breast cancer now

Genetic testing for faulty breast cancer genes

Information for people eligible for genetic testing due to their family history

Quick facts

- **Faults in breast cancer genes, such as BRCA1 or BRCA2, are rare.**
- **People with a faulty breast cancer gene have an increased risk of developing breast cancer, and sometimes other cancers too.**
- **If you have a confirmed family history of breast cancer and are estimated to have at least a one in 10 chance of having a faulty gene, you may be able to have genetic testing on the NHS. Genetic testing involves an explanation of the possible outcomes, followed by a blood test.**
- **Genetic testing can sometimes confirm which members of a family have inherited an increased risk of breast cancer.**
- **However, other times the results are inconclusive.**

What are 'faulty breast cancer genes'?

Some of the genes in our bodies can contain a fault (a mutation). Often this doesn't have any effect, but other times it might make our cells behave differently to usual.

Experts have identified some faults in genes linked to breast cancer, including BRCA1, BRCA2 and TP53. People with these faults are more likely to develop breast cancer during their lifetime.

Some people with a family history of breast cancer have a fault in a known breast cancer gene but some do not.

What is genetic testing?

Genetic testing checks the DNA in a sample of blood for faults in genes linked to a particular condition (such as an increased risk of breast cancer).

The test cannot prevent breast cancer developing, but it can give an indication of how likely it is that a person will develop breast cancer in the future. It can sometimes confirm which people in a family have an increased risk and which do not.

Am I eligible for genetic testing?

Both men and women with a confirmed family history of breast cancer can have genetic testing for faulty breast cancer genes, if they meet certain criteria.

Your risk assessment at a genetics clinic will determine whether you are eligible for genetic testing on the NHS. People are eligible if they have at least a one in 10 (10%) chance of having inherited a faulty breast cancer gene.

If you are interested in having genetic testing, but have not yet had your family history assessed, the first step is to visit your GP (or breast care team if you are receiving treatment for breast cancer). Find out more in our fact sheet *Exploring your family history of breast cancer: Information your GP will need.*

If you have not been to a genetics clinic yet, but your family history specialist thinks you are at high risk for breast cancer and might benefit from further assessment at a genetics clinic, they can refer you.

Where does the test take place?

People go to a specialist genetics clinic for genetic testing.

What are the first steps?

If you're eligible for a test, you will have genetic counselling before it takes place. This is to help you decide whether or not you want to have the test and to explain what the results could mean for you and your family. If you decide to go ahead, a sample of your blood will be taken. This is sent to a laboratory where your DNA will be checked.

Who in my family will be tested first?

If possible, a person in your family who has been diagnosed with breast cancer will be tested first. This is called mutation searching (or diagnostic testing). If a person with breast cancer is not available, a relative without breast cancer can be tested first if their chance of having a faulty gene is high enough. The results take about eight weeks to come back. Waiting times may vary. For example in Wales, tests can take up to 16 weeks to complete. Check with your genetics specialist if the waiting time for results is not clear.

If a faulty gene linked to breast cancer is found, other family members can be tested too, to see whether they have the same faulty gene. This second stage is called predictive testing and the results take about two to four weeks to come back.

The order of genetic testing for you and your relatives will depend on the patterns of cancer in your family and who is available for testing. Find out more about the likely steps for your family at **familyhistory.breastcancernow.org**

Which gene faults are tested for?

There are hundreds of faults (mutations) that genetic testing can look for, most of which are in two genes known as BRCA1 and BRCA2. The DNA code for these two genes is checked from start to finish to look for any faults. Sometimes possible faults in other genes, such as TP53, are looked for instead depending on a person's family history.

Some people of Ashkenazi Jewish descent may be offered testing for very specific BRCA gene faults that are frequently associated with a family history of breast cancer in this ethnic group.

Does genetic testing always find a faulty gene?

For many families with an increased risk of breast cancer, genetic testing is not able to pinpoint a genetic cause of their increased risk. This is because although some of the faulty genes associated with breast cancer are known, there may be other rarer, or more complex inherited faults that are not yet known, and so geneticists can't test for them yet. In addition, sometimes a fault is found but it is not clear whether this will increase the person's risk of breast cancer. Researchers are working to discover more gene faults linked to breast cancer and other cancers.

What do the results mean?

If you are found to have a faulty breast cancer gene, this means you are at increased risk of developing breast cancer and associated cancers in the future. There is a 50% chance that you will pass on the gene to each child you have. Your genetic counsellor will be able to explain your risk, steps you can take to manage your risk, and what this means for you and your family.

If a faulty gene is found in your family but your own test shows that you do not carry this gene, this means that you are not at increased risk of breast cancer. There is no chance that your children could inherit the faulty gene from you.

If no faulty gene linked to breast cancer is found in your family, this means a genetic cause for your family's increased risk could not be found. Each person's risk of developing breast cancer can still be estimated, based on the patterns of cancers in your family. At some time in the future, your family's situation can be reviewed.

About Breast Cancer Now

We're Breast Cancer Now, the UK's largest breast cancer charity – and we're dedicated to funding research into this devastating disease. We believe that if we all act now, by 2050, everyone who develops breast cancer will live. Join us at **breastcancernow.org**

This leaflet is based on up-to-date research evidence and aims to give the best information available. All information was reviewed by appropriate experts and members of the public. We advise readers with concerns about breast cancer to discuss them with a health professional.

If you would like more information about the sources of evidence on which this publication is based, please contact **info@breastcancernow.org** or call 08080 100 200.

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