



## **Genetic testing and hereditary ovarian cancer**

A guide for women with ovarian cancer  
and their families



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# Introduction

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If you have had a diagnosis of ovarian cancer, it is a common reaction to wonder what caused it, or worry that other family members may be at risk. This guide talks through what a family history of ovarian cancer means, whether your family members are at risk of developing hereditary cancer, whether you and your family should have a genetic test, and what the implications are for you and your family if you do have hereditary ovarian cancer.

The guide has been written by health professionals and experts in ovarian cancer and genetics. It has been developed with the help of women who either have hereditary ovarian

cancer themselves or are at risk and have been through many of the experiences you are facing.

We hope it provides you with sufficient information to make an informed decision about whether you should have a genetic test, and supports you and your family in dealing with the implications of the results of the test.

We understand how isolating it can feel to have an ovarian cancer diagnosis, which is why we provide a full programme of support and information across the UK. Please get in touch if you would like to know more, receive support or meet others going through similar experiences.







## Is my ovarian cancer hereditary?

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Cancer is a common illness so it is not unusual to have more than one family member with different types of cancer. However, in some cases families may be predisposed to certain types of cancer.

About 15 to 20 per cent of cases of ovarian cancer occur because of a genetic cause – a mutation in one or more genes known to increase the risk of ovarian cancer that has been passed on from your mother or father. Some types of ovarian cancer are more likely to be caused by a mutation in a gene than others.

## How do mutations in genes cause cancer?

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Genes are chemical instructions made from DNA, which tell the cells in our bodies how to work. Every cell in our body contains genes that determine and regulate our body's functions, growth and appearance. Each person has two copies of each gene, one inherited from their mother and one from their father.

In general, our genes enable our cells to function normally. However, sometimes the genes that were inherited have small changes, known as alterations or mutations. If one of our genes is mutated this can result in an increased risk of developing different illnesses.

A person with a gene mutation has a 50/50 (one in two) chance of passing it on to each of his or her children.

### **Mutation? Fault? Spelling mistake? Alteration?**

When a change occurs in an inherited gene it is called a mutation. You might hear it called a faulty or an altered gene elsewhere, but in this guide it is referred to as a mutated gene.

## What is meant by hereditary or familial ovarian cancer?

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'Hereditary' means the passing on of specific characteristics in your DNA from one generation to another – in this case, passing on a mutation in a gene. The term 'familial' means something that occurs or tends to occur in families.

'Familial cancer' or 'hereditary cancer' is a cancer which has developed as a result of carrying a mutation in a gene. Throughout this guide, we refer to it as hereditary ovarian cancer.

## Why should I consider if I have hereditary ovarian cancer?

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There are a number of considerations to think about if you find out you have hereditary ovarian cancer. Firstly, there's the impact on you – some hereditary ovarian cancers also increase your risk of developing other cancers, including breast cancer and so you would need to consider ways to manage this increased risk. It may also have some implications for the treatment of your ovarian cancer. Secondly, there could be an impact on family members who may also have inherited the gene mutation which could put them at increased risk of ovarian and breast cancer. This guide talks through these considerations and gives more information on each.





## How do I know if I have hereditary ovarian cancer?

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Women diagnosed with non-mucinous ovarian cancer in the UK, irrespective of family history, should be offered genetic testing for mutations in their BRCA1 and BRCA2 genes. Over 90 per cent of ovarian cancers diagnosed are non-mucinous. Some women may not be eligible, due to the specific type of cancer they have or the age at which they first developed ovarian cancer, but it is important all women discuss genetic testing with their GP or clinician to find out if testing is appropriate.

It's also important to note that mutations in genes such as BRCA1 and BRCA2 can occur in any family; however, they are more common in some communities. For example, members of the Ashkenazi Jewish population are approximately 10 times more likely to have a mutated BRCA1 or BRCA2 gene, and so are at greater risk of developing hereditary ovarian and/or breast cancer than people from the general population.

*"I had no previous family history of cancer, on either side of my family, other than an uncle who died of a bowel cancer. I didn't even know about possible familial risk until I attended a Target Ovarian Cancer Being Together day, after a question was asked to the expert panel."*

Shani





## Which genes are most likely to have a mutation which increases the risk of ovarian cancer?

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Hereditary ovarian cancer is most commonly caused by a mutation in either the BRCA1 or BRCA2 gene. Some mutations in other genes are also known to be associated with an increased risk of ovarian cancer however scientists have not yet found all of the genes associated with an increased risk.





## What are the BRCA1 and BRCA2 genes?

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The BRCA1 and BRCA2 genes were discovered in the mid-1990s in families that had a lot of cases of breast cancer (BREast CAncer genes). A mutation in the BRCA1 or BRCA2 gene accounts for the majority of hereditary ovarian cancer.

About 10–15 per cent of women who develop ovarian cancer have a mutation in either their BRCA1 or BRCA2 gene.

BRCA1 and BRCA2 genes are protective against cancer. However, mutations can occur or be inherited in these genes which can increase the risk of developing breast cancer (for both men and women) and increase the risk of developing ovarian cancer from two per cent (among women generally) to 30–50 per cent for BRCA1 and 10–25 per cent for BRCA2 gene mutations.

It is important to note that not everyone who inherits a mutation in the BRCA1 or BRCA2 gene will develop cancer, but it does increase the risk.



## Which other gene mutations put me at risk?

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Mutations in the BRCA1 and BRCA2 genes account for the majority of hereditary ovarian cancer cases. However, mutations in other genes can also increase risk of ovarian cancer:

- Lynch Syndrome (also known as hereditary non-polyposis colorectal cancer or HNPCC) is a type of bowel cancer predisposition which is linked to mutations in one of the genes **MSH2**, **MLH1**, **MSH6** and **PMS2**. A woman with a mutation in one of these genes has an estimated nine to 12 per cent chance of developing ovarian cancer at some point during her lifetime. The chance of developing cancer of the stomach, liver, kidney, bladder, skin and brain can also be increased by a mutation in one of the Lynch Syndrome genes.
- Mutations in **RAD51C** and **RAD51D** genes can increase a woman's risk of ovarian cancer. However these mutations are very rare. Women carrying a RAD51D mutation have about a 10 per cent risk of developing ovarian cancer by the time they reach the age of 80.
- Mutations in the **STK11** gene may also increase the risk of developing ovarian sex cord-stromal tumours – a different type of ovarian cancer. Mutations in this gene cause Peutz-Jeghers syndrome, which is an extremely rare condition estimated to affect one in 100,000 people. People with Peutz-Jeghers syndrome have an 18 per cent risk of developing gynaecological cancers by the age of 70.

- Mutations in the **BRIP1 (FANCJ)** gene increase the risk of ovarian cancer from approximately two per cent to six per cent. It is estimated that two women in every 2000 carry a mutation in their BRIP1 (FANCJ) gene. There is currently no routine test available to check for mutations in the BRIP1 (FANCJ) gene.

Mutations in different genes carry different increases in risk. A clinical geneticist (a doctor who specialises in genetics) or a genetic counsellor will be able to give you a more accurate and personalised risk assessment.

This guide focuses on information about the BRCA1 and BRCA2 gene mutations as this is what is most commonly tested for at the moment. Testing is currently in development for a number of other genes.





## Getting tested for a BRCA gene mutation



### What should I do if I think I have hereditary ovarian cancer?

If you have been diagnosed with ovarian cancer and not offered genetic testing, arrange to talk with your oncologist and discuss whether you may be eligible.

As guidance on genetic testing is relatively new, in some areas of the UK testing may not be automatically offered. If you haven't been offered a test, you may still be able to ask for a referral to a genetics centre via your GP or oncologist. You can find your nearest centre here – **[bsgm.org.uk/information-education/genetics-centres](https://www.bsgm.org.uk/information-education/genetics-centres)**

You will then be offered the chance to speak with a genetic counsellor to help you make a decision about whether to have the test or not, and what the implications would be for you and your family members.



## What should I consider before having a genetic test?

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There are some important things to think about before going ahead with genetic testing; in particular you should think about the impact of the testing on yourself and on those who are close to you.

## Considerations for me

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Some women find it helpful to have an explanation for why they developed ovarian cancer. However, there will be other implications for you if you are identified as having a hereditary cancer risk as you will have an increased risk of developing other cancers. In the case of the BRCA1 or BRCA2 genes you will have a higher risk of developing breast cancer (in comparison to the general population). What is recommended for you in terms of addressing this risk will be discussed in the context of your current cancer diagnosis and treatment. There is more information about this later in this guide.

The results of genetic testing may also have implications for the treatment you are offered for ovarian cancer. There is more information about this later in the guide.

## Considerations for my family

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A second consideration is the impact on your family. Other members of your family may also have the gene mutation if you do; including your mother or father, siblings and your aunts, uncles and cousins, depending on side of the family the mutation is passed down through. The children of someone with a BRCA1 or BRCA2 gene mutation have a 50 per cent (one in two) chance of having inherited it and therefore being at increased risk. They also have a 50 per cent (one in two) chance of having inherited a normal copy of the gene from the parent with the gene mutation.

Women with a BRCA1 or BRCA2 gene mutation have a high risk of developing breast and ovarian cancer and men may have an increased risk of developing prostate cancer and male breast cancer. There may also be a small increased risk of pancreatic cancer for men and women who carry BRCA2 gene mutations.



There is information later in this guide about discussing your result with your wider family.

Due to the potential implications, it can be very helpful to discuss genetic testing with your relatives at an early stage. If you are seen by your local Clinical Genetics Service prior to deciding about genetic testing, they will cover these issues with you in more detail.

### **When should I have genetic testing?**

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If you are eligible for a genetic test, it is important to ask yourself: “when is the right time for me to have genetic testing?” There is evidence that having genetic testing soon after a cancer diagnosis can be more distressing. If you feel that it is not the right time to have genetic testing now, you may choose to revisit it at a later stage. Some women have DNA stored (from a blood sample) so that it is available for genetic testing at a later date.

*“I felt that I was on a hamster wheel of cancer since being diagnosed. It’s hard to keep your head in gear while being practical the whole time.”*

*Berni*







## What is a genetic test and what are the possible results?

.....

There are a few stages to having a genetic test. Your specialist will refer you to the local genetics centre. You will then receive an appointment to attend your nearest genetics centre, at which point you will be asked to complete a family history form. At the appointment a clinical geneticist or genetic counsellor will discuss with you what is involved in a test and what the implications will be for you and your family.

If you wish to go ahead, giving consent to the test, you will then have a blood test. You will be given a choice of how you'd like to receive the results – whether by phone or in writing, or sometimes face to face.

The time it takes for the blood test to be analysed and the results to be available varies slightly between the genetics centres and you will be advised how long it will take for yours. It is usually between six and ten weeks.

When you are informed about your test result, you will be invited to speak with the genetics teams again to explain the result and what the next steps are.

## What are the possible results of a genetic test?

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

- 1 Mutation present – sometimes called a positive test result:** Testing identifies a mutation that causes a significant change to the gene and is therefore very likely to be the cause of your cancer.
- 2 Variant of unknown significance (VUS) – sometimes called an inconclusive test result:** Testing may identify a mutation or an alteration in the gene where it is uncertain whether or not this would cause an increased cancer risk. This is known as a variant of unknown/uncertain clinical significance because it is not clear if it causes significant changes to the gene.
- 3 Mutation not present – sometimes called a negative test result:** Testing has not identified any gene mutations.

"At the first meeting, they talked about the likelihood of it being genetic, and asked me to complete the family history forms. Because an aunt on my dad's side had died of ovarian cancer, and another of breast cancer, the counsellor said this indicated a strong link."

Berni











## What is the impact of the different results for me and my family?

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### **What is the impact of ‘mutation not present’?**

Some people can feel relief and reassurance if they are not found to have a genetic cause for their cancer.

If you have this result, and do not have a family history of ovarian or breast cancer, it is most likely that your ovarian cancer was ‘sporadic’ or a one-off in your family. It means your risk, and the risk of your children, of developing breast cancer and other cancers associated with the BRCA1 and BRCA2 genes is probably the same as the general population.

If you have this result, but have a strong family history of ovarian or breast cancer, the result can be confusing and less informative. Given that scientists still do not know all the mutations in different genes that may increase a woman's risk of ovarian cancer, it is possible that you have a mutation in a different gene not detected by this test. This means there is still a level of uncertainty even after genetic testing.

### **Can my family members still receive a genetic test?**

Although the result makes it less likely that a very strong predisposition to cancer runs in your family, close relatives could receive risk reducing advice, based on the family history.

The results of genetic testing may also have implications for the treatment you are offered for ovarian cancer. There is more information about this later in the guide.

*"My test came back negative which was good but also left a lot of questions unanswered for me and my relatives. I still have a strong family history which leaves us wondering why and what the unknown possible risks are."*

Sue

## **What is the impact of 'variant of unknown significance' (VUS)?**

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If you have been told the result showed a genetic variant of unknown significance (or VUS), this means that the test has found a gene mutation or alteration in the BRCA1 or BRCA2 gene where it is not known if it causes an increased risk of ovarian cancer or not. The number of women who are given a VUS result is decreasing as scientists learn more and more about the different mutations that increase the risk of ovarian cancer. It may also mean that your VUS is reclassified over time. As more is discovered about genes, there may be further information about the VUS in the future and you may like to contact your Clinical Genetics Service to discuss this in a few years' time.

### **Can my family members still receive a genetic test?**

Based on a VUS, predictive genetic testing would not be available for the rest of the family. However, close relatives could receive risk reducing advice, based on the family history.

If a VUS is identified, it may create a higher level of uncertainty for a family. You may find the uncertainty very difficult to understand or deal with emotionally, as it is less clear whether you and other family members have an increased risk.



It is, therefore, important that you ask your doctor or genetic counsellor for support and advice about the options available to you.

## What is the impact of 'mutation present'?

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If you've been told there is a known mutation present, you will be told whether you have a mutation in your BRCA1 or BRCA2 gene. The result will have an impact of your risk of developing other cancers, the risk of your family members developing cancer, and it may also have implications for your treatment. You will receive additional counselling to explain the impact of this result.

"I was very stunned. After I'd completed the family history form I'd been told I was at a very low risk of having the gene mutation so didn't have any counselling about what the impact of a positive test would be."

Shani

"The genetic counsellors were brilliant and very sensitive in the way they approached it. They offered further support and counselling."

Berni

## How might I feel?

"It adds another dimension to the journey, just when you think you're through it and can get off the wheel, finding out about BRCA throws you off kilter. It's the hardest part to deal with because you know cancer is not going away."

Berni

Naturally people can have very different feelings and reactions when they find out they have a gene mutation that explains their cancer diagnosis. Initially many women feel shock, anger or anxiety. Some people expect to have a gene mutation and are glad to have an explanation for the cancers in their family.

"I feel like I've been diagnosed all over again, which sounds dramatic, but I was really shocked. I got over this shock by reassuring myself that at least my children would know about their risk."

Shani

"If I'm honest, knowing I have a faulty gene has given me a sense of relief because it helped explain why I was unlucky to get cancer twice. But then I realised the implications for my family."

Adele

With time, most women adjust to this new information. Additionally, many are pleased that although it does not change their own diagnosis, there is important information available for other family members.

"My sister pointed out that now she can be monitored and will not have to go through the same as me."

Berni

It is important for women to remember that they cannot control the genes they inherit from their parents or the ones they pass on to their children.

"I felt a bit guilty that I could have passed this on to my children, but you can't choose what genes you pass on."

Shani

You will have the option of a follow up appointment with your local Clinical Genetics Service to further discuss how you are feeling and the impact the result can have on you and your family. They will be able to provide you with further information and direct you to where you can access further support.



*"When I was diagnosed 15 years ago as being a carrier of the BRCA1 gene mutation following my ovarian cancer diagnosis, I wasn't offered information about preventative surgery as less was known about it then. I did unfortunately go on to be diagnosed with breast cancer 10 years later, and now get regular follow ups. The clinicians are always approachable and I have their phone numbers if I'm ever worried over anything. I had a recent scare and called them, and was seen by the clinician the same day."*

*Diane*

## **What is the impact on my treatment and my risk of other cancers?**

For women with ovarian cancer who have a mutation in the BRCA1 or BRCA2 genes, the risk of developing breast cancer is also increased. For a woman in the general population the lifetime risk of developing breast cancer is about 12 per cent (one in eight). If she has a BRCA1 or BRCA2 mutation, the risk is around 50 to 80 per cent. Remember, an increased risk does not mean you definitely will develop breast cancer. There are a number of risk management options which we talk more about later in the guide.

There is ongoing research into new treatment options for women with ovarian cancer who carry known mutations in the BRCA1 or BRCA2 gene. Studies indicate that the prognosis for BRCA1 or BRCA2 carriers can be better than for those with no gene mutation because carriers have an increased responsiveness to specific therapies. We talk more about the impact having a BRCA1 or BRCA2 gene mutation has on your treatment later in this guide.



## What is the impact on my family members?

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### What is predictive testing?

If your test for a BRCA1 or BRCA2 gene mutation is positive (mutation present), your relatives can have the predictive genetic test to see if they have the same mutated gene. It is 'predictive' because a positive result for them means they have an increased risk of developing cancer. It doesn't mean they have cancer or are definitely going to develop it.

Having a mutation in your BRCA1 or BRCA2 genes may also have important health and social implications for family members, including future generations. If you have been found to have a gene mutation, genetic testing (known as predictive testing) will be available to adult members of your family who wish to know whether or not they have inherited the gene mutation.

- Your siblings will have a 50 per cent chance of also carrying the BRCA gene mutation, and will be able to access predictive testing.
- Your children will also have a 50 per cent chance of inheriting the BRCA gene mutation. Testing is only available to adults over the age of 18 because the risk management options are either not available or not yet necessary for those under 18 and it is important that they

make their own informed choice about whether to be tested.

- The mutation in your BRCA1 or BRCA2 gene will have been inherited from your mother or father. If they are unaware of their BRCA status, they can access advice about testing, as can their siblings, nieces and nephews (your aunts, uncles and cousins).
- It's important to note that both men and women who inherit a BRCA1 or BRCA2 mutation, whether or not they develop cancer themselves, may pass the mutation on to their sons and daughters.
- Remember, not all individuals with a BRCA mutation will develop cancer.

*"My mum had never had cancer, and wasn't aware of BRCA until I got ovarian cancer and was tested. She subsequently also tested positive and at the age of 75 was diagnosed with breast cancer – caught and treated very early because she was put on a screening programme because of her BRCA status."*

*Sharon*

## How do I tell them and how might they feel?

If your relatives are aware that you are having genetic testing, a conversation about the results may arise naturally. Depending on how you are feeling (physically and emotionally) it can feel like a difficult discussion to have. It can be particularly challenging if there are young family members you want to share the information with. Please remember you are not alone. Some women find that confiding in their partner or a friend can be beneficial. Your local Clinical Genetics Service is also available to provide support.

In many cases women find that their relatives are wondering whether they may have an increased cancer risk and are very accepting of the news about a gene mutation. It means they are able to have a predictive genetic test that can allow them to take action if they are at increased risk.

Many people choose to have predictive genetic testing because they would rather be aware of an increased risk of cancer and take up additional screening and/or surgical options that are available. However, knowledge about a possible increased cancer risk may cause anxiety for relatives and some people are concerned about the possible impact on their work or insurance. There is more information about this later in this guide.

*"I was really worried for my children, but relieved to know they can be tested and be alert for the signs. I was open with my family the whole way through about what it might mean for them."*

Shani

*"When I was first diagnosed as a BRCA gene carrier, I wanted my daughter tested straight away, but was counselled against it as she was only 13. They said she should be at least 18 and should be fully aware of the implications of the possible results."*

Adele



Some women find they have relatives who are resistant to genetic testing and who prefer not to receive the information. Some have relatives they are not in contact with. These situations are challenging and may be distressing. Your local Clinical Genetics Service will be able to discuss this with you. If you think it will be helpful, they can provide an 'open letter' that you can send to relatives that explains about the gene mutation and the genetic testing available to them.

*"I was given a letter by my genetic service to share with my brothers and children explaining the gene and about being tested."*

*Shani*

Ultimately, whether and when your relatives decide to have predictive genetic testing is up to them. People are all different and some adjust to the news more quickly than others. You will have given them an option that may not otherwise have been available to them.



*"All my family are aware, and some have already been tested. My three daughters have had very different reactions. One is finding it difficult to cope with cancer deaths in the family and then my subsequent diagnosis, but says she will get round to getting tested. One does not want to be tested, and did not even want to know the result of my test, but I felt it was important she had all the information. One has been tested and also carries the BRCA1 mutation, and says her team have been fantastic. She's been offered counselling and will be talking to them about preventative measures soon. My eldest sister is reluctant to be tested – she says she 'hasn't gotten around to it yet'."*

*Berni*

There is a section later in this guide on implications for a family member who are also found to have a BRCA1 or BRCA2 mutation.







## I have a BRCA1 or BRCA2 gene mutation – what now?



### How can I manage my increased risk of breast cancer?

Your genetic counsellor will discuss with you the different ways of reducing your risk of developing breast cancer.

There are three options to consider – **screening, risk reducing surgery and drug treatment**. The choice will take into account your current health and the prognosis of your diagnosis of ovarian cancer.

## Screening

The NHS runs a breast screening programme for women throughout the UK. Women between the ages of 50 and 70 are typically invited for breast screening every three years, but women at high risk can access screening tests before the age of 50 and after the age of 70. Women with a BRCA1 or BRCA2 gene mutation will be considered for yearly MRI scans and mammograms from the age of 30 onwards. Ask your genetic counsellor or GP for further information. 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.



## Risk-reducing surgery – bilateral mastectomy (removal of both of your breasts)

For women with a BRCA gene mutation, having surgery to remove both your breasts will greatly reduce your risk of developing breast cancer, but cannot guarantee completely that you will not develop breast cancer. You will be offered the option of having reconstructive surgery to rebuild your breasts using implants and/or tissue from another part of your body. This may be carried out at the same time as your operation, or it may be done at another time as a separate surgery.

Surgery will have a very big impact on you both physically and emotionally, especially following an ovarian cancer diagnosis. It is important that you take your time and discuss all the advantages and disadvantages with a counsellor and your medical team before making a decision. Your medical team will recommend that surgery is not advisable until after you have recovered from your ovarian cancer treatment.

“My counsellor explained about the increased risk of breast cancer and that I should be screened every year.”

Shani

*"As I'm still dealing with the aftermath of cancer treatment, the counsellors and clinical team have agreed to discuss preventative surgery in a few more months as I feel I need to take a breath. I think I will have it, but haven't decided about reconstructive surgery – I have an appointment with the plastic surgeon to help me decide."*

*Berni*

## **Drug treatment and lifestyle choices**

In some cases, 'chemoprevention' may be considered to reduce the risk of breast cancer. Chemoprevention is the use of drugs, vitamins, or other agents to try to reduce the risk of, or delay the recurrence of, cancer. This includes treatment with drugs such as tamoxifen and raloxifene. Tamoxifen is usually used as a treatment for breast cancer, and raloxifene is used to treat or prevent osteoporosis (bone thinning) after the menopause. Your genetics doctor or genetic counsellor can discuss this with you and give you written information on the absolute risks and benefits of chemoprevention, including the side effects of the drugs, and the extent of risk-reduction before you make a decision.

Women with ovarian cancer who are carriers of a BRCA1 or BRCA2 gene mutation should be offered advice about other factors which may affect their risk of breast cancer. These include:

- The use of the oral contraceptive pill which has long term protective effects for the risk of ovarian cancer but a short term reversible increased risk of breast cancer.
- The use of hormone replacement therapy (HRT) which depending on your clinical circumstances may increase your risk of developing breast cancer. Your clinician should discuss with you whether the increased risk outweighs the benefit, and what alternatives there are.
- Reducing your alcohol consumption, stopping smoking, and maintaining a healthy weight through healthy eating and exercise to reduce your risk.

## **What are the implications for my treatment for ovarian cancer?**

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There are a number of drugs in use and in development that are targeted at cancers in women with mutations in one of their BRCA genes so it is important to ask your clinician if your genetic test result has an impact on your ability to access different treatments or clinical trials.

## PARP inhibitors

PARP inhibitors are a group of drugs which work by stopping cancer cells repairing themselves. There are currently a number of PARP inhibitors at different stages of development and research.

The PARP inhibitor called olaparib (also known as Lynparza) has been licensed to treat ovarian, fallopian tube, and primary peritoneal cancer in women who carry mutations in the BRCA1 or BRCA2 genes and whose cancer has come back. However, access to olaparib is different across the UK, and it depends on the number of recurrences of ovarian cancer you have had so you will need to have a detailed discussion with your clinician about your eligibility for this and other PARP inhibitors.

## Participation in treatment clinical trials

The best time to look into participating in clinical trials is when you are first diagnosed with cancer or when you have a recurrence before starting treatment. Some studies have restrictions on the number or the types of prior cancer treatments that a person has received. Talk to your clinician or visit Target Ovarian Cancer's Clinical Trials Information Centre – [clinicaltrials.targetovariancancer.org.uk](https://clinicaltrials.targetovariancancer.org.uk) – for information about research on PARP inhibitors and other studies specifically designed for people with BRCA mutations or hereditary cancers.

*"I was diagnosed with ovarian cancer in 2009, and subsequently tested positive for a BRCA1 mutation. Following surgery and chemotherapy, I had about 10 months of remission before the cancer returned. I then joined a clinical trial for a PARP inhibitor rather than have more chemotherapy. I was happy to be generally living a normal life with no nasty side effects; my cancer shrunk to "non-measurable disease," and my CA125 tumour marker went from 204 to 10. I am very grateful for the trial that gave me a fantastic year without chemo. Although the drug stopped working for me after 10 months, it is still working for others, and I am hopeful that my involvement helps others (I have two BRCA-positive daughters)."*

*Sharon*



## Tumour profile testing

All tumours have genetic mutations that arise as the tumour develops. These are called 'somatic' mutations and are not present in the normal healthy cells of a person who has cancer. The inherited mutations referred to throughout the rest of this guide which may increase the risk of developing cancer are known as 'germline' mutations.

When a tumour is removed during a biopsy or surgery, the tissue will be sent to a pathologist who will study the tissue under a microscope and arrange additional tests on the tissue to profile it. This tumour profiling test gives information about the cells in the somatic mutations, and can help the medical team identify which treatments the tumour is most likely to respond to, and in some cases whether the woman is eligible for certain clinical trials.







## What are the implications for a family member with a BRCA gene mutation?

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### **Risk reducing options**

If a family member has predictive testing and been found to carry a gene mutation, they will have had genetic counselling to discuss the implications and risk management options.

We have already outlined in the previous section what the options are for breast cancer. However, women may also want to consider risk reducing options for ovarian cancer.



"My sister was diagnosed with breast cancer about a month before getting her BRCA result. She took immediate action, with a bilateral mastectomy and reconstruction. A few months after completing treatment, she had her ovaries removed, minimising her risk of having ovarian cancer. Six years later she remains cancer free."

Adele

### **Removal of both ovaries and fallopian tubes – bilateral salpingo-oophorectomy**

This operation will greatly reduce the risk of developing ovarian cancer and may, to a lesser extent, reduce the risk of breast cancer, although the evidence is unclear about this. There is still a small chance of developing ovarian cancer after the surgery because microscopic cancer cells invisible to the naked eye may have started to grow in the abdomen or pelvis before the ovaries are removed, and these cells will not be removed during the operation.

The operation gives the greatest benefit if it is carried out before your relative goes through the menopause. Removal of both ovaries will mean the immediate start of the menopause. Their medical team will discuss with them the advantages, disadvantages and risks of taking hormone replacement therapy (HRT) to manage the side effects of early menopause.





## Contraceptive pill

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. Using the contraceptive pill can reduce the risk of developing non-mucinous ovarian cancer (over 90 per cent of ovarian cancers diagnosed are non-mucinous), by as much as 50 per cent. Although the contraceptive pill has been shown to reduce the risk of ovarian cancer, it is not suitable for all women and it can slightly increase a woman's risk of developing breast cancer, although this risk is reversible within five years of stopping the oral contraceptive pill.

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

## Other implications

### Family planning

The genetic counsellor will be able to provide more information to your family members about having children and the impact on the children they've had already.

The family planning options for men and women with a BRCA mutation are:

- To have children without any intervention – each child would have a 50 per cent chance of inheriting the mutation
- To not to have children at all because of the risk of passing on the BRCA gene mutation
- Adoption
- Egg or sperm donation
- Prenatal testing – some couples may choose to get pregnant naturally and have a test during the pregnancy to see if the baby 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 fertilize in a test tube (IVF). When the embryos reach a certain size (at a few days old), a cell is removed and is tested for the hereditary disease. Once the genetic status is determined, the parents can decide which embryos they want implanted.

Your family members may want to discuss their options with their geneticist who can explain the implications and the funding options available, but it's important to note that there is no right or wrong answer.



*"My niece plans to have surgery on completion of her family."*

*Adele*

"I was originally told about the possibility of using PGD to have children early on when talking to a genetic counsellor about having the BRCA mutation. The genetic counsellor also told me about the other options including having children naturally in the hope that they would be born without the gene or conceiving naturally then having the embryo tested at 12 weeks. It was through her that I was referred onto a hospital that carries out PGD where I was further informed about PGD in detail.

All parents want to protect their children from harm and although I am aware that PGD can't get rid of all the health risks to my unborn child, it does mean that my child won't have the genetic make-up that puts it at such a high risk of breast and ovarian cancer. I also don't want them to have to make the decisions I've

had to make on whether to have risk reducing surgery or not, or for them to have to decide on how to have children or for them to worry about it. For me and my family using PGD means that the gene fault stops with me; future generations of my family will never have to worry about a gene that puts them at such high risk of cancer."

Carly

## Insurance

An agreement is in place which guarantees that anyone who has had a predictive genetic test (eg for breast and ovarian cancer), can take out life and critical illness insurance cover without disclosing the results. This agreement, known as the Concordat and Moratorium on genetics and insurance, has been in place since 2001 and has been extended until 2019.

However, you should check the **Association of British Insurers (ABI)** for the latest guidance at [abi.org.uk](http://abi.org.uk) or on 020 7600 3333.











## Further sources of support

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### **More from Target Ovarian Cancer**

This guide is for women who have been diagnosed with ovarian cancer and who would like to know more about hereditary ovarian cancer.

If you would like more information on dealing with the practical and emotional needs of ovarian cancer we have a lot of information on our website and a range of information guides for women offering advice and support.



- **What happens next?** – a guide for women with a recent diagnosis of ovarian cancer.
- **Back here again** – a guide for women with recurrent ovarian cancer.
- **Looking after me** – a guide for women living with terminal ovarian cancer.
- **A younger woman's guide to ovarian cancer** – information on treatment, relationships, fertility and facing an early menopause.

All of our guides can be downloaded from our website [targetovariancancer.org.uk/guides](https://targetovariancancer.org.uk/guides) or sent out as a printed guide.

## Other sources of support and information

Genetic Alliance UK

[geneticalliance.org.uk](https://geneticalliance.org.uk) | 020 7704 3141

Macmillan Cancer Support

[macmillan.org.uk](https://macmillan.org.uk) | 0808 808 0000

Cancer Research UK

[cancerresearchuk.org](https://cancerresearchuk.org) | 0808 800 40 40

Breast Cancer Care

[breastcancercare.org.uk](https://breastcancercare.org.uk) | 0808 800 6000

Breast Cancer Now

[breastcancernow.org](https://breastcancernow.org)

[familyhistory.breastcancernow.org](https://familyhistory.breastcancernow.org)

## BRCA specific support

BRCA Umbrella Support and Social Network:  
an online support forum for BRCA carriers  
[brcaumbrella.ning.com](https://brcaumbrella.ning.com)

FORCE: a support group for BRCA carriers based  
in America and in Essex UK

[facingourrisk.org](https://facingourrisk.org)

[facingourrisk.org/essex](https://facingourrisk.org/essex)

The National Hereditary Breast Cancer Helpline  
and Information Centre

[breastcancerogenetics.co.uk](https://breastcancerogenetics.co.uk)

Helpline: 01629 813 000

There are several BRCA peer support groups on  
Facebook – [facebook.com](https://facebook.com)

## Websites for further information:

Association of British Insurers (ABI)

[abi.org.uk](https://abi.org.uk) | 020 7600 3333

National Institute for Health and Care Excellence  
(NICE guidelines for familial breast cancer)

[nice.org.uk/Guidance/CG164](https://nice.org.uk/Guidance/CG164)

## About Target Ovarian Cancer

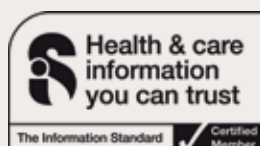
Target Ovarian Cancer is the UK's leading ovarian cancer charity. We work to:

- improve early diagnosis,
- fund life-saving research,
- provide much-needed support to women with ovarian cancer

We're the only charity fighting ovarian cancer on all three of these fronts, across all four nations of the UK. Ovarian cancer can be devastating. But there is hope – Target Ovarian Cancer. We work with women, family members and health professionals to ensure we target the areas that matter most for those living with ovarian cancer.

As an Information Standard accredited organisation all our publications and information are subject to an information production system that ensures quality and impartiality. All our publications are peer reviewed by experts in their field, health professionals and those affected by ovarian cancer.

We hope that you have found this publication useful, if you have any comments or suggestions please do let us know.



To access our list of references we used for this publication please contact us directly.

We make every effort to ensure that the information we provide is accurate. If you are concerned about your health, you should consult your doctor. Target Ovarian Cancer cannot accept liability for any loss or damage resulting from any inaccuracy in this information or third party information on websites to which we link.





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