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# A beginner's guide to *BRCA1* and *BRCA2*



Patient Information



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## Introduction

### What does it mean to have a *BRCA* gene mutation?

This information booklet was put together to provide answers to some of the many questions people have when they receive the news that they are *BRCA* gene mutation carriers. Healthcare professionals working in Cancer Genetics have met many people who are *BRCA* carriers and we can share our experience to some extent. We can provide information and make referrals to other healthcare professionals. We can tell you that other people in your situation have had similar feelings and anxieties but we do not truly know what it is like for you and how it may affect your life.

Many people have questions that, for the time being, cannot be answered fully because we are still studying the *BRCA* genes and their impact. One of the challenges that *BRCA* carriers face is coping with the uncertainty of what the future holds, both for you and your family. You may be asked to make life-changing decisions about cancer screening and risk-reducing surgery without the benefit of all the facts you would like. This may be difficult and could cause you to feel quite anxious, frustrated or even angry at times. It may take you some time to feel that you have absorbed enough information to feel confident about your decisions.

Unlike other health-related issues, there is an extra set of challenges for *BRCA* gene mutation carriers – the risks for your relatives. If you are the first person in the family to have been diagnosed with a *BRCA* gene mutation, you will have to find a way to share this information with your family. Even if you are not the first person in the family to be tested, there may be family pressures to face. Will relationships be affected if one person has a positive gene test and their relative has a negative result? What about starting a new relationship – when do you tell someone and what do you say? What if you have young children or are planning a family?

We hope that this guide will be useful in providing you with some of the information you need and also help you to think about questions you may wish to discuss with your family and the genetics department. If you have any comments about the guide, do let us know. We would like to make this booklet as useful and accessible as we can to support you and your relatives.



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## Background information about the *BRCA1* and *BRCA2* genes

### Is breast cancer inherited?

Generally speaking, cancer is not an inherited illness. Breast cancer is common, affecting about one in eight women at some point during their life, more commonly after the menopause. It is not unusual to have a relative who has been affected by breast cancer. Most breast cancer occurs sporadically or out of the blue. However, we know that faulty genes are the underlying cause for about 5–10% (5 to 10 in 100) of breast cancers. There are two genes that, if mutated, greatly increase an individual's chance of developing breast and ovarian cancer. These are known as the *BRCA1* (Breast Cancer 1) and *BRCA2* (Breast Cancer 2) genes. These two genes account for approximately a fifth (20%) of familial breast cancers. Research to identify new genes that also contribute to a high risk of breast cancer or modify the risk associated with a *BRCA1* or *BRCA2* mutation is ongoing.

### What are breast cancer genes?

Genes are pieces of the DNA code that we inherit from our parents. We have two copies of each gene – one copy is inherited from our mother and one from our father. We have approximately 20,000 genes, each with a specific function that helps our bodies grow and function normally. Some genes work to protect against cancer by correcting damage that can occur in the DNA during cell division. *BRCA1* and *BRCA2* are two examples of these tumour suppressor genes. If an individual has a mutation in a *BRCA1* or *BRCA2* gene, they have a greatly increased risk of developing breast and ovarian cancer.

The *BRCA1* and *BRCA2* mutation does not cause cancer to occur on its own. The individual is at greater risk of developing cancer because their cells' ability to repair DNA damage may be affected by the *BRCA1* or *BRCA2* mutation. It is the build up of DNA damage which causes a normal cell to change into a cancerous cell. We do not yet fully understand the whole process that changes a normal cell into a cancerous cell. We also do not yet understand why the *BRCA1* and *BRCA2* mutations primarily give a high risk of breast, ovarian and prostate cancers, rather than other types of cancer.

## What cancer risks are associated with *BRCA1* and *BRCA2*?

Gene	<i>BRCA1</i>	<i>BRCA2</i>
Breast cancer in unaffected women (up to age 80)	60–90%	45–85%
Women with breast cancer (unilateral)	50%	50%
Lifetime risk of a new cancer in the other breast	5 year risk of new breast cancer ~10%	5 year risk of developing a new breast cancer ~5–10%
Ovarian cancer, lifetime risk	40–60% Risk increases from age 40	10–30% Risk increases from mid-late 40s
Male breast cancer, lifetime risk	0.1–1%	5–10%
Prostate cancer, lifetime risk	~10% Similar to population risk	20–25%

For further information please refer to the ICR/RMH BRCA carrier management guidelines:

[www.icr.ac.uk/Media/research-divisions/protocol-3-brca-mutation-carrier-20150209-v4.pdf?sfvrsn=2](http://www.icr.ac.uk/Media/research-divisions/protocol-3-brca-mutation-carrier-20150209-v4.pdf?sfvrsn=2)

## Managing breast cancer risk

Women who are carriers of a *BRCA* gene mutation have an increased risk of developing breast cancer during their lifetime.

### What is my risk compared with other women?

Women in the UK have a one in eight or 12.5% chance, on average, of developing breast cancer in their lifetime. Women with one or two close relatives affected with breast cancer have a lifetime risk of 17–30% depending on the specific family history.



Women who have a *BRCA1* gene mutation have a 60-90% lifetime risk and women who have a *BRCA2* gene mutation have a 45–85% lifetime risk. Women with *BRCA* mutations have an increased risk of developing breast cancer at a younger age. Also, *BRCA* mutation carriers who have had breast cancer have a higher risk of developing another new breast cancer, compared to women in the general population who develop sporadic breast cancer.

## **How can I manage my risk of developing breast cancer?**

There are two approaches to managing your risk. You can choose to have cancer screening such as mammography to try to detect breast cancer at an early stage so it is possibly easier to treat. You can choose to have risk-reducing surgery to remove your breast tissue, before the diagnosis of breast cancer. Screening does not prevent or reduce the risk of breast cancer. Surgery has potential side effects that need careful consideration. Many women choose to have screening initially and some may also consider risk-reducing surgery.

There is no right choice. Everyone has to decide which option is best for them. We can help you make that decision by ensuring you have accurate information and by referring you to speak to the appropriate specialists.

## **Early detection: Breast cancer screening**

### **How is breast screening organised for *BRCA1* and *BRCA2* carriers in England?**

Women with *BRCA1* and *BRCA2* mutations are offered annual breast screening from age 30 through their local NHS Breast Screening Programme (NHSBSP) centre. The NHSBSP have always delivered breast screening to women over the age 50 in a population screening programme. Since April 2013 they have been funded to deliver the screening to higher risk women including *BRCA1* and *BRCA2* mutation carriers. Mutation carriers are referred by their Genetics unit to the specific NHSBSP centre which is funded to cover their postcode. Women cannot choose which centre they want to have their screening at nor can they request to have their screening at a hospital that is not an approved NHSBSP centre, such as The Royal Marsden.

## What breast cancer screening is recommended for *BRCA* mutation carriers?

### Breast screening

Age	Annual mammography	Annual MRI
30–39	No	Yes
40–49	Yes	Yes
50+	Yes	Depends on breast density

### Breast awareness

Breast awareness involves being aware of changes in the breast and feeling the breast with the flat of the hand in a systematic way, once a month, 5–10 days after the menstrual period (if applicable). It is also important to take time to look at the breasts in different positions. If there are any changes at all, women should inform their doctor. All women should practise this from their early twenties.

### If I have breast cancer, what is the chance that it will be detected on a mammogram?

Mammography has been shown to be effective in detecting breast cancers in women over 50. If a woman over 50 has breast cancer, there is about an 85% chance that mammography will detect it. This is called the sensitivity of the test. There is less evidence available about the sensitivity of mammography in women under 50. For example, in some studies of women aged 40–49, the sensitivity of mammography ranged from 62–76%. The sensitivity of mammography in young *BRCA* carriers (aged 35–49) has been demonstrated to be around 40%. This difference in sensitivity is one of the reasons we suggest that women have an annual mammogram rather than every two or three years.



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## **If I have breast cancer, what is the chance that it will be detected on a MRI scan?**

Studies on young women (aged 35–49) who carry *BRCA* mutations, have demonstrated that MRI screening is more effective at detecting breast cancer than mammography (77% versus 40%). Also, studies showed that combining both MRI and mammography screening improves the detection rate for both *BRCA1* and *BRCA2* mutation carriers to 94%. One drawback of MRI screening is that there is a higher recall rate to investigate abnormalities that may not be cancer, compared with mammography. Further research is ongoing into the usefulness of MRI screening for *BRCA* mutation carriers.

## **What are the risks associated with breast screening?**

Mammography involves X-rays, a form of radiation. People who are regularly exposed to high levels of radiation are known to have an increased risk of cancer. Thankfully, mammograms deliver a very small dosage of radiation. It is about the same as the dose a person receives by flying from London to Australia and back. The risk of developing cancer as a result of having regular mammography is therefore considered to be very low.

Overall, the benefit of early diagnosis and treatment of breast cancer far outweighs the risk of the small amount of radiation women receive during a mammogram. This is particularly true for women with a *BRCA1* or *BRCA2* mutation, who have a high risk of breast cancer. The radiation dose delivered by mammograms is continually monitored to ensure that it remains as low as possible while still providing a good quality image.

## **What does it mean if I am called back for another scan?**

If something unusual is found on your mammogram or MRI scan, you will be recalled for further examinations to clarify what the abnormality is. It is not unusual to be recalled following your first mammogram or MRI. After your first screening examination, the doctors will have images to compare, making it easier to tell if further investigations are needed. You may be recalled because of suspicious results or technical issues. If you are recalled, you may have a clinical exam,



a mammogram taking views from different angles, an ultrasound scan and, if needed, a biopsy to determine the nature of the abnormality. Not everyone who is recalled will be diagnosed with breast cancer, but the extra investigations can be uncomfortable and it can be a stressful time waiting for the results. This is why it is important that a screening test is good at detecting cancer when it is present and has a low recall rate.

### **Why can't I have ultrasound scans to screen for breast cancer?**

Studies have shown that an ultrasound scan is not an effective tool for routine screening for breast cancer. Ultrasound examinations are used to clarify the finding of a clinical breast examination, mammogram or MRI scan, where a lump or thickened area has been detected. It is also used when biopsies are taken.

### **What should I do if I find a lump in my breast and I am worried it is a sign of breast cancer?**

We recommend that you see your GP urgently for a clinical breast exam. Your GP can refer you to have an urgent assessment of the breast lump at a hospital. Patients referred to hospital with a suspected cancer diagnosis have to be seen within two weeks of the referral date, so this is a fast way to receive a clinical assessment. The genetics team and the NHSBSP centre cannot arrange for you to be seen and assessed more quickly than this route.

### **Where can I find more information about breast screening?**

The NHS Breast Screening Programme website has more information about breast screening.

[www.gov.uk/topic/population-screening-programmes/breast](http://www.gov.uk/topic/population-screening-programmes/breast)



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## Risk reducing strategies: Chemoprevention and lifestyle factors

### What choices can you make to help to reduce your risk of developing breast cancer?

**Risk-reducing surgery:** Women who have risk-reducing mastectomies reduce their risk of developing breast cancer to less than 5% over their lifetime, which is less than the risk in the general population. Women who have their ovaries removed before the menopause reduce their risk of developing breast cancer by up to 50%, even when hormone replacement therapy is given.

**Chemoprevention:** Chemoprevention describes drugs that are used to reduce the risk of cancer developing. This is different from chemotherapy which describes drugs that are used in the treatment of cancer. Guidance from NICE (National Institute for Health and Care Excellence) published in June 2013 recommends that chemoprevention should be considered in women at increased risk of breast cancer based on their family history. Two drugs have been recommended by NICE – tamoxifen and raloxifene. Both these drugs have anti-oestrogen properties. Both drugs have been shown to reduce the risk of breast cancer developing in women with a strong family history, but can cause a number of side effects which need to be taken into consideration.

Women with a high risk family history of breast cancer (who have been offered annual mammography between ages 40–60) and women with a *BRCA2* mutation may wish to consider chemoprevention. Firstly, they should have a full discussion with their doctor regarding the potential benefits and side effects associated with these drugs. Chemoprevention with these drugs is not recommended to women with a *BRCA1* mutation because most women with a *BRCA1* mutation develop a breast tumour that is not oestrogen sensitive, and there is no evidence that tamoxifen or raloxifene would reduce their risk of breast cancer. A leaflet about chemoprevention is available from the Cancer Genetics department or can be downloaded from The Royal Marsden website.

**Lifestyle and diet:** Currently there is no definitive scientific evidence to suggest that there are specific lifestyle or dietary factors that reduce or increase the risk of breast cancer for *BRCA* mutation carriers.

Many scientific studies in the general population have shown that limiting alcohol consumption, maintaining a healthy weight, being physically active, and breast feeding can help women to reduce their overall risk of developing breast cancer. Having a healthy diet that includes plenty of fruit and vegetables, pulses and whole grain foods can help you maintain a healthy weight.

**The pill:** There may be a small increase in breast cancer risk if the oral contraceptive pill is used for more than four years but evidence is still being gathered on this. It is not known if the mini-pill is safer than the combined pill but it is one alternative that women may consider. Another alternative is the Mirena coil. We advise that women discuss their contraception with their GP to assess the range of options.

**HRT:** Studies have shown that women who take hormone replacement therapy (HRT) after the natural menopause (especially the combined HRT, rather than oestrogen only HRT) have a slightly increased risk of developing breast cancer compared with women who do not take HRT. However, once women stop taking HRT, their risk goes back to normal within five years of stopping HRT. We do not know if this same effect occurs in *BRCA* mutation carriers. However, women who have risk-reducing surgical removal of their ovaries (bilateral salpingo-oophorectomy) at a younger age are usually offered HRT to prevent significant menopausal symptoms. Evidence indicates that these women still have a reduction in breast cancer risk (despite taking HRT) if their ovarian surgery is performed before age 50.

## Risk-reducing mastectomy

Women who have a faulty *BRCA1* or *BRCA2* gene may choose to have screening to manage their increased risk of breast cancer. However, screening does not prevent cancer. If a woman wishes to reduce her risk of developing breast cancer, she could consider risk-reducing mastectomies.

This is the surgical removal of breast tissue to try to prevent the development of cancer. There are various types of operations, but, in general, as much breast tissue as possible is removed. There will be some breast tissue remaining but the lifetime risk of breast cancer would generally be reduced by 90–95%.



Women who have risk-reducing surgery will no longer need annual mammography screening, because they will have very little breast tissue left. Reconstructive surgery can be offered at the same time as the risk-reducing mastectomies.

Making a decision to have risk-reducing surgery is very personal and very complex. There are many factors to consider. There are decisions to be made about the type of surgery you wish to have. The surgical team will explain the options to you over several appointments. You will have a chance to look at pictures with the breast nurse and speak with a clinical psychologist about the potential emotional impact of the surgery. One advantage of having risk-reducing surgery is that you can pick a time to have surgery and make arrangements in advance, regarding work and childcare for your recovery period.

A booklet called *Understanding risk-reducing breast surgery – prophylactic mastectomy and reconstruction*, produced by Macmillan Cancer Support, provides information about many aspects of this surgery for women who are considering this option. The booklet includes information about the surgery, reconstruction techniques, recovery, possible complications, the emotional effects of having the surgery and sources of additional information. If you are interested in finding out more about having risk-reducing surgery, you can ask for a referral to the breast team. You would be under no obligation to go ahead with the surgery. We recognise as much information as possible is needed before this type of decision is made.

## Breast surgery options for women affected by breast cancer

When a woman with a faulty *BRCA1* or *BRCA2* gene develops breast cancer, a breast surgeon will discuss with her the best way to treat her cancer. The surgery will be part of a treatment plan that may include chemotherapy, radiotherapy and hormone therapy with the aim of treating the cancer and preventing it coming back.

For *BRCA* carriers, the risk of a cancer recurring either in the breast or in another part of the body is not increased compared to a woman with the same type of breast cancer who does not have a *BRCA* mutation. However, *BRCA* carriers have an increased risk of developing a new breast tumour in either the same or the opposite breast.

This risk is approximately 1–3% per year. So depending on the age at which a woman is first diagnosed with breast cancer, their cumulative (total) risk of developing a new breast cancer may be significant and for this reason, a woman may want to consider more extensive surgery.

Having both breasts removed (bilateral mastectomy and reconstruction) will reduce the long-term risk of developing a new breast cancer to less than 5%. It will avoid the need for future breast screening by mammograms and MRI (magnetic resonance imaging). It may also provide an improved sense of control over risk and may avoid the worry associated with having breast imaging.

However, having both breasts removed does not reduce the risk of the original breast cancer coming back in another part of the body. We also do not know whether this more extensive surgery is necessary to maximise a woman's survival chances.

For these reasons, a decision about breast surgery includes thinking about the risks associated with the breast cancer that has already been diagnosed and the risk of developing a new breast cancer in the future. Other important factors to consider include how a woman feels about the impact of her surgery on her body image and relationships. The breast cancer team will help each woman decide what the best choice is for her.

Some women choose to have lumpectomy and radiotherapy initially and then consider surgery to both breasts later on. If considering this option, it is important to bear in mind that skin elasticity after radiotherapy can be less which can affect the type of reconstructive choices available.

## Managing ovarian cancer risk

Women who are carriers of a faulty *BRCA* gene have an increased risk of developing ovarian cancer during their lifetime.

### What is my risk?

On average, women have about a 2% chance of developing ovarian cancer in their lifetime. Women with a close relative affected with ovarian cancer may have a slightly increased risk of around 4–5%. Women who have a faulty *BRCA1* gene have a 40–60% lifetime risk of developing ovarian cancer. Women who have a faulty *BRCA2* gene have a 10–30% lifetime risk of developing ovarian cancer.

The risk of developing ovarian cancer starts to increase from around the age of 40 for *BRCA1* carriers and in the mid 40s for *BRCA2* carriers.

### What are the symptoms of ovarian cancer?

It is important to be aware of ovarian cancer symptoms and discuss them with your GP. Ovarian cancer was once known as a ‘silent’ disease, because its symptoms can be vague. Evidence now shows that any of the following three symptoms, if they occur on most days, may suggest ovarian cancer:

- persistent pelvic and abdominal pain
- increased abdominal size or persistent bloating (not bloating that comes and goes)
- difficulty eating, and feeling full quickly.

Occasionally, women may also experience other symptoms, such as urinary symptoms, changes in bowel habit, extreme fatigue or back pain, on their own or at the same time as those listed above. These symptoms are unlikely to be ovarian cancer, but may be present in some women with the disease. If you regularly experience any of these symptoms and they are not normal for you, see your GP.

There is further information available at:

[www.nhs.uk/conditions/cancer-of-the-ovary/pages/symptoms.aspx](http://www.nhs.uk/conditions/cancer-of-the-ovary/pages/symptoms.aspx)

## Is ovarian cancer screening available?

Currently there is no proven test or group of tests that reliably diagnoses ovarian cancer at an early stage. Therefore no routine screening for ovarian cancer is currently available. Several large studies have assessed the role of ovarian screening, but so far none have shown a benefit in high risk women (such as those with a *BRCA* mutation).

## What is available to reduce ovarian cancer risk?

We know that taking the oral contraceptive pill is protective against developing ovarian cancer. However, if women with *BRCA* mutations take the pill for an extended period of time (more than four years), it may slightly raise their risk of developing breast cancer.

## Risk-reducing ovarian surgery

The other option women can consider is having risk-reducing surgery to remove their ovaries and fallopian tubes, before any cancer is diagnosed.


### **What is a risk-reducing bilateral salpingo-oophorectomy (BSO)?**

A risk-reducing bilateral salpingo-oophorectomy (BSO; bilateral (both sides) salpingo- (fallopian tube), oophor- (ovaries), ectomy (excision of)) is the surgical removal of a woman's ovaries and fallopian tubes before an ovarian cancer has occurred. This surgery is carried out to reduce the risk of developing ovarian or fallopian tube cancer, and lowers the risk to less than 5%.

### **How do the ovaries normally work?**

Normally a woman has two ovaries and each month an egg is released from one of the ovaries. If the egg is fertilized, a pregnancy may result. If a pregnancy does not occur, the lining of the womb (uterus) falls away and the woman has her period.

As well as storing and producing eggs, ovaries produce the female sex hormones called oestrogen and progesterone.



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These hormones cause a woman's breasts to develop, help make periods regular and work to build up the lining of the womb each month to support a pregnancy (if it occurs). The ovaries also produce a hormone called testosterone that influences hair growth and sex drive (libido). As a woman ages and gets closer to the menopause, the ovaries make less of these hormones and her periods gradually stop. For most women this usually happens between the early 40s and mid-50s.

### **Who is offered a risk-reducing BSO?**

Some women, for example *BRCA* mutation carriers, are advised to consider risk-reducing BSO because they have a high risk of developing cancer of the ovaries. In this situation it is also recommended that the fallopian tubes be removed at the same time, as this tissue is similar to that of the ovaries. Women may choose to have risk-reducing BSO after they have considered their individual risk of ovarian cancer against the risks of the surgery, the risk of premature menopause and depending on personal circumstances, such as their family planning situation.

### **What if I prefer not to choose any of these options?**

After discussion with your doctor, you may decide that none of these options are appropriate for you at this time. It may be that you are younger than the recommended age for surgery, or it may be that you wish to stay fertile as you have not completed your family. It is important to make the decision that is right for you, and this decision can be discussed with your doctor, nurse or genetic counsellor at any time.

For women who choose not to have surgery, there are certain advantages and disadvantages that need to be considered. The advantages include that you will remain fertile (if you have not yet gone through the menopause) and that there are no side-effects (see **complications of surgery** below). The disadvantage of not having surgery is that it is not easy to recognise the signs and symptoms of ovarian cancer during the early stages. If ovarian cancer does occur and is only found at an advanced stage, it is much more difficult to treat effectively.



## **When should I consider having risk-reducing surgery?**

The age at which a woman's risk of developing ovarian cancer becomes significant will vary depending on her family history and which gene fault is present.

The risk of ovarian cancer in women who carry a *BRCA1* or *BRCA2* gene fault does not begin to rise markedly until about the age of 40 for *BRCA1* carriers and in the mid-40s for *BRCA2* carriers. Risk-reducing BSO is generally performed in women in their early to mid-40s.

For most women, the value of risk-reducing BSO before the age of 40 is minimal. However, a very small group of women, who have a strong family history of cancer of the ovaries before the age of 40, can be offered risk-reducing BSO earlier.


## **How is risk-reducing BSO performed?**

There are two main surgical ways of removing the ovaries – keyhole (laparoscopy) or open surgery (laparotomy). Both are carried out under general anaesthetic. The majority of women will be able to have the surgery performed laparoscopically (as keyhole surgery), but some women may need a laparotomy, especially if they have had major abdominal surgery before. Whichever approach is used, after the operation, both the ovaries and tubes will be sent to a laboratory to check if ovarian cancer is already present. You will get an outpatient appointment about four weeks after the procedure to discuss the final pathology results.

## **What other procedures are carried out?**

The womb is not usually removed as part of the risk-reducing BSO operation, although some women with a history of gynaecological problems may consider having a hysterectomy (removal of the womb) during the surgery.

A hysterectomy is a much bigger operation and will lengthen the hospital stay and recovery time. Some gynaecologists prefer to obtain a sample of lining from the womb (endometrium) during the procedure, known as curettage.



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Before the procedure, it is common practice to carry out a trans-vaginal ultrasound scan to assess the womb and ovaries, and a blood test to check the levels of an ovarian tumour marker called CA125. If the surgery is delayed following these investigations, for whatever reason, the gynaecologist may wish to repeat both.

## Hormone Replacement Therapy (HRT)

Women who have a risk-reducing bilateral salpingo-oophorectomy (RR-BSO) before 50 are usually offered hormone replacement therapy to prevent significant menopausal symptoms and to protect their general well-being (including heart and bone health). We recommend that, after RR-BSO, women (who have not had Estrogen-Receptor-Positive breast cancer) have HRT up until age 50 to 51, as this is the usual age at which menopause would have otherwise occurred naturally. Studies show that women who have had RR-BSO before age 50 actually have less breast risk (despite taking HRT) than women who still have ovaries.

If a woman has had an ER-positive breast cancer, HRT should not be used until discussed with her oncologist. Menopausal symptoms can vary greatly from one individual to another. Some women may have no menopausal symptoms after surgery, others may have some, such as hot flushes, night sweats, tiredness, loss of libido, vaginal dryness and mood changes. The use of HRT after BSO is best discussed with your gynaecologist and your GP before you decide about proceeding with ovarian surgery. There are also specialist Menopause Clinics where HRT and alternatives can be reviewed if a woman does not feel that her supplementation is suiting her, and we can refer you to these.

Women who have an earlier menopause are also at risk of developing osteoporosis (loss of bone density). We recommend that your bone density is monitored if your RR-BSO is done before age 50. The measurement is usually done after surgery as a baseline (which we request your GP to arrange), and it is then repeated every three to five years if the baseline scan is normal. If the baseline shows reduced density, then scanning is done more often, usually every two years. There are a number of alternatives to HRT for the prevention and treatment of osteoporosis, and these can also be discussed with your GP or gynaecologist.

## Sharing information with your family

### Genetic testing for your relatives

#### Who needs to know that a *BRCA* gene mutation has been identified?


During your genetic appointment, the doctor or the genetic counsellor will go through your family tree with you to identify who is at risk of inheriting the faulty *BRCA* gene. Cancer susceptibility due to a faulty *BRCA1* or *BRCA2* gene is inherited. Your close relatives (brothers, sisters and your children) will have a 50% chance of inheriting the faulty *BRCA* gene. In most families it will be easy to predict if the faulty *BRCA* gene came down through your mother's or your father's family, because of the family history of cancer. The only way to prove this is to test family members to see who has the faulty gene. Only your relatives from that side of the family will have a risk of inheriting the faulty *BRCA* gene.

#### What do I tell people?

You will have received quite a lot of information about the *BRCA* gene, your risks and your options. It can be daunting to know how to share all this information with someone else. Many people worry that they will be asked questions and they will not know the answers. Your genetics unit will not approach your relatives about your result, so usually it is you who would provide this initial information about the presence of a faulty *BRCA* gene to the family. The geneticists can provide you with a **to whom it may concern** letter for you to share with your relative(s) which contains all the required information so that they can ask for a referral from their GP to their local genetics unit.

#### Why do other people in my family need this information?

Genetic information is different from most medical information an individual receives, because it is not only relevant to the individual but also to their family members. Genetic information can provide an explanation as to why someone has a particular health problem, but it can also predict future poor health or the risk of having a child affected with a particular genetic problem.



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If someone is aware that they have an increased risk of developing cancer, they have the chance to make choices about genetic testing, cancer surveillance or preventative surgery. They may also decide to make changes to their lifestyle to help decrease their risk of developing cancer, and they may wish to consider the family planning implications and insurance issues. Knowing about the risk gives your relatives a chance to take action to reduce their risk of getting cancer or help ensure that cancer is detected at an early stage so it can be treated more effectively.

### **When should I share this information?**

There is probably no right time to tell people. Sometimes families are aware of the testing process and are waiting to hear the results. Other people choose to wait until they know their results before they mention it to anyone else. There will be a variety of factors that affect your decision to share this news, for example you may need time to get used to the information first, or someone may have been bereaved recently and you may feel it is better to wait a bit. Sometimes people are too young to be told everything or maybe you are waiting until you have the chance to see someone in person rather than telling them over the telephone or by mail. Women can start breast screening from 30 years of age, so if your female relative is over 30, she could start breast cancer screening right away if she is at risk of having the faulty *BRCA* gene. Anyone in the family who has been affected with breast, prostate or ovarian cancer should be told as soon as possible, because their doctors may need this information to make appropriate treatment decisions. It is important to highlight that women who are closely related (sister, daughter) to a person with a faulty *BRCA* gene can also have breast cancer screening, without undergoing genetic testing. We realise that not everyone wishes to have the genetic test.

### **How can I avoid upsetting my relative?**

Many people are concerned that sharing this information will cause their relative to feel very anxious or guilty and feel responsible for causing this upset. It is always difficult to share bad news. It might be useful to think of other times that you have had to share bad news, how you did it and what you learned from that experience.

It may be useful if you think about this information in a positive light; that members of your family, with this information, are in a better position to make choices that could reduce their risk of cancer or ensure it is detected as early as possible. It is important to remember that if someone has this *BRCA* gene mutation it is nobody's fault – we cannot control which genes were passed on to our children. It is also important to remember that if someone has a faulty *BRCA* gene, they have always had it since the moment they were conceived, so what is different now is that we can identify it and give people options to manage their risk.

### Where can I get help with telling my family?

The genetics team can help you to identify who needs to be informed in your family and provide you with a **to whom it may concern** letter. We can provide you with a copy of a letter that was written by a woman with *BRCA* gene mutation to her family, which is an example of how someone else in your situation chose to share information. There is a resource list which can direct you and your family to additional sources of information. It can be a burden to have this responsibility. Perhaps there is someone else within your family that you could share the information with and who could then help by taking on some of the responsibility of telling other people within the family. In some families, we see the information being passed on to one generation and it is then passed down within family units. The genetics team can discuss strategies for sharing information with you to make it as easy as possible.

### Who is at risk of having the faulty *BRCA* gene in my family?

If you are a *BRCA* mutation carrier, your close relatives have a 50/50 risk of having the faulty *BRCA* gene. The faulty gene would have been present in your family for many generations, it is not new within your family. It is only that we are now able to identify who actually carries the faulty gene. You would have inherited the *BRCA* mutation from either your mother or your father. This faulty *BRCA* gene causes the increased risk of developing cancer.



## **Your brothers and sisters:**

Your brothers and sisters each have a 50/50 risk of having the faulty *BRCA* gene. Each person would need to have genetic testing to determine if they have the faulty *BRCA* gene or not. You cannot predict if someone has the gene on the basis of their brothers' or sisters' genetic test results or if people look alike in the family.

## **Your children:**

Your children (or future children) each have a 50/50 risk of having the faulty *BRCA* gene. We do not offer predictive testing to children because screening or risk-reducing treatment is not generally needed or available for children. Once they are older, each of your children could choose to have genetic testing to find out if they have inherited the faulty *BRCA* gene or not.

## **Your extended family:**

The faulty *BRCA* gene would either have come down through your mother's or your father's family. So if you inherited the faulty *BRCA* gene from your mother, for example, then only your aunts, uncles and cousins on your mother's side of the family are at risk of having the faulty *BRCA* gene.

## **Does the faulty *BRCA* gene skip a generation?**

The faulty gene does not skip a generation. Not all individuals with a *BRCA* mutation will develop cancer (particularly men). For this reason, it may look as if the mutation has skipped but in reality it was present in the previous generation.

## **Is the faulty gene only important if you are a woman?**

When thinking about who is at risk of having the mutation in the family, it does not matter if you are a man or a woman. Both men and women have *BRCA1* and *BRCA2* genes and men can also carry a faulty *BRCA1* or *BRCA2* gene. The risk of developing cancer for men is not as great as it is for women, but they can still pass the faulty *BRCA* gene on to their offspring.

## How do I know who to tell about the gene?

When you have your appointment in the genetics clinic, the doctor or genetic counsellor will go through your family tree with you and explain who is at risk of having the faulty *BRCA* gene and who is not at risk. We can provide you with a letter that explains about the gene fault for you to share with your at-risk relatives. They can then decide if they want to have a genetics appointment to find out more information.

## What does dominant inheritance mean?

You may have heard the term **dominant inheritance** at your genetics appointment when the doctor or the genetic counsellor was describing how the gene mutation is passed down within the family. When we understand how a gene is passed down in a family, we can then determine who is at risk of having the faulty gene in the family.

We have about 20,000 genes. These genes are the instructions that our bodies need to develop and function normally. All genes come in pairs – we get one copy from our mother and the matching copy from our father.

A dominant condition is one where a person only needs one of the two genes to have a mutation, in order to be affected. In a *BRCA* carrier, having one faulty (mutated) copy is enough for someone to have the increased risk of developing breast, ovarian and prostate cancer, even though the other copy of the gene, which they inherited from their other parent, works normally.

Therefore we know that if a person carries a *BRCA* mutation, there is a 1 in 2 or 50% chance they will pass the faulty copy of the gene to their offspring; hence the 50% risk for children of *BRCA* carriers. We can also work out the risk for extended family members by looking at how they are related to the *BRCA* carrier in the family.



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## Predictive genetic testing

### What does predictive genetic testing mean?

This term refers to a genetic test that is offered to someone who is healthy (for example, has not had a diagnosis of breast or ovarian cancer). The purpose of the test is to determine if the person has inherited the faulty *BRCA* gene that has already been identified in one of their close relatives. It is called a predictive test because if the person has the faulty *BRCA* gene, it means that they have an increased risk of developing these cancers over their lifetime. Not everyone who has a faulty *BRCA* gene goes on to develop cancer, but the risk is increased.

### Who can have predictive testing?

Close relatives of an individual with a faulty *BRCA* gene can ask to be referred to their local genetics service by their GP to discuss the option of genetic testing. Your genetic doctor or genetic counsellor will go through the family tree with you to show you who in your family is at risk of having the faulty *BRCA* gene. We do not offer predictive genetic testing to children because this is an adult onset disorder so cancer screening or interventions are not necessary for children.

### What happens when someone is referred for genetic testing?

If someone is coming for a predictive genetic test, they may have one or two appointments to discuss the issues around testing, depending on their age and their understanding of the genetic information.

The first step for the genetics department is to confirm that there is a faulty *BRCA* gene in the family and obtain a copy of the relative's *BRCA* mutation report from the genetics centre where they had their test. If the report is not available, then predictive genetic testing may not be possible. At their first appointment, the doctor or counsellor will explain what the faulty gene is, how it is inherited, the associated cancer risks, and their risk of having the faulty gene and options for cancer screening or risk-reducing options. They will also discuss whether or not the individual wants to have a test at this point in their life,



who they have for support and who they have told about the test. We also discuss how a positive or negative result will impact on their lives and their relationships within the family. A letter summarising the consultation is sent to the individual after the clinic appointment. If they decide to have the test, results are given a few weeks later at an appointment (which is usually face-to-face, rather than by telephone). Individuals are welcome to bring a friend or relative with them to the results appointment for support.

### **What if someone does not want genetic testing?**

Not everyone who is at risk of having a faulty *BRCA* gene wishes to have a predictive genetic test. Even if someone does not want genetic testing, it is still useful to talk to them in the genetics clinic. We can explain the options and give them a chance to ask any questions. If people are at 50% risk of having a faulty *BRCA* gene, they are still eligible for extra cancer surveillance, such as breast screening from age 30, even if they do not want testing.

### **Does predictive genetic testing have a negative impact on insurance?**

Sometimes people worry that if they have a genetic test, they will have problems taking out life insurance. There is an agreed code of practice amongst insurance companies that results of predictive testing for high risk cancer genes will not be used by ABI insurers to determine premiums or eligibility for life insurance. This agreement was recently extended. Further information can be found on the website [www.abi.org.uk](http://www.abi.org.uk)

### **Planning to emigrate?**

If you are planning to emigrate to a new country, it would be good to check with that country's relevant office to determine if having a predictive genetic test could impact either your emigration application or your ability to obtain health insurance and/or health care provision before undertaking genetic testing.



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## Men and *BRCA* genes: What do men need to know?

Everyone has two copies of the *BRCA1* gene and two copies of the *BRCA2* gene. These genes are part of our normal complement of genes. Genes are the individual instructions which make our bodies develop and function normally. The *BRCA* genes' role is to help the body correct mistakes that occur in the DNA code during cell division and therefore help to prevent cancer. If someone has a faulty copy of either the *BRCA1* or *BRCA2* gene, it means that their body is less efficient at correcting mistakes that occur when cells divide, and are therefore at a higher risk of developing cancer during their lifetime. For reasons we do not fully understand, *BRCA1* and *BRCA2* gene faults mainly cause an increased risk of breast and ovarian cancer.

Many people mistakenly believe that either men do not carry *BRCA* genes at all or that having a faulty *BRCA* gene is not relevant to men within the family. This is not true. Men have *BRCA* genes and they can carry faulty *BRCA* genes. Men have a slightly increased risk of developing certain types of cancer and they can pass on the faulty gene down to both sons and daughters.

### **What are the cancer risks for men who have a faulty *BRCA* gene?**

Men who have a *BRCA* gene mutation have a slight increase in breast and prostate cancer risk as adults, and this is seen mainly in men with a change in the *BRCA2* gene. Men who have a *BRCA1* gene mutation have a 0.1–1% risk of breast cancer and a prostate cancer risk which is similar to (or may be slightly more than) the population risk. A man with a *BRCA2* gene mutation may have a 5–10% lifetime breast cancer risk and 20–25% lifetime risk of prostate cancer.

### **Can men be referred to genetics clinics for information about testing and cancer surveillance?**

Yes, men can ask their GP or hospital consultant to refer them to their local genetic service to discuss their family history, the option of genetic testing and arrange appropriate cancer surveillance plans.

## **What are the risks to the children of men who have a faulty *BRCA* gene?**

Men who carry a faulty *BRCA* gene have a 50/50 risk of passing the faulty gene to their children. The risk of inheriting the faulty gene is the same for both sons and daughters. However, daughters who inherit the faulty gene have a higher risk of developing cancer during their lifetime, compared with sons, because the risks are greater for women than men. Women need earlier and more frequent cancer surveillance.

## **What cancer surveillance can be offered to men who have a faulty *BRCA* gene?**

Male carriers of *BRCA* mutations are advised to practise breast awareness and to inform their doctors of any changes in texture of breast tissue (due to the small risk of male breast cancer). They may also be offered the opportunity to take part in a research project which is looking at the efficacy of prostate screening using PSA (prostate specific antigen) blood tests annually from the age of 40 to 69. There is a small risk of some other cancers for which screening is not usually considered to be useful. However, this is individualised depending on the family history. As in all cases, men (and women) are advised not to smoke. If there are any new symptoms, they should discuss this with their doctor without delay.

## **How common is prostate cancer?**

Prostate cancer is the most common cancer in men in the UK. Cancer Research UK estimates that 1 in 9 men may be affected by prostate cancer in their lifetime and usually after the age of 70 years. As faults in the *BRCA1* and *BRCA2* genes may increase the risk of prostate cancer, the IMPACT study is conducting a multi-centre international study of prostate cancer screening in men with mutations in *BRCA1* or *BRCA2*. It has enrolled several thousand men aged between 40 and 69 and who are from a family who are known to have a fault in either *BRCA1* or *BRCA2*. The study has now completed enrolment and the results will be reported in 2019 after all men have undergone five years of screening. More information can be obtained from [www.impact-study.co.uk](http://www.impact-study.co.uk)



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## Talking to children and young people about the familial *BRCA* mutation

Many parents find the prospect of talking to their children about the presence of a faulty *BRCA* gene within the family difficult and distressing. Studies show that parents' first instinct is to protect their children and they find it difficult to know what and when they should tell their children. Parents are often dealing with their own concerns about their health and future wellbeing, and can feel a sense of guilt that their children may also be at risk.

### **What helps children and young people?**

In most cases, children cope better when the family is more willing to discuss what is happening to different family members. Talking to children helps them feel valued and respected and helps them cope better than when they are left feeling confused and unsure how or what to ask.

Children get information from many places including school, television and friends. They are likely to already have some knowledge about cancer and possibly about hereditary cancer. By talking to them, you can help them sort out what is accurate and what is inaccurate and clarify things they are not sure about.

Children will probably be most worried about their parent developing cancer, so they will need reassurance and reminders that having the *BRCA* gene mutation does not always result in cancer. However, knowing you carry the gene mutation means that you can have regular tests to ensure that if a cancer arises, it can be detected very early on and treated.

### **When is a good time to tell your children?**

There is no right age but try not to keep secrets. Children and young people place great emphasis on trust and honesty from parents. Children often observe changes in their parent's behaviour and may try asking questions or may be waiting for you to discuss what is happening. Watch for any changes in your child's behaviour as this may indicate that they are worried or concerned about what they have observed or overheard.

By the age of eight years, children learn not to ask difficult questions unless their parent(s) gives them permission because they fear upsetting their parent(s). Therefore, you may have to prompt your child, and let them know you are willing to talk with them about the *BRCA* gene. This applies to older children too.

### **What information do you tell children?**

Try to respond to children's questions using language appropriate to their age. Providing small amounts of information gradually is likely to help children understand and cope best. Check on the question being asked so that you find out what your child actually wants to know.

Explain and provide the name '*BRCA* gene' – children cope better because knowing the name allows them to discuss it with you, and this knowledge gives them a sense of control. Parents often place a positive emphasis on the importance of knowing about the *BRCA* gene because it means there are improved screening and perhaps treatment options. When children become adults, there may also be even better treatments available, which you can explain to your child.

### **Communication tips**

- Children and young people prefer informal discussion often while doing other things together, for example, driving, cooking or gardening.
- Check their understanding because children worry about upsetting their parents and so may not always ask.
- Talking about *BRCA* is an ongoing discussion rather than a one-off conversation. Like adults, children probably need information given to them more than once. They may need time to digest information and then want to come back and discuss it with you.
- Discuss information young people find on the internet or in newspapers.
- Discuss emotions and provide reassurance they are not alone.
- Explain parents' behaviour if they are anxious or upset.
- Being with peers, for example cousins, in similar circumstances might be helpful.

- Support and guide decision-making, especially with young people, who usually like to make their own decisions but with advice from parents.
- If you do not know the answer, explain some questions do not have answers or that you will try to find out for your child.
- Agree appropriate times to discuss *BRCA* and cancer if your child asks questions at difficult or awkward moments.

### **What are children likely to know about genes and inheritance?**

**8–11 years:** They have a very basic understanding of heredity and that they share characteristics with parents. They may talk about genes but not fully understand what they are. Often children of this age cope with simple explanations in response to their questions and are not easily upset, although you may have to reassure them that having an altered *BRCA* gene is not the same as having cancer. Children and young people can easily confuse this, so it often needs repeating throughout development into adulthood.

**12–14 years:** Children are beginning to develop more insight about hereditary. They will begin to recognise that you having the gene mutation may have implications for them but will usually cope well if you explain there is only 50% chance of them having the *BRCA* gene alteration.

**15–17 years:** Children recognise the risks to their parent, themselves and often their future children and can begin to consider genetic testing. By this age, young people will be learning about *BRCA* and other hereditary diseases in school curricula.

Most children are quite pragmatic in response to genetic risk in families affected by inherited genetic conditions. Children and young people are often focused on developing friendships, school and their personal interests, so do not dwell on the risk.

### **What helps parents talk to their children?**

It has been observed that the following points have helped parents talk to their children:

- Younger children do not have the experience to recognise and anticipate the fuller implications so there is a gradual realisation.
- Not feeling under pressure to talk before an impending event, for example, a school science lesson.
- Talking was a relief for parents and ultimately easier than keeping the secret.
- Parents can be the role models for young people – giving them insight into how to cope with the risk.
- Recognising siblings may all have different needs and trying to find out what each understands at different times in their development.
- Ensuring children and young people understand a positive genetic test is not a cancer diagnosis – some get quite confused about this. Belief in a child's right to know.
- Support of other family members, friends and health professionals.
- Attendance at support groups gave focus to regular discussion with children and young people, with parents discussing where they were going and what had been discussed when they returned.

## Preparing to talk to your children

It might be worth considering the following benefits and drawbacks in preparing to talk to your child but try to take naturally occurring opportunities where possible.

Benefits	Drawbacks
<ul style="list-style-type: none"><li>• Makes the family closer.</li><li>• Provides support for children.</li><li>• Gives insight and helps them realise that parents being upset about <i>BRCA</i> is not down to them or their behaviour. It is no fault of theirs.</li><li>• Gives children confidence to talk to close friends.</li><li>• Children and young people feel valued by parent(s).</li><li>• Allows discussion of altered <i>BRCA</i> and cancer risk without centralising it to life.</li><li>• A shared reality and understanding helps children and young people cope.</li><li>• Reduces risk of children getting inaccurate information from elsewhere.</li></ul>	<ul style="list-style-type: none"><li>• It can be emotionally taxing dealing with questions.</li><li>• Children and young people can remind you about <i>BRCA</i> when you do not want to be reminded.</li><li>• Questions can arise at difficult or awkward moments – explain when it is appropriate to discuss it.</li><li>• Children may want to talk to peers but networks can be limited.</li><li>• Can affect schoolwork for a short time (but so can worrying about what is happening in their family if there is secrecy).</li></ul>

[Alison Metcalfe & Gill Plumridge (June 2010 Version 2)]




## Did you know?

### Family planning

If you are found to have a *BRCA* mutation before you have your family, you may have some questions about risk for your future children and you may wish to know if anything can be done to minimise this risk. Anyone with an inherited condition has a risk of passing it on to their children. The risk depends on the specific genetic condition, as there are several ways an inherited condition can be passed on in a family.

If you have a *BRCA* mutation, it is a dominant condition. Therefore there is a 50/50 or one in two chance that each of your children could inherit the *BRCA* mutation. This risk is the same for each pregnancy. The concept of dominant inheritance is explained in more detail in an earlier section of this booklet. Everyone who has a genetic condition can choose from a variety of family planning options and each option has pros and cons. It is a highly personal decision for you and your partner, and many factors will influence your choice. There is no right or wrong decision. You have to make the choice that best suits your circumstances, feelings and beliefs. In summary the options are:

1. Have your children without any intervention. Each child would have a 50% chance of inheriting the *BRCA* mutation and if they inherited the mutation, they would be at an increased risk of developing cancer as an adult.
2. Some couples decide not to have a family at all because they are very concerned by the risk of passing on the *BRCA* mutation.
3. Adoption – some couples may chose to adopt a baby to avoid the risk of passing the *BRCA* mutation to the next generation.
4. Egg or sperm donation – by using donor eggs (if the mother is the *BRCA* carrier) or donor sperm (if the father is the *BRCA* carrier) couples can avoid passing the *BRCA* mutation to the next generation.

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5. Prenatal testing – some couples may chose to get pregnant naturally and have a test during the pregnancy to see if the baby has inherited the *BRCA* mutation. The couple would plan to continue the pregnancy if the baby has not inherited the *BRCA* mutation. However, if the baby has inherited the *BRCA* mutation, they would choose to terminate the pregnancy.
  6. Pre-implantation genetic diagnosis (PGD) – some couples may choose this option to avoid passing the *BRCA* mutation to their offspring and avoid considering prenatal testing and possibly termination. There is no guarantee this technique will result in a successful pregnancy and funding for it on the NHS may not be available to all patients. (For further information, see below.)

Some of these options may seem drastic at first glance. Please bear in mind that these are the options for couples at risk of passing any genetic condition to their children. Some genetic conditions cause severe learning or physical disabilities, or often both, and some genetic conditions may be life-limiting, meaning that the child would not survive into adulthood due the condition. For most genetic conditions, there is no cure and limited options to treat the symptoms.

In our experience of talking to couples about family planning, most choose to have their family without any intervention. If anyone expresses an interest in finding out more about any of the other options, we can help make referrals or direct people to the appropriate services. We are happy to talk about these options in more detail at a genetic counselling appointment, if you would find that helpful.

As we get asked about PGD often, we have provided a brief overview of it here as well as websites to get more detailed information.

## Pre-implantation Genetic Diagnosis (PGD)

### What is it?

It is a technique used to select embryos that have not inherited the gene mutation which causes the genetic illness or cancer predisposition in a family.

## **Why would people consider having PGD?**

Some couples wish to ensure that they avoid passing on a gene mutation to their children. They want to be confident that their children will not be affected with the genetic illness or cancer predisposition (although it is important to point out that a risk of cancer would still be present in a child without the genetic mutation, just like for everyone else). Alternatives to PGD could include prenatal testing and termination of affected pregnancies, using donor sperm or eggs, adoption or deciding not to have children.

## **How is it done?**

PGD is carried out in conjunction with an IVF (in vitro fertilisation) procedure. The egg and sperm cells are collected from each parent and the embryos are created outside the woman's body. The embryos are then tested for the genetic mutation in the family. Only embryos that have not inherited the genetic mutation are selected to be implanted.


## **Who can have PGD?**

If an individual has a specific gene mutation that is known to cause a predisposition to cancer, they may be able to have PGD. If there is a strong family history of cancer, but no gene mutations have been identified or genetic testing is not possible in your family, then PGD is not an option.

If you have had cancer and stored embryos prior to treatment and your treatment has left you unable to have children naturally, you would need to have an in depth discussion about the pros and cons of using PGD technology with the PGD team.

## **Where can I find out more about PGD?**

If you are interested in considering this option, please ask to be referred to a PGD centre. There is one NHS PGD centre in London which offers testing for inherited cancer syndromes. At your first appointment they will discuss all aspects of PGD, including fertility assessment, funding applications, licence applications, time frame, risks and success rates.



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It can take many months from the first appointment to starting treatment, so you may wish to ask for an initial consultation long before you actually want to start your family. Please refer to the centre's website for more detailed information. The Centre for Pre-Implantation Genetic Diagnosis [www.pgd.org.uk/home](http://www.pgd.org.uk/home)

## Current studies and trials

Individuals attending the cancer genetics clinic may be offered an opportunity to take part in appropriate research trials. Specific details about research projects that are currently open can be obtained from the genetics team.

### **Taking part in research**

Taking part in research studies is voluntary, and if you prefer not to take part in research that is not a problem at all. We invite everyone we meet in the clinics, who is eligible, to take part in research. If you would prefer that we did not ask you, then please let one of the clinicians know so that we can make a note of your preference. We have many research projects running at one time, and it is likely that you will be invited to take part in several studies. You will be given an information sheet about each study and given as much time as you need to think about whether you would like to take part in the study. You will also be given the opportunity to ask any questions about the research project, including any benefits or disadvantages of taking part. With all research, if you decide to take part, you are free to withdraw at any time, without giving a reason and this would not affect the standard of care you receive.

### **Confidentiality**

If you decide to take part in a research project, any information you give us for the purposes of the study will be treated like all medical information. It will be kept strictly confidential and will be accessible only to the immediate study team. Information is stored on databases adhering to guidelines set out in the Data Protection Act. If specimens (for example, blood or urine) are obtained from you as part of the study, they will be stored using identifying codes that will be known only by the research team.

If you decide to withdraw from a study and you would like us to destroy any samples that you have provided for research, we will be happy to do so.

### **Types of research project**

**Epidemiological studies:** studies looking at the interaction of environmental, lifestyle and genetic factors in the development of cancer.

**Screening studies:** studies looking at new methods of screening for cancer.

**Psychosocial studies:** studies looking at the emotional and societal impact of disease.

**Drug trials:** studies looking at the use of new drugs for treatment or prevention of cancer.

**Tissue bank:** the storage of samples in a sample bank for future research.

### **New cancer treatment options for *BRCA* carriers**

It is useful for the oncology team to know when patients have a *BRCA* mutation to help guide their choice of treatment. Knowing that a patient carries a *BRCA* fault can affect treatment choices for newly diagnosed cancer as well as cancer that has come back after treatment. Treatment choices may be between established chemotherapy drugs or may include entry into clinical trials.

For example we know that *BRCA* carriers with ovarian cancer receiving platinum chemotherapy have higher response rates and longer times to relapse than women with non-hereditary cancer. Similarly, *BRCA* carriers who have breast cancer that has returned have been shown to be more likely to benefit from platinum chemotherapy than another standard drug. We know about the benefits of platinum chemotherapy because of previous clinical trials.

Your oncologist may discuss a clinical trial with you. These trials help to establish the best treatment for a particular type of cancer and sometimes will include the use of new drugs.



If you are seeking information on cancer trials you should discuss this with your oncology team and you can also refer to these websites:

*[www.cancerresearchuk.org/about-cancer/find-a-clinical-trial/what-you-should-be-told-about-a-clinical-trial](http://www.cancerresearchuk.org/about-cancer/find-a-clinical-trial/what-you-should-be-told-about-a-clinical-trial)*

and *[www.cancer.gov/about-cancer/treatment/clinical-trials](http://www.cancer.gov/about-cancer/treatment/clinical-trials)*

Sometimes a clinical trial may be available at your own hospital or you may need to go to a different hospital. It is important to know that trials for a particular drug may not be open to new patients at the time you are discussing treatment options with your oncologist.

## Insurance

### **Insurance implications of genetic testing for individuals who have had cancer**

If you have been diagnosed with cancer and have been offered a genetic test because of your cancer diagnosis, you are having a diagnostic genetic test. This includes cancer diagnoses that occurred in the past, even if treatment has been completed. The diagnostic genetic test may either be a full test of one or more genes associated with developing cancer such as *BRCA1* and *BRCA2*, or a specific test for a gene mutation that has been identified in your relative.

For Life, Income Protection and Critical Illness insurance policies, insurers can ask for information about your diagnostic genetic test to set the level of cover and cost of your premiums, in the same way as they ask about the rest of your medical history. For example, if you have been diagnosed with breast cancer that is due to a *BRCA1* mutation, that genetic information is part of your personal medical history.

However, insurers cannot ask you to take a genetic test and can only ask for the results of a genetic test you have already had.

If you are offered a diagnostic genetic test after your insurance policy is in place, then you do not have to disclose that you have had a genetic test or the results of the test.

If you are taking out a new insurance policy after you have had a genetic test, you will need to disclose that you have had the test along with the results.

Genetic test results do not have to be disclosed for health or travel insurance policies, but associated medical conditions such as cancer diagnoses do need to be disclosed.

If close relatives without cancer are taking out a new insurance policy (Life, Income Protection or Critical Illness) they would need to tell the insurance company about both the cancer diagnoses in the family and if any are due to a genetic diagnosis, if they are aware of this information. Relatives without cancer do not have to disclose the results of their own predictive genetic testing to insurance companies, but may choose to do so, particularly if their test result is negative.


### **Insurance implications of genetic testing for individuals without cancer**

If you have not had cancer and have been offered a genetic test because there is a known mutation in the family or because no family members with cancer can be offered a genetic test, you are having a predictive genetic test.

There is an agreed Code of Practice amongst members of the Association for British Insurers (ABI) that information about **predictive** genetic tests for cancer predisposition gene mutations are not used by insurance companies to determine if a policy is offered, or to determine the cost of the policy. The agreement relates specifically to Life, Income Protection and Critical Illness insurance products only. The agreement is currently in place until 1 November 2019. Health insurance and travel insurance policies are not specifically covered by the agreement, but also do not require disclosure.

It is important to note that insurance companies will assess risk based on family history information, as they have always done.

If any of your relatives have had cancer due to a cancer predisposition gene mutation, both the cancer diagnoses and the genetic test results are part of your family history information that should be disclosed.



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You are only required to provide the requested information that you are aware of at the time of taking out a new policy. If a relative has had cancer and/or genetic testing but you were not aware of it when you took out the policy, this will not subsequently be used to alter the policy.

You can choose to disclose your genetic test result. For example, if your test shows you do not have the gene mutation that caused cancer in your relatives this may have a favourable impact on your premiums as it reduces the impact of family history on your risk of cancer.

Further information can be found on these websites:

[www.abi.org.uk/Insurance-and-savings/Topics-and-issues/Genetics](http://www.abi.org.uk/Insurance-and-savings/Topics-and-issues/Genetics)  
[www.geneticalliance.org.uk/insurance.htm](http://www.geneticalliance.org.uk/insurance.htm)

## Organ donation

### **Can I still be on an organ donor?**

*BRCA* carriers have asked if they can still register to be an organ donor, given their *BRCA* status and/or personal history of cancer. The NHS Organ Donation information team has provided the following information:

There are only two conditions where organ donation is ruled out completely. A person cannot become an organ or tissue donor if they have been diagnosed with HIV or have, or are suspected of having, Creutzfeld-Jakob disease (CJD). Cancer is one of the diseases that can be passed from donor to the recipient. However the risk that this will happen depends to some extent on the type of cancer, the treatment received and the length of time since the disease was diagnosed and treated. Corneas can always be used. At the time that donation is being considered:

- The family would be asked about any history of cancer.
- The medical records would be reviewed.
- A report would be obtained from the GP, although this is not always available pre-transplantation.



So if the potential donor had been confirmed as having a genetic predisposition to cancer this would be reported as part of the assessment process pre-donation and would be included as part of the information provided to the transplant surgeon. A decision will then be made by a healthcare professional, at the time of death, about whether or not the organs can be used.

### **How can I register to be an organ donor?**

If you do decide to register please make sure that you have informed your family, both so that they are aware of your wishes and to ensure the information about your genetic status is available for the assessment. People can sign up to the register at the following web page [www.organdonation.nhs.uk/about-donation/what-is-donation/](http://www.organdonation.nhs.uk/about-donation/what-is-donation/)

### **Where can I get more information?**

This list of resources is intended to help you find further information or additional sources of support. Some patients may find these websites, books and charities helpful, but not all information will be relevant to all individuals.

Please consider the source of the material, for example clinical practice in the US may differ to clinical practice in the UK. Responsibility for the content of the website remains with the organisation that publishes the website.

#### **Articles:**

- My Medical Choice by Angelina Jolie (Op-Ed piece in the New York Times)  
[www.nytimes.com/2013/05/14/opinion/my-medical-choice.html](http://www.nytimes.com/2013/05/14/opinion/my-medical-choice.html)
- Diary of a Surgery by Angelina Jolie-Pitt (Op-Ed piece in the New York Times)  
[www.nytimes.com/2015/03/24/opinion/angelina-jolie-pitt-diary-of-a-surgery.html](http://www.nytimes.com/2015/03/24/opinion/angelina-jolie-pitt-diary-of-a-surgery.html)
- Cancer and the Maiden by Jessica Queller (Op-Ed piece in the New York Times)  
[www.nytimes.com/2005/03/05/opinion/cancer-and-the-maiden.html](http://www.nytimes.com/2005/03/05/opinion/cancer-and-the-maiden.html)



## Books:

- *Pieces of Me: Genetically Flawed – Surviving the Cancer I May Never Have* by Veronica Neave (2011)  
ISBN: 0980658209, 9780980658200
- *Pieces of Me (DVD): Genetically Flawed – Surviving the Cancer I May Never Have* by Denny Neave. A documentary about three Australian sisters who tested positive for the *BRCA2* gene mutation that had been identified in the family The DVD can be purchased [www.helpelisha.com.au](http://www.helpelisha.com.au)
- *Singing the Life: The Story of a Family Living in the Shadow of Cancer* by Elizabeth Bryan Vermilion (2008)  
ISBN-13: 978-0091917166
- *Am I Next In Line?: How I Conquered My Risk Of Hereditary Breast And Ovarian Cancer* by Monique Achtman (2006)  
ISBN-13: 978-0973373202
- *Positive Results: Making the Best Decisions When You're at High Risk of Breast or Ovarian Cancer* by Joi Morris & Ora Gordon  
Prometheus Books (2011)  
ISBN 978-1-59102-776-8
- *Apron Strings: Inheriting Courage, Wisdom And Breast Cancer* by Diane Tropea Greene Books (2007)  
ISBN-13: 978-1568251080
- *Pretty is What Changes* by Jessica Queller Spiegel & Grau; (2008)  
ISBN-13: 978-0385520409
- *Why I wore Lipstick to my Mastectomy* by Geralyn Lucas St. Martin's Griffin (2005)  
ISBN-13: 978-0312334468
- *Previvors: Facing the Breast Cancer Gene and Making Life Changing Decisions* by Dina Roth Port (2010)  
ISBN-13: 978-1583334058
- *The Genesis Breast Cancer Prevention Diet* by Michelle Harvie & Roy Ackermann Rodale International Ltd (2006)  
ISBN-13: 978-1405093460

### **Websites for further information:**

- Association of British Insurers (ABI)  
*www.abi.org.uk*
- Genetic Alliance UK  
*www.geneticalliance.org.uk*
- National Cancer Institute (US)  
*www.cancer.gov/types/breast*
- Genetics Home Reference (US)  
*ghr.nlm.nih.gov/condition/breast-cancer*
- Cancer Research UK (CRUK)  
*www.cancerresearchuk.org*

### **Charities and support groups:**

- Macmillan Cancer Support  
*www.macmillan.org.uk*
- FORCE: a support group for *BRCA* carriers based in America  
*www.facingourrisk.org*
- BrCa Umbrella Support and Social Network:  
*www.brcaumbrella.ning.com*
- Pink Hope: an Australian website for *BRCA* carriers  
*www.pinkhope.org.au*
- Breast Cancer Now  
*www.breastcancernow.org*
- Breast Cancer Care  
*www.breastcancercare.org.uk*
- The National Hereditary Breast Cancer Helpline and Information Centre  
*www.breastcancergenetics.co.uk*
- Ovacome: The Ovarian Cancer Support Network  
*www.ovacome.org.uk*
- Ovarian Cancer Action  
*www.ovarian.org.uk*
- Target Ovarian Cancer  
*www.targetovariancancer.org.uk*

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Revised May 2016

Planned review May 2019

This booklet is evidence based wherever the appropriate evidence is available, and represents an accumulation of expert opinion and professional interpretation.

Details of the references used in writing this booklet are available on request from: The Royal Marsden Help Centre

Freephone: 0800 783 7176

Email: [patientcentre@rmh.nhs.uk](mailto:patientcentre@rmh.nhs.uk)

The Royal Marsden NHS Foundation Trust

Fulham Road

Downs Road

London SW3 6JJ

Sutton, Surrey SM2 5PT

[www.royalmarsden.nhs.uk](http://www.royalmarsden.nhs.uk)

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No conflicts of interest were declared in the production of this booklet.

The information in this booklet is correct at the time of going to print.



Typeset by

Print Room, The Royal Marsden NHS Foundation Trust

Downs Road, Sutton, Surrey SM2 5PT

GE-1200-04

The Royal Marsden publishes a number of booklets and leaflets about cancer care. Here is a list of information available to you.



## Diagnosis

- A beginner's guide to the BRCA1 and BRCA2 genes
- CT scan
- MRI scan
- Ultrasound scan
- Lynch Syndrome



## Treatment

- Central venous access devices
- Chemotherapy
- Clinical trials
- Radiotherapy
- Radionuclide therapy
- Your operation and anaesthetic



## Supportive care

- Eating well when you have cancer
- Lymphoedema
- Reducing the risk of healthcare associated infection
- Support at home
- Your guide to support, practical help and complimentary therapies



## Your hospital experience

- Help Centre for PALS and patient information
- How to raise a concern or make a complaint
- Making your stay with us safe
- Your health information, your confidentiality



Please visit [www.royalmarsden.nhs.uk/patientinformation](http://www.royalmarsden.nhs.uk/patientinformation) where several patient information booklets are available to download.

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SERVICE  
EXCELLENCE

