

## Important information about your data



Your genomic data and samples will be stored as part of your health record. DNA samples may be used anonymously for quality control. All data is kept securely and confidentially in line with UK law and NHS policy. More information can be found at [www.england.nhs.uk/contact-us/privacy-notice](http://www.england.nhs.uk/contact-us/privacy-notice)

Information in this guide should be used to supplement professional advice specific to your circumstances. If you have any questions, it is important to ask your medical team or contact

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Telephone Number/Email.....  
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# Genetic testing for PARP inhibitor treatment in breast cancer (Test Code R444)

## Patient Information Leaflet



South West  
Genomic Laboratory Hub

## Why have I been offered a genetic test for PARP inhibitor treatment in breast cancer (R444)?



You are being offered this test because it may help to inform your cancer treatment. Most breast cancers do not have a genetic cause. Those that do, can sometimes benefit from specific medicines. At present, we know that people with a BRCA gene variant may benefit from drugs called PARP inhibitors. If you were found to have a BRCA1 or BRCA2 gene variant, you may be offered these drugs.

## What happens if I have this genetic test?



This test (Code R444) is carried out on a blood sample.

If you decide to have the R444 genetic test, your healthcare professional will discuss this with you. There is a form to complete which records your choices. Your blood sample will be sent for analysis. Results usually take around 6 weeks. This test is about your cancer and inherited breast cancer risk, it does not look for other health conditions.

We will look at a number of genes associated with breast and ovarian cancer risk. The precise list of genes may change over time, as we learn more about cancer genetics. Only genes with a proven influence on breast cancer risk will be added. If you have specific questions about the genes currently included in the R444 test, please discuss these with your healthcare professional.

If required, you have the option to speak to a genetic specialist before making your decision about testing. Everyone who tests positive, or has a significant family history, will be offered a referral to a genetic specialist.

## Are other cancers associated with these genes?



The genes on this panel primarily influence breast and ovarian cancer risk. Some influence the risk of additional cancers, but to a smaller extent. Other relevant cancers might include pancreatic, skin, or prostate cancers (in men). If a gene variant is found you will be given detailed advice and screening options, tailored to your situation.

This section is for any notes you'd like to make during your appointment

## 2. What does it mean if no variants are found on my genetic test?

2

This is the most common result. This result means it is unlikely that your breast cancer was caused by an inherited gene variant. The risk of future cancers (in you or your relatives) is unlikely to be raised unless you have a significant family history of cancer.

If you have a significant family history of cancer you will be referred to Clinical Genetics for further discussion.

## 3. What does it mean if genetic variation is found on my test, but is of uncertain significance?

3

We are all different and there is a lot of variation within our genes. Some gene variants have no effect on how a gene functions. If a new or rare variant is picked up, it can hard to predict whether it affects cancer risk.

If the scientists cannot be sure of a variant's effect, they will report it as being as of uncertain significance (VUS). If an uncertain variant is found, you will be referred to Clinical Genetics so that they can assess this for you and your family.

## What else might the test tell me?



The test may give us information about why you developed breast cancer. It could tell us about your risk of developing new cancers in the future. As this test looks for inherited (germline) gene variants, it may have implications for some of your relatives.

If we find that you have an increased risk of future cancers we will discuss screening and risk reducing methods with you.

Please take time to ask all the questions that you need to.

## Is breast cancer inherited?



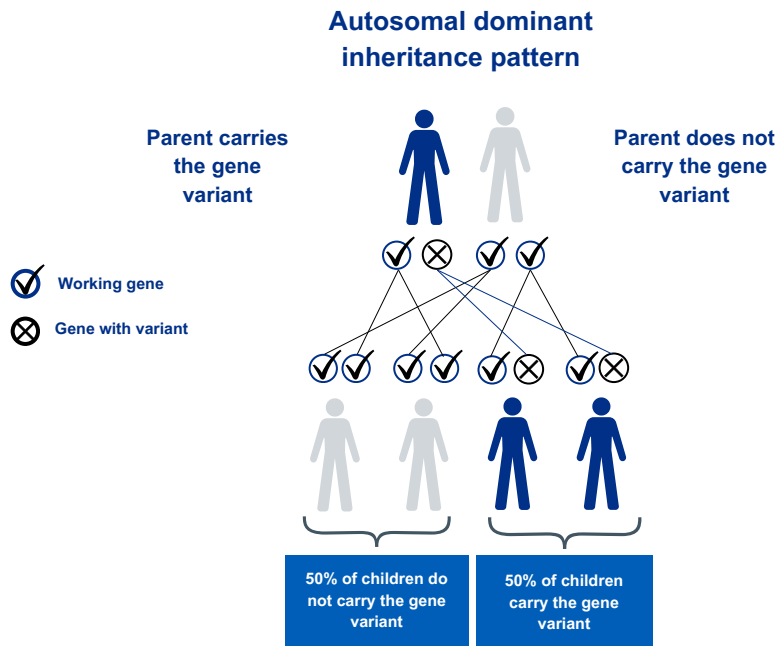
Most breast cancer is not inherited. Breast cancer is a common condition, it affects around one in seven women in the UK. Only a minority of breast cancers, around 5-10%, are strongly influenced by a genetic variant. Genes which influence breast cancer risk generally fall into one of two categories:

- High risk breast and ovarian cancer genes. These genes can cause a high lifetime risk of breast cancer. High risk is defined as a lifetime breast cancer risk of 30-80%. Examples of high risk genes include BRCA1, BRCA2 and PALB2. These genes also influence the risk of ovarian cancer. There may be some increased risk of other cancers, depending on family history.

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- Moderate risk breast or ovarian cancer genes. These genes increase cancer risk, but not as strongly as the high risk genes. Moderate risk genes typically cause a lifetime breast cancer risk of 20-30%, or an ovarian risk in isolation, depending on family history. Examples of moderate risk genes include CHEK2 and RAD51C. Moderate risk genes were discovered more recently, so we are still learning about their effect on cancer risk.

**Inheritance:** If someone has a gene variant, there is usually a 50% chance of their close relatives (parents, children and siblings) also having it. This is not influenced by sex or gender, as we all have these genes. This type of gene variant only affects cancer risk in adults.



## What results might I get?



Once your sample has been taken the results are returned to your healthcare professional who will share them with you. There are three outcomes to this genetic test:

1. A variant has been found in a breast cancer gene.
2. No variants were found in the genes tested.
3. Some genetic variation was found but it is of uncertain significance.

## 1. What does it mean if a variant is found on my genetic test?

1

This is likely to explain why you developed breast cancer. Your breast cancer team will inform you of the result and consider this information when planning your treatment. In some cases, you will be offered PARP inhibitors as part of your care.

You will be referred to Clinical Genetics so that you can discuss your result in more detail.

Clinical Genetics will explain what the test result means for your future risk of cancer, your options for cancer screening and measures to reduce these risks.

A test will be available for members of your family to assess their genetic cancer risk. Clinical Genetics will support you in sharing the information with family members, so that everyone gets the care they need.