



Understanding genetic testing in breast cancer

You have received this leaflet because a member of your cancer care team has made a decision to discuss genetic testing with you.

This booklet has been produced and funded by AstraZeneca.

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What is genetic testing?

What is a gene?

Genes are a set of instructions for cells in the human body

It is becoming more common for women with breast cancer to be offered genetic testing

Genetic testing – sometimes called genomic testing – finds changes in genes that can cause health problems. These changes are often referred to as genetic variants. In cancer, genetic testing can be used to look for certain genetic variants which can tell us something about your cancer including:

- What your risk of developing cancer is (hereditary risk)
- Whether your cancer is more or less aggressive (prognostic)
- Whether your cancer might be susceptible to specific treatments (therapeutic)

Genetic testing in breast cancer is done through a blood or saliva test:



- These tests can be used to detect “**germline variants**”
- Germline variants are faults in genes which are present in every cell of your body and are there from birth
- If you have a germline variant it will in most cases have been passed onto you from a parent. This is known as the variant being ‘hereditary’ or ‘inheritable’

For more information please visit these websites or scan the QR codes:

- <https://www.nhs.uk/tests-and-treatments/genetic-tests-for-cancer-risk/>



- <https://www.nhs.uk/tests-and-treatments/genetic-and-genomic-testing/>



- <https://www.cancerresearchuk.org/about-cancer/causes-of-cancer/inherited-cancer-genes-and-increased-cancer-risk/genetic-testing-for-cancer-risk>



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Which genetic tests are used in breast cancer?

In breast cancer, genetic testing such as BRCA, may be offered to patients. Knowing your genetic status means that your doctor may be able to tailor your treatment plan for your type of cancer.

BRCA testing

BRCA is a gene (BReast CAncer gene). There are two forms of BRCA gene, *BRCA1* and *BRCA2*. These genes are responsible for helping cells repair themselves and preventing cancer development. Faults in either of these BRCA genes can increase the risk of developing some cancers

Genetic testing is offered to see if you are carrying a BRCA variant. The result of this test is often described as ‘finding out your BRCA status’.

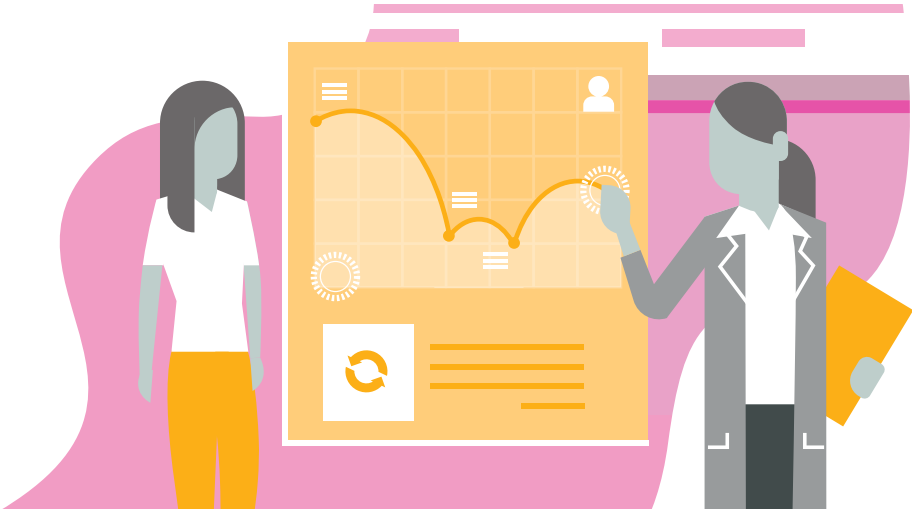
Variants in BRCA can be both germline (found in all cells) or somatic (found only in tumour cells) and about 1 in 20 women with breast cancer have a germline BRCA mutation.

In most cases, breast cancer is a one-off occurrence that is unrelated to any gene variant or family history. However, if the results of your germline test detect a *BRCA1* or *BRCA2* variant, this means your relatives may also carry the BRCA variant. Depending on the results, other members of your family may be offered germline BRCA testing, but this is a personal decision for each individual.

How do you perform genetic testing?

Your healthcare professional can make arrangements for you to have a sample taken. The sample is then sent to a genetics laboratory. They will look for any changes in the genes associated with breast cancer.

How do I consent to genetic testing?

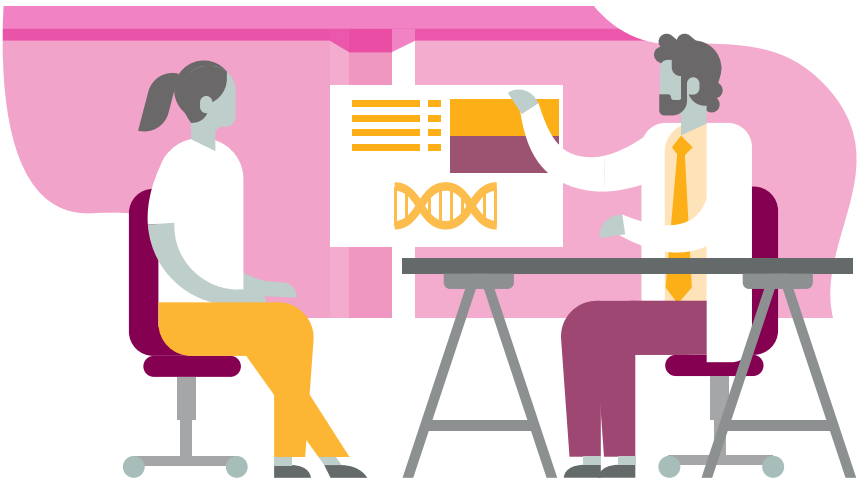


Before you undergo genetic testing, it is important that your healthcare professional explains the test and the possible consequences of the test results to you. This is so that you can fully understand the procedure, including the intended benefits and any possible risks. This is called informed consent. Once your healthcare professional has explained the test to you, you can let them know verbally whether you are happy to have the test.

What support can I get if I need to have genetic testing?

Genetic counsellors can provide you and your family with support if needed

Your cancer care team will talk to you about genetic testing and what the results of these tests mean. This is not the same as genetic counselling. If a relevant genetic mutation is identified you will be referred for genetic counselling. Talk to your cancer care team if you have any questions about this.



What happens after my genetic testing?

The testing lab should return your results to your healthcare team within 6-8 weeks. Your healthcare team will discuss the implications of your genetic testing results with you and may also involve a genetic counsellor, if appropriate.

Where can I find support?

There are several UK charities who provide support to anyone affected by breast cancer

These charities offer telephone support, face to face or email support groups, patient hubs, leaflets and resources. For more information click the links or scan the QR codes:



<https://brcaumbrella.ning.com/>



**BREAST
CANCER
NOW** The research & support charity

<https://breastcancer.org/about-breast-cancer/awareness/breast-cancer-in-families/>



**MACMILLAN
CANCER SUPPORT**

<https://www.macmillan.org.uk/cancer-information-and-support/worried-about-cancer/causes-and-risk-factors/brca-gene>



These organisations are independent patient support groups and AstraZeneca are not responsible for the content of their websites. The organisations have not been involved in the development of this material.

BReast CAncer Genes and me

This project has been developed and funded as part of a collaborative working initiative between the NHS South West Genomic Service and AstraZeneca UK to support consultations you may have with your healthcare team. Please scan the QR code to access six short animations providing an overview of the genetic testing pathway.



<https://www.youtube.com/@SWGenomics>

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References

NHS. Predictive genetic tests for cancer risk genes. Available at: <https://www.nhs.uk/tests-and-treatments/genetic-tests-for-cancer-risk/> (Accessed October 2025)

NHS. Genetic and genomic testing. Available at: <https://www.nhs.uk/tests-and-treatments/genetic-and-genomic-testing/> (Accessed October 2025)

Cancer Research UK. Genetic testing for cancer risk. Available at: <https://www.cancerresearchuk.org/about-cancer/causes-of-cancer/inherited-cancer-genes-and-increased-cancer-risk/genetic-testing-for-cancer-risk/> (Accessed October 2025)

Notes



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