couple of months after this I was very low. I cried a lot but then I also knew I could now take control and do whatever I could to reduce my risk. Not knowing felt like putting your head in the sand for me." – Michelle, age 39

Results are usually available between four and six weeks although it can sometimes take longer.

After you get your test result, you will speak with the genetic counsellor again, who will explain the result and what the next steps are.

THE STEPS TO HAVING A GENETIC TEST

1. Referral by a health professional or self-referral to your local genetics clinic
2. Getting an appointment at a family cancer centre with a clinical geneticist or genetic counsellor
3. Completing a family history form before your appointment
4. Speaking at the appointment about what is involved in a test and what the implications will be for you and your family
5. Signing a consent form if you go ahead with the test
6. Having the blood test
7. Receiving the results.

"I thank my lucky stars every day I was given the opportunity to be tested and find out I was BRCA positive when I did. Being pro-active, and an enormous dollop of luck, has given me and my family outcomes that have been advantageous to us. I will always support women who choose to have genetic testing as it can have enormous benefits and save your life." – Christine, age 52

Results of genetic testing

Possible results of genetic testing

There are three possible results for testing for a BRCA1 or BRCA2 gene fault (mutation):

- **Mutation not identified** (sometimes called a 'negative' result): Testing has not found a gene fault.
- **Variant of unknown significance (VUS)** (sometimes called an 'inconclusive' result): Testing has found a change in the gene, but it is uncertain whether this could cause an increased cancer risk.
- **Mutation present** (sometimes called a 'positive' result): Testing has found a fault that causes a significant change to the gene and is therefore likely to be the cause of your cancer.

Results of genetic testing

What do the different results mean for my family and me?

MUTATION NOT IDENTIFIED (NEGATIVE RESULT)

Finding out your cancer is not hereditary/genetic often brings a sense of relief and reassurance.

If no faulty gene is found, and you do not have a family history of ovarian or breast cancer, it is most likely your ovarian cancer has happened by chance. It means your risk, and the risk to your children, of developing breast cancer and other cancers associated with the BRCA1 and BRCA2 genes is likely to be the same as the general population.

If you receive this result but have a strong family history of ovarian or breast cancer, the result may be confusing. Scientists are still identifying all the different genes that may influence a woman's risk of ovarian cancer. It is possible that you have a fault in a different gene that is not yet known, and therefore cannot be detected by current tests. This means there is still some uncertainty, even after genetic testing. After going through the counselling and test, this may be challenging to accept. Your genetic counsellor will be able to provide support if you need it.

Can my family members still have a genetic test?

If your test does not find the faulty gene, your family members don't need to have a genetic test.

If you have a strong family history of cancer, your close relatives may like to receive advice about reducing their risk.

Results of genetic testing

VARIANT OF UNKNOWN SIGNIFICANCE (VUS) (UNCERTAIN RESULT)

If your result comes back showing a genetic variant of unknown significance (VUS), this means the result is unclear. It means you have a fault in the BRCA1 or BRCA2 gene, but it is not known if this fault causes an increased risk in ovarian cancer or not. This can be confusing to hear.

Fewer women are receiving this result, as scientists are learning more about the different gene faults that increase cancer risk.

In time, further information about the VUS may be discovered. If you've received a VUS result, it's recommended you contact a genetic counsellor every one to two years to see if any additional gene faults have been discovered.

Can my family members still have a genetic test?

If you receive a VUS result, this means free government-funded genetic testing is not available for your family. But based on family history, they still may like to get advice about reducing their risk of developing cancer.

Getting a VUS result can cause higher level of uncertainty for a family as it is less clear whether you or your family have an increased risk. This can be emotionally difficult to cope with and many people need further support. Talk with your doctor or genetic counsellor about possible options.

MUTATION PRESENT (POSITIVE RESULT)

A positive result means a gene fault has been found that is known to be disease-causing.

The result will influence:

- your risk of developing other types of cancers
- the risk of your family members developing cancer
- the type of treatment you may have.

Your genetic counsellor will explain the impact of your result and offer you support on how to deal with this. Sometimes you can get your results and support via a telephone genetic counsellor provider. Your medical team will guide you about the type of treatment that is best for you.

Results of genetic testing

"Genetic testing has given me an opportunity I would not have otherwise had. It changed my narrative (story) and I will always be grateful for that." – Christine, age 52

How might I feel?

Finding out you have a faulty gene that has caused your cancer may bring up many feelings and reactions. At first you may feel shocked, angry and upset. Being given a cancer diagnosis, as well as finding out your cancer is hereditary, can be difficult to accept. Some women say it feels like they have been diagnosed all over again, as they now know they may get another cancer in the future and/or a family member may develop a cancer. For some women this may be the hardest part to deal with. Feeling anxious about what this means for you and your close family is very natural.

There can also be a sense of relief, as you now have an explanation for the cancers in your family.

Some women feel guilty. Feeling responsible for possibly passing the gene on to your children and grandchildren can be very difficult. But it is important to remember **you can't control the genes you inherit from your parents or the ones you pass on to your children. You are not to blame.**

"Mum certainly didn't pressure my sister and I to have the tests and the first thing she said to us when she found out about her result was, 'I am so sorry darling'." – Lisa, age 45

"It is not your fault if you pass this gene on to our children, and if you do then we will deal with it as things happen." – Adrian, partner of Michelle, age 39

"I knew it wasn't Mum's fault I had the inherited gene and I didn't want her to feel it was either." – Michelle, age 39

With time, most women adjust to finding out they carry a faulty gene. Although in some cases it does not change their own diagnosis and possible outcomes, they feel empowered to be able to pass on important information to their family

Results of genetic testing

members. It allows their family to make their own choices about genetic testing.

"My two daughters were tested when they turned 25 – one daughter has the faulty gene and the other doesn't. As a mother, my whole life has been about doing all that I can to ensure my children grow up healthy and happy. Thinking I may have passed something on to my children that would undermine their health has been very challenging. My husband and I will always be there for her and as a family we just move forward in the direction we need to in order to make the best of things for each of us." – Christine, age 52

How will my result affect family members?

Mutations in BRCA1 and BRCA2 genes increase a woman's risk of developing several other cancers including:

- breast cancer
- fallopian tube cancer
- peritoneal cancer
- pancreatic cancer
- melanoma.

Men with BRCA2 mutations (and sometimes BRCA1 mutations) are at an increased risk of breast, prostate, pancreatic and peritoneal cancer.

It is important to know that carrying the BRCA1 or BRCA2 mutation **does not** mean you will definitely develop breast cancer or another type of cancer. And there are also ways to manage your risk, which we discuss later in this booklet.

Christine

Results of genetic testing

"Having the BRCA gene mutation does not mean I am definitely going to get cancer – it is not a death sentence. I tried to flip the information around and make something good out of it. I found it empowering to find out I had the gene as now I am in control as much as I can be. I'm doing everything I can now and if I end up with cancer before I have my surgeries, that will be very sad and tragic but at least I know I have done everything I possibly could. That brings an element of peace to it all." – Michelle, age 39

TELLING YOUR FAMILY ABOUT GENETIC TESTING

Telling your family you are having a test to find out if your cancer is hereditary can be hard. You may be especially worried about young family members. This is natural, and many women feel the same.

Confiding in your partner or someone close to you can help ease anxiety. You can also gain support from your medical team and genetic counsellor.

Your family are likely to be wondering whether your cancer diagnosis means they are at an increased risk of cancer. They will want to understand their risk. Most people will be grateful to know they have the choice of finding out more about testing. They can then make their own decisions about managing their risk with additional screening or surgical options.

Finding out you have an increased risk of cancer can also cause anxiety for your relatives. Some may have concerns about how this might impact their work, insurance and personal relationships. They may be reluctant to have the test, which is understandable. We discuss this in more detail later in this booklet.

How will my result affect family members?

If you have a positive result for a faulty BRCA1 or BRCA2 gene, this means your adult family members (age 18 and over) will be able to have a test to see if they have the same gene fault. This is called a 'predictive test'.

A positive test result means the person has an increased chance of developing cancer, but it doesn't mean they have cancer or that they are definitely going to get it.

Results of genetic testing

There are other implications for family members if they inherit a faulty BRCA1 or BRCA2 gene.

- Your siblings and children will have a 50% chance of carrying the faulty gene.
- You inherit the faulty gene from your mother or father. If they did not already know about their BRCA status, they may want to seek advice about testing, as can their siblings, nieces and nephews (your aunts, uncles and cousins).
- Everyone who inherits a faulty gene can pass the gene faults on to their children during conception, whether they develop cancer themselves or not.

Genetic testing is only available to people over the age of 18. If you are worried about younger children, talk to your doctor or genetic counsellor.

It is important to understand the implications of having a genetic test and to make an informed choice about having it.

"I found out I had the BRCA gene and so did one of my daughters – she carried both gene mutations. She had surgery and is thinking about having a double mastectomy. It has affected her a lot mentally and she has suffered a lot of anxiety since." – Vicki, age 59

How will my family feel?

Everyone is different. Your relatives may have mixed responses when you tell them you have had genetic testing, and that they may carry the gene fault.

Some women say their relatives did not want to know their result. Others may be reluctant to have genetic testing. This can be challenging and upsetting. If this happens, your family member may speak to your genetic counsellor to find out their risk and options for testing.

"My original thought was: why would I want to get tested and find out I might have a ticking time bomb inside me? I thought I don't look like my Mum, so I am unlikely to have the faulty gene and Mum had always told me I am more like Dad than her. I did not want to know." – Lisa, age 45

Ultimately, the decision to have predictive genetic testing is up to each person.

Results of genetic testing

You will have given them an option that may not otherwise have been available.

"My sister had a different approach to me. She was more reluctant to get tested and did not want to talk about it. But I wanted to know and do something as soon as possible to prevent my risk of getting cancer." – Lisa, age 45

Results of genetic testing

Family members WITH a BRCA gene fault: what now?

If a family member has genetic testing and is found to carry a gene mutation, they will have genetic counselling to discuss what this means for them and their options for risk management.

The previous section outlined some of the options for reducing the risk of breast cancer, and that screening options for other cancer types may also be suitable. Women may also want to consider risk-reducing options for ovarian cancer.

There is no reliable screening test for ovarian cancer. Research has shown that routine pelvic ultrasounds and measuring CA125 (a protein in the blood) are not sufficiently sensitive to detect ovarian cancer early enough.

To reduce the risk of developing ovarian cancer, women with a faulty BRCA gene may have their ovaries and fallopian tubes removed (risk-reducing salpingo-oophorectomy or RRSO). A hysterectomy (removal of the uterus) may sometimes be recommended but is not always necessary. Discuss with your specialist or genetic counsellor when the right time might be to have this operation.

It is important to discuss with your treating team the pros and cons for having the surgery and whether it is the best option for you.

REMOVAL OF BOTH OVARIES AND FALLOPIAN TUBES

This operation, known as a bilateral salpingo-oophorectomy (BSO), will greatly reduce the risk of developing ovarian cancer. It may also reduce the risk of developing breast cancer, although the evidence for this is currently unclear.

Having a BSO does not guarantee 100% that a woman won't develop ovarian cancer. This is because there is a small chance that cells invisible to the naked eye (microscopic cells) may begin to grow in the abdomen or pelvis before the ovaries are removed. These cells can't be taken out during this operation.

If a woman decides to have a BSO, research suggests it is best to do this before starting menopause. This is because she will gain the greatest benefit of decreasing her risk of developing ovarian cancer.

Results of genetic testing

It is often a difficult decision to make as it will mean the woman will go through an 'immediate' or 'surgical' menopause. Instead of the gradual transition into menopause that usually happens with age, symptoms may develop suddenly. This can be challenging and adjusting emotionally can take time. There are effective treatments to help manage the symptoms of surgical menopause, which you can discuss with your treating team.

The Ovarian Cancer Australia fact sheet *Early menopause from ovarian cancer treatment* explains what to expect, helpful ideas for managing the short and long-term effects, and who to contact for more information and support (www.ovariancancer.net.au or **1300 660 334**).

"I aim to have my children and then have a removal of my ovaries and tubes as soon as possible after that. I think I will also potentially have breast surgery, but I am not 100% sure about that yet." – Michelle, age 39

CONTRACEPTIVE PILL

Research suggests that women who have used the oral contraceptive pill for five years or more are less likely to develop ovarian cancer than women who have never used it. It is not suitable for all women and it can slightly increase a woman's risk of developing breast cancer.

A woman thinking of taking the oral contraceptive pill should first discuss her options with her GP or genetic counsellor.

FAMILY PLANNING

A genetic counsellor can talk to your family members about having children and the impact on existing children.

There are several family planning options for men and women with a faulty BRCA gene, some of which are listed below. There are also some reproductive technologies available using in vitro fertilisation (IVF) that can significantly reduce the chance of having a child who carries a known genetic fault.

Couples who are interested in learning more about their options are strongly advised to speak with their genetics specialist/counsellor.

Results of genetic testing

The following options might be discussed during genetic counselling:

- to have children without any intervention – each child would have a 50% chance of inheriting the BRCA mutation
- egg or sperm donation
- pre-natal testing – some couples may choose to get pregnant naturally and have a test during the pregnancy to see if the foetus has inherited the BRCA mutation (the couple could then decide whether to continue with the pregnancy)
- pre-implantation genetic diagnosis (PGD) – some couples may choose this option to avoid passing the BRCA mutation to their children. PGD involves removing a woman's eggs to fertilise in a test tube (IVF). When the embryos reach a few days old, a cell is removed and tested for the hereditary disease. Once the genetic status is determined, the parents can decide which embryos they want implanted
- to choose not to have children because of the risk of passing on the BRCA gene mutation
- adoption.

Your family members may also want to discuss with their genetic counsellor the implications for them, the costs and possible funding options available. It's important to remember there is no right or wrong decision. Everyone must feel comfortable to do what feels right for them.

Results of genetic testing

"I talked to my counsellor about having a baby and the possibility of passing the gene on to my child. I needed to work through how this would make me feel and how would I support my child if I do in fact pass it on. But even having worked through these scenarios and finding my peace, I am still hopeful for the future in terms of medical advances for screening and support. My partner and I have had many conversations and I have not stopped asking questions and seeking out information. I wanted to know everything and look at every possible scenario before deciding to have children. I am now comfortable with my decision and know that even if it is not okay – it will be okay. And from that, I am finally in a good place where I am ready to start trying for a baby now and know that if I am lucky enough to become a parent, I will tell them when they are old enough to know and that Mum and Dad are here to support you if you have the gene. I will be there EVERY step of the way." – Michelle, age 39

Results of genetic testing

Family members WITHOUT a BRCA gene fault (mutation): what now?

It is usually a relief for family members who receive a negative result. However, if they have relatives who have been given a positive result, it can be extremely difficult. It may bring up feelings of guilt knowing you do not have the mutation whereas others in your family do.

For family members with a positive result, it may bring up thoughts of "Why me?" or "I wish I was the one without the gene". This is understandable even though they are also pleased for their relatives who tested negative. Being sensitive to each other's feelings is important and will help everyone involved feel less anxious.

Will my result affect my ovarian cancer treatment?

There are several drugs in use and in development that target cancers in women with BRCA mutations. Ask your specialist doctor about the results of your genetic test and the implications for treatment. You may be eligible to have treatment with PARP inhibitors or take part in a clinical trial.

PARP INHIBITORS

PARP is a protein in our body that helps damaged cells to mend themselves. Cancer cells with altered BRCA genes depend on PARP to keep their DNA repair mechanisms functioning. PARP inhibitors, such as olaparib, stop PARP from working, meaning the cancer cells can't mend themselves.

There are several PARP inhibitors at different stages of development and research.

Olaparib is for women who have a fault in a BRCA gene. Currently it is only for women who have had a recurrence of their cancer and have had a response to further treatment with platinum chemotherapy drugs. There is strong evidence olaparib maintenance can lengthen the remission in these women.

Recent research has shown promising results for using olaparib as a first-line treatment in women with a BRCA gene fault.

Results of genetic testing

Guidelines for the use of treatments are regularly developed and revised as new evidence becomes available.

Talk with your oncology team about the latest treatments. Ask your specialist doctor about your eligibility for olaparib and other PARP inhibitors.

PLATINUM-BASED CHEMOTHERAPY DRUGS

Research shows that women with ovarian cancer who carry a BRCA mutation may respond well to platinum-based chemotherapy. Chemotherapies are drugs used to help destroy cancer cells. Platinum chemotherapy uses platinum-based drugs to fight several types of cancer. It can have less severe side effects than certain other chemotherapy drugs.

Your specialist will be able to tell you whether platinum-based drugs are the best choice of treatment for you.

PARTICIPATION IN CLINICAL TRIALS

Treatment for ovarian cancer is constantly being improved through clinical trials. In clinical trials, new treatments are developed, evaluated and compared with current treatments. Participating in a clinical trial may provide individual women with a better outcome from ovarian cancer, as well as benefiting other women who may eventually receive the treatment being trialled.

Your doctor may suggest you take part in a clinical trial or ask if there is a trial you can be part of. Not every woman will be eligible for all trials. You will need to meet the guidelines for any proposed trial. If there is a trial suitable for you, speak with your healthcare team and the people close to you.

Talk to your doctor or go to the following websites for more information about research on PARP inhibitors and clinical trials in Australia designed for people with BRCA mutations or hereditary cancers:

- Ovarian Cancer Australia (www.ovariancancer.net.au) – search for clinical trials or call the helpline on **1300 660 334**
- Australia New Zealand Gynaecological Oncology Group (www.anzgog.org.au)
- Australian New Zealand Clinical Trials Registry (www.anzctr.org.au)
- Cancer Australia (www.australiancancertrials.gov.au).
- Cancer Council Australia has an excellent booklet, *Clinical trials for cancer* (www.cancer.org.au or **13 11 20**).

Results of genetic testing

BENEFITS OF GENETIC TESTING FOR WOMEN WITH OVARIAN CANCER

- May help you and your doctor take steps to reduce the risk of a future hereditary cancer diagnosis.
- Women who carry a BRCA gene mutation can respond better to platinum-based chemotherapy.
- Helps doctors to decide if a person is suitable for clinical trials.

BENEFITS OF GENETIC TESTING FOR FAMILY MEMBERS

- Empowers family members with knowledge about their risk for ovarian, breast and other cancer, allowing them to make an informed decision about their care.
- May help family members to take steps to decrease their risk of future cancers by having surgery, using preventive drugs or making lifestyle changes.

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

If you are told you have a BRCA1 or BRCA2 gene fault (mutation), your genetic counsellor will discuss ways you can reduce your risk of **breast cancer**. What you decide to do will depend on your current health status and what stage your ovarian cancer is. It is important to discuss your result with your specialist doctor, who will explain how it will affect you, including monitoring for any future cancers.

If one of your family members has been told they carry a BRCA1 or BRCA2 gene fault, and they currently do not have ovarian cancer, there are risk-reducing options they may consider. See the next section.

There are three options to think about:

- regular screening
- risk-reducing surgery
- drug treatment.

REGULAR SCREENING

Mammogram screening aims to detect tumours that are too small to be felt by you or your doctor. Breast screening will not stop women from developing breast cancer, but it will help detect tumours at an early stage, when they are easier to treat and can be curable.

BreastScreen Australia encourages women aged 50 to 74 to have a free mammogram every two years. However, women with a high risk of developing breast cancer, like women who have a BRCA1 or BRCA2 gene fault, can begin screening tests before the age of 50 and continue after the age of 74.

Women with a BRCA1 or BRCA2 gene fault may be offered yearly magnetic resonance imaging (MRI) scans, and sometimes a mammogram, from the age of 30. There is a Medicare rebate for annual breast MRI for women under the age of 50 who are at high risk of developing breast cancer. This includes women who carry a BRCA gene mutation. There is further information on the Department of Health website (www.health.gov.au).

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

Some clinics recommend breast surveillance from the age of 25.

Screening advice is tailored to an individual's family situation. Individuals may need different screening programs, which can include screening for other types of cancers, depending on the genetic fault found. Your specialist cancer team will discuss a monitoring plan with you based on your test results.

It is important to also have your breasts checked by your GP each year.

"My sister has opted for six-monthly breast checks and yearly screening whilst I decided to go ahead and have a hysterectomy (removal of my tubes and ovaries) and double mastectomy not long after finding out my result." – Lisa, age 45

Talk to your GP about the potential risks and benefits of breast screening. For more information you may like to read the Cancer Council fact sheet about the early detection of breast cancer (www.cancer.org.au).

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

RISK-REDUCING SURGERY (REMOVAL OF BOTH BREASTS)

If you have a BRCA gene mutation and are at a moderate to high risk of developing breast cancer, you can significantly reduce your risk (by 90–95%) by having surgery to remove both breasts. This operation is called a 'bilateral mastectomy'. This is often referred to by medical professionals as 'prophylactic' (preventive) or 'risk-reducing' surgery.

Your surgeon cannot give a 100% guarantee that you won't develop breast cancer. This is because it is impossible to remove every bit of breast tissue, and breast cancer may still grow in a tiny amount of remaining tissue.

> Based on an estimated risk reduction of 95%, if you are a BRCA carrier and have risk-reducing mastectomy, your lifetime risk of breast cancer after the surgery is about 4%. This is less than half the risk of women in the general population who don't have a BRCA gene fault and still have both breasts.

If you decide to have risk-reducing surgery, you can also have breast reconstruction surgery. This surgery 'rebuilds' your breasts using implants and/or tissue from another part of your body. It can be done at the same time as having the bilateral mastectomy or later as a separate surgery.

Before having risk-reducing surgery, carefully consider the physical and emotional impact it may have on you. Most women worry about:

- possible scars and their appearance (see the section titled 'Concerns about appearance')
- what others will think
- how the operation and possible side effects are going to make them feel.

Your medical team can discuss the advantages and disadvantages of the operation with you. Don't be afraid to ask questions. Seek further advice if you are unsure.

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

We recommend that you fully recover from your ovarian cancer treatment before having risk-reducing breast cancer surgery.

"I tested positive for the BRCA1 gene fault and immediately made the decision to have a bilateral salpingo-oophorectomy (BSO) to decrease the risk of future ovarian cancer. During this operation it was found I already had an early ovarian cancer, which was a huge shock for me. I had to have three-and-a-half months of chemotherapy after the operation. Shortly after chemotherapy finished, I made the decision to have a prophylactic bilateral mastectomy with reconstruction. The surgery went smoothly with no surgical complications. I did feel quite low after that final operation as there had been so much to face during the previous six months. But I knew I had to do the surgery as I could never live with the fear, and risk, of getting breast cancer after having been given an ovarian cancer diagnosis." – Christine, age 52

Lisa

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

CONCERNS ABOUT APPEARANCE

Understandably, a big concern for women who are thinking about having a risk-reducing mastectomy is what they will look like (sometimes known as the 'cosmetic' result).

"I didn't want any reconstructive surgery when considering my mastectomies and didn't think I would care about the cosmetic outcome as long as my risk was reduced. However, my surgeon persuaded me to have reconstruction given I was 35 years old, and I have to say I am so glad because my body image has remained more intact this way, making it easier to cope with." – Anne, age 39

For many women it is very important to maintain their body image. Research has shown that women who are offered nipple-sparing mastectomy are more likely to go ahead with the surgery.

For more information, see the Cancer Council booklet *Breast prostheses and reconstruction* (www.cancer.org.au). Although the information is for women who have breast cancer, it will be similar for women having a reconstruction after risk-reducing surgery.

"I would strongly recommend thinking it through carefully before making a decision about having surgery. Don't be afraid the cancer will get you before you have the surgery. You have enough time to make the decision, think about everything, the cosmetic effect and if you want to have children, even if it fails. Just think about every scenario." – Lisa, age 45

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

DRUG TREATMENT

Another way to try to reduce the risk of breast cancer is chemoprevention (prevention using drugs).

The drugs are called 'selective oestrogen receptor modulators' (SERMs). The two used are:

- tamoxifen for pre-menopausal women; tamoxifen is also used to treat breast cancer
- raloxifene for post-menopausal women; raloxifene is also used to treat osteoporosis (very weak bones) in post-menopausal women.

It is thought these drugs can reduce breast cancer risk by about 40%. Before you decide, your genetics doctor or genetic counsellor can discuss and give you written information on the risks and benefits of chemoprevention, including drug side effects and the extent of risk reduction.

Reducing breast cancer risk for women with ovarian cancer who carry a BRCA mutation

LIFESTYLE CHOICES

Women with ovarian cancer who are carriers of a BRCA1 or BRCA2 gene mutation should consider other factors that can affect their risk of developing breast cancer. These include:

- The oral contraceptive pill, which has long-term protective effects for the risk of ovarian cancer, but a short-term increased risk of breast cancer. This risk is reversible within five years of stopping the oral contraceptive pill.
- The use of hormone replacement therapy (HRT), which, depending on your clinical circumstances, may increase your risk of developing breast cancer.
- Reducing/limiting your alcohol consumption, quitting smoking and maintaining a healthy weight by eating a healthy diet and exercising regularly to reduce your risk.

Your doctor should advise you about the best way to reduce your risk of breast cancer by making changes to your lifestyle. They will also be able to advise you about whether your increased risk of breast cancer outweighs the benefit of taking HRT, and what alternatives are available.

"I don't want other people to be afraid. I don't want others to go through what my mum went through with her cancer and how that has affected our family. If I can stop this happening to me and for my future children – then why wouldn't I? I know everyone is different but if my story of getting tested and finding out I carry a BRCA gene mutation can give one person courage then it is worth it."
– Michelle, age 39

Michelle

Testing and counselling costs

A consultation with a genetic counsellor and genetic testing at a family cancer centre may be funded by the government. All family cancer centres are based in public hospitals. Ask your GP or specialist doctor for a referral to a family cancer centre nearest you.

For more information about family cancer centres, visit Cancer Council Australia website or phone the Cancer Information Helpline on **13 11 20**.

There are also several private genetic services in Australia.

Medicare funds testing for BRCA1 and BRCA2 gene mutations for women with breast or ovarian cancer who have a high probability of having a faulty gene. The test is also funded for relevant family members of women who are found to have a gene fault. Genetic testing for other faulty genes may not be covered by Medicare. Talk with your specialist doctor about this.

INSURANCE

If you have ovarian cancer and/or a positive BRCA1 or BRCA2 gene mutation, it may affect taking out any future insurance policies (but usually only income-related policies). You will need to discuss this with the insurance company.

You may also find the following links helpful:

- Watch the webinar Genetic testing and insurance discrimination on Pink Hope's website (www.pinkhope.org.au).
- Read fact sheet 20, *Life insurance products and genetic testing in Australia*, at the NSW Health Centre for Genetics (www.genetics.edu.au).

Finding information online

The internet has an enormous amount of information about ovarian cancer, genetic testing and hereditary cancer. We don't recommend using online information as a **substitute** for the information from your doctor and other members of your healthcare team.

Not all information online is accurate or will be suitable for you. While there are some excellent websites, some sites provide wrong or biased information. Focus on websites from reputable cancer organisations and universities. Look in the next section for links to further information and support or call our helpline on **1300 660 334**.

When you are unwell it can be overwhelming to try to sort through information. Ask a family member or friend to help if you are feeling unable to do this. Your medical team can also clarify whether the information you find on the internet is accurate.

A note on celebrities with BRCA gene mutations

Some celebrities who carry the BRCA mutation have made the decision to undergo prophylactic (preventive) surgery for breast and ovarian cancer. This always attracts a lot of attention worldwide, especially for women in a similar situation.

As we have discussed in detail in this booklet, Ovarian Cancer Australia recognises that preventive surgery for ovarian cancer, based on genetic testing results, is an extremely complex and emotional issue. We strongly recommend anyone thinking about this type of surgery to **seek expert advice**. It is extremely important to receive all the information you need to make an informed decision.

Women who are concerned about their genetic risk of ovarian cancer should discuss this with their GP or cancer specialist. They will explain where to get further advice and counselling if necessary.

Further information and support

More information on hereditary cancer and genetics can be found on the following websites:

- **Cancer Council Australia** (www.cancer.org.au) has information about family history and cancer and genetic testing. They also have a link to a list of all the family cancer clinics in Australia. You can also phone the Cancer Helpline on 13 11 20.
- **Cancer Council Victoria's** (www.cancervic.org.au) *Challenging choices* webinar discusses issues arising from being a carrier of the BRCA1 and/or BRCA2 gene fault.
- **Breast Cancer Network Australia (BCNA)** (www.bcna.org.au) has information on breast cancer in the family.
- The **Health Centre for Genetics Education** (www.genetics.edu.au) has many excellent fact sheets relating to cancer in the family, gene mutations, genetic counselling, breast and ovarian cancer and inherited predisposition.
- **Pink Hope** (www.pinkhope.org.au) has information about genetic risk as well as several excellent educational videos relating to hereditary cancer.
- The US website **FORCE** (www.facingourrisk.org) has more information about family history and hereditary ovarian cancer and you can sign up to their regular ebulletins.

If you would like information about dealing with the practical and emotional needs of having ovarian cancer, call Ovarian Cancer Australia on **1300 660 334**. We can provide information about support groups and networks that can help you connect with other women in similar circumstances. We can also suggest how you can connect with counsellors and psychologists who specialise in helping people with cancer.

If you have continuous feelings of sadness, anxiety and fears relating to your cancer or your cancer coming back, we strongly advise you to seek medical help. Asking your specialist or GP for a referral to a counsellor or psychologist can help with managing your fears.

Glossary

After a diagnosis of ovarian cancer, it may feel like you have a whole new language to learn. Medical professionals often use unfamiliar terms. Don't be afraid to ask them to explain any words or phrases you do not understand. It is important you feel sure about everything to do with your treatment and care. Here are some of the common terms you may hear and read.

ABDOMEN

The area between the ribs and hips containing the stomach, intestines, liver, gall bladder, bladder, kidneys and spleen. In women, the abdomen also includes the uterus, fallopian tubes and ovaries. The abdominal cavity is lined with the peritoneum.

BILATERAL MASTECTOMY

Surgical removal of both breasts.

BILATERAL SALPINGO-OOPHORECTOMY

Surgical removal of both fallopian tubes and both ovaries.

BIOPSY

Removing a sample of tissue so it can be examined under a microscope to help diagnose cancer or another disease.

BRCA1 AND BRCA2 MUTATION

Faults in two genes, known as BRCA1 and BRCA2, which are associated with a higher risk of developing breast, ovarian and some other cancers. These faulty genes can be passed from parent to child.

BREAST RECONSTRUCTION

Surgery to rebuild your breasts using implants and/or tissue from another part of your body. This surgery can be done at the same time as having a bilateral mastectomy or later as a separate surgery.

Glossary

CANCER
A disease where cells in the body behave and grow abnormally and form a malignant growth called a tumour. Malignant tumours can spread into other parts (tissues) of the body.

CHEMOTHERAPY
The use of drugs to slow the growth of or destroy cancer cells.

CELLS
The 'building blocks' of the body. A human is made up of billions of cells, which are adapted for different functions. Cells can reproduce themselves exactly, unless they are abnormal or damaged, as are cancer cells.

DNA (DEOXYRIBONUCLEIC ACID)
An extremely long chain of particles (molecules) containing all the information necessary for the life functions of a cell. It is our genetic coding.

EPITHELIAL OVARIAN CANCER
This is the most common type of ovarian cancer, accounting for nine out of 10 cases.

FAMILIAL
A condition/disease that happens more often in family members than is expected by chance alone.

FAMILY (OR FAMILIAL) CANCER
When a gene fault causes higher rates of particular cancer/s within a family. These cancers often affect family members at a young age.

FAMILY OR FAMILIAL CANCER CENTRE/SERVICE
A centre where genetic counsellors can talk to you about genetic testing if you have a family history of cancer and wish to consider testing for yourself, adult children or other family members.

Glossary

FAULTY GENE (GENE MUTATION)

A change in the structure of a gene. The gene fault (mutation) may be passed from parent to child during conception and can also be passed from generation to generation.

GENES

Small pieces of DNA: the material that acts as a 'master blueprint' for all the cells in your body. Your genes determine such things as what colour hair and eyes you have and how tall you are. If you inherit specific gene mutations, it may mean you have an increased risk of certain types of cancer.

GENETIC

Relating to genes. A genetic condition is one caused by a fault in one or more genes and may have been inherited.

GENETIC COUNSELLOR

A healthcare professional who is experienced in cancer genetics.

GENETICIST

A doctor who specialised in genetics.

HEREDITARY CANCER

When a gene fault linked to a particular cancer/s is passed from parent to child during conception. These are called 'inherited cancer genes'. You may develop a hereditary cancer from these genes. All cancer is triggered by faulty genes, but only 5–10% of cancers are hereditary.

HORMONE REPLACEMENT THERAPY (HRT) OR HORMONE THERAPY (HT)

Prescription medicines that supply the body with hormones that are no longer produced in the body after menopause. Used to help relieve menopausal symptoms.

HYSTERECTOMY

Surgical removal of the uterus (womb) without removal of the ovaries and tubes.

Glossary

LYNCH SYNDROME (ALSO KNOWN AS 'HEREDITARY NON-POLYPOSIS COLORECTAL CANCER SYNDROME' OR HNPCC)

Inherited faulty genes that increase the risk of colorectal, ovarian, endometrial, renal tract and other gastrointestinal cancers.

MUTATION

A change or alteration in a gene's DNA, resulting in a gene fault (mutation) that may be passed on to following generations.

OOPHORECTOMY

Surgical removal of one or both ovaries.

PROGNOSIS

An assessment of the possible future course and outcome of a person's illness.

RAD51C AND RAD51D

Inheriting faulty RAD51C or RAD51D genes has been found to increase the risk of developing ovarian cancer. These mutations are very rare, and together may contribute to less than 2% of ovarian cancers diagnosed.

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