NHS North Thames Genomic Laboratory Hub | The Royal Marsden NHS Foundation Trust | ICR The Institute of Cancer Research | NHS North Thames NHS Genomic Medicine Service Alliance

BRCA-DIRECT

Genetic Testing for Breast Cancer Patients

Please read this booklet to learn more about genetic testing that is available to you to identify **heritable gene faults**. If you would like to proceed with genetic testing, follow the instructions on the back cover to:

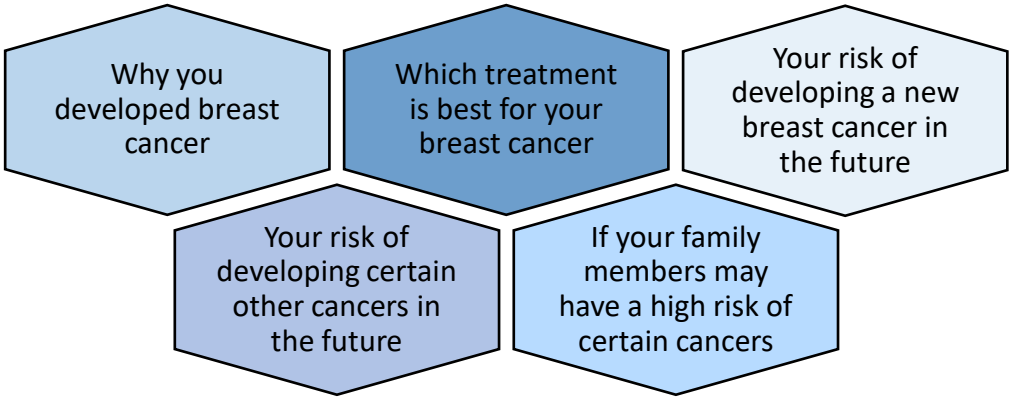
1. complete a genetic test consent form ☐
2. complete a personal details form ☐
3. provide a saliva sample ☐
4. return the forms and sample at your local post office ☐

If you have any questions or would like to discuss further, please contact us:

Genetics Helpline: +44 20 3437 6514
Monday to Friday, 9:00am to 5:00pm
Extended hours to 7:00pm on Wednesdays

Why is this genetic test helpful?

- The results of the genetic test may give useful information to you and your clinicians about:



- Approximately 5% of women diagnosed with breast cancer (1 in 20) have a gene fault in a breast cancer susceptibility gene.
- These gene faults are heritable: they are inherited from a parent and can be passed down in a family.

1 in 20

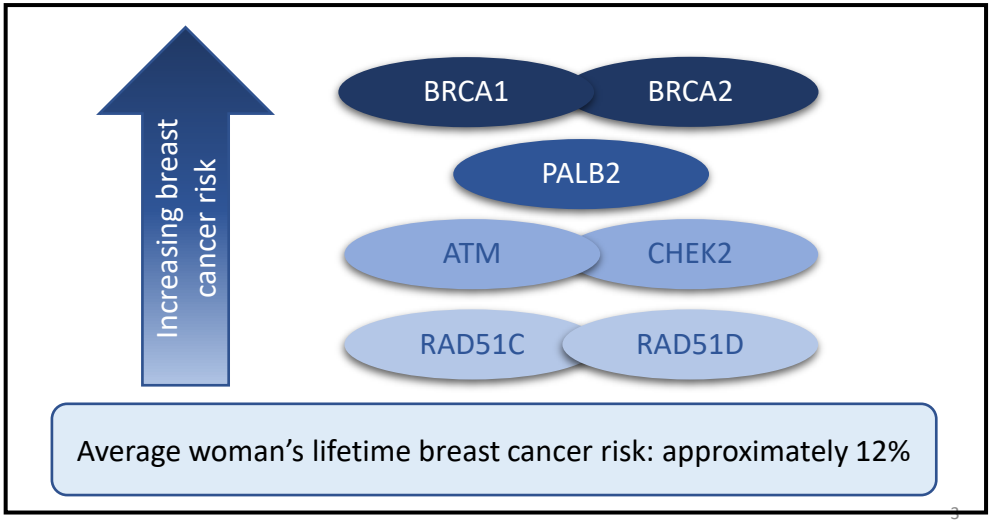
Why might I choose not to have this test now?

- It can be emotionally difficult for some people to learn that they have a gene fault. Some people may feel that now is not the right time to have this test.
  - BRCA-DIRECT is a time-limited program being offered in some hospitals to any woman with a new diagnosis of breast cancer. You may or may not be eligible for NHS BRCA genetic testing in the future. If you are interested in genetic testing, but would like to defer to a later date, you may speak to your clinician or call our genetics helpline to learn more about your options.
- If you already know that there is a gene fault in your family, you may wish to be referred by your GP directly to clinical genetics for testing and support.

What can the test tell me about my breast cancer?

This genetic test helps find out if your breast cancer was caused by a gene fault. This information can help you and your clinicians make choices about treatments and/or breast surgery.

- The average woman has a 12% risk of developing breast cancer in her lifetime. A gene fault can increase a woman's breast cancer risk further.
- The genetic test looks for faults in seven breast cancer susceptibility genes: BRCA1, BRCA2, PALB2, CHEK2, ATM, RAD51C, and RAD51D.
  - Gene faults are also called "pathogenic variants," "harmful alterations," or "mutations."
- Faults in these genes are associated with differing levels of increase in breast cancer risk.
  - For example, faults in BRCA1/BRCA2 are associated with a higher lifetime risk of breast cancer of approximately 70%.
  - For faults in the other genes, the increase in breast cancer risk is less.
  - Breast cancer risk will also be influenced by other factors in addition to the gene fault, such as family history.



What can the test tell me about my risk of new cancers in the future?

- There are different cancer risks associated with each of the seven genes on this test.
  - The increased risk for developing a new breast cancer in the future varies gene-by-gene.
  - Some of the genes are also associated with an increased risk of certain other types of cancer in the future.
    - For example, gene faults in BRCA1 and BRCA2 also increase the risk of ovarian cancer.
- Your individual risk of developing different cancers will also depend on your family history and personal factors (such as weight and alcohol intake).

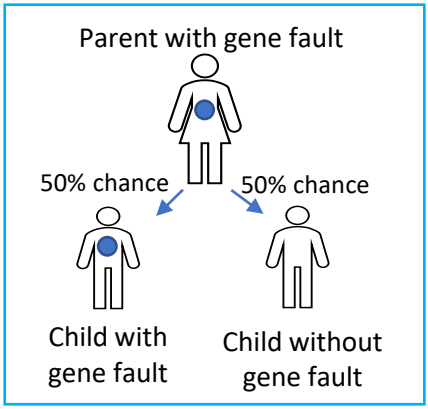
How can I manage my future cancer risk if I have a gene fault?

- Knowing that you have a gene fault can open more options for managing your risk.
- Cancer risk management options are different for each of the seven genes on this test.
- Options may include additional screening (for example, additional mammograms) or surgery (for example, removal of breast tissue or ovaries).

If the genetic test finds that you have a gene fault, you will have appointments in Clinical Genetics and be given detailed information about your risks and management options related to that gene.

What do the results mean for my family?

- If you have a gene fault, other people in your family may have the same gene fault.
- Both men and women can have and pass down gene faults.
- Every time a person with a gene fault has a child, there is a 50% chance (1 in 2) that the gene fault is passed on to the child and a 50% chance (1 in 2) that the gene fault is NOT passed on.



- If you have a gene fault, other adults in your family can access genetic testing to learn if they have this gene fault too.
- Sharing of genetic information in your family can be done in discussion with you or through a process that will not personally identify you.

Genetic testing and insurance

- This genetic test is termed a **diagnostic test** because you have previously been diagnosed with breast cancer. Insurance companies may ask you for the results of your genetic test if you open a new life, income protection or critical illness insurance policy. Any policies already in place will not be affected.
- If you have a gene fault, and then your relative chooses to have genetic testing, their test is considered a **predictive test** (unless they have also had a relevant cancer). Insurance companies cannot ask for the results of predictive genetic tests for the majority of policies.
- You can find more information about insurance from the Association of British Insurers
  - www.abi.org.uk/data-and-resources/tools-and-resources/genetics/genetic-testing/

Results

- If you decide to proceed with genetic testing, you will be notified of your results by post within approximately 6 weeks of submitting your saliva sample (see back page for sample instructions).
- If you would like to have support from a genetic counsellor while opening your results, please telephone the genetics helpline (+44 20 3437 6514).

There are three possible test results:

Gene fault found

- This gene fault likely played a part in the development of your breast cancer.
- A telephone appointment with a genetic counsellor will be scheduled for within one week of your result being posted to you. If you prefer, you may telephone the genetics helpline after receiving your result and speak to a genetic counsellor sooner, and at a time of your choosing.
- You will also be referred to your local clinical genetics unit for further consultation and ongoing management.

No gene fault found

- It is highly unlikely that your breast cancer was caused by a gene fault in one of the seven genes on this test
- Your risk of developing a new breast cancer in the future (not a recurrence) is the same as for any other woman with her first breast cancer at the same age as you and with your family history.
- It is unlikely that your close family members are at particularly high risk of breast cancer, unless you have a significant family history of breast cancer.

Variant of uncertain significance (VUS) found

Very occasionally the laboratory identifies a rare alteration in a gene, but they cannot classify this as either harmful or not harmful. This is called a VUS. Usually, as research advances, these alterations are reclassified as not harmful. Therefore, a VUS is treated the same as when no gene fault is found.

Additional information and support

Genetic counsellors are available to discuss any of this information in more detail. They are here to support you with any individual concerns or questions. To speak with a genetic counsellor, please call the genetics helpline.

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With extended hours to 7:00pm on Wednesdays

Alternatively, you may contact us by email at BRCADirect@rmh.nhs.uk

Below are some links for further information and support. These are generic resources, and some information may not be relevant to you.

- Breast Cancer Now: breastcancernow.org
- Macmillan: www.macmillan.org.uk
- Prevent Breast Cancer: preventbreastcancer.org.uk
- Royal Marsden NHS Foundation Trust, Beginners Guide to BRCA1 and BRCA2: patientinfolibrary.royalmarsden.nhs.uk/brca1brac2

Management of data and samples

The genetic testing will be completed by the Centre for Molecular Pathology at the Royal Marsden, part of the North Thames Genomic Laboratory Hub.

To understand and classify gene alterations, we need to compare information about the genetic alterations we have found nationally across the NHS and internationally (without sharing any patients' names). Sometimes, with new information, we will reclassify a gene alteration. You may be recontacted in the future if new information about the classification of your gene alteration becomes available.

Your DNA sample will be stored as per normal NHS laboratory practice. It may be used for future analysis (for example if additional gene tests are required) and/or to ensure that other testing (for example that of family members) is accurate and of high quality.

To Proceed with Genetic Testing, Please Complete the Steps Below

1. Consent Form

- Read and sign the Genetic Test Consent Form.
- Please note that we cannot process your sample if your Genetic Test Consent Form is incomplete

2. Personal Details Form

- Fill in the requested information

3. Saliva Sample

- Wait at least 30 minutes after eating or drinking before sample collection
- Wash your hands thoroughly
- Rinse your mouth well with water to clear away any food
- Fill the saliva collection tube (smaller tube in plastic wallet) with your saliva up to the line
- Screw on the lid and check it is tight
- Wash your hands again thoroughly
- Write the date of saliva collection on the adhesive label, then stick the label on the saliva tube
- Place the labelled saliva tube into the transport tube (larger tube with absorbent padding), making sure the transport tube is shut tightly
- Place the transport tube into the small padded envelope

4. Post from a Post Office

- Place both forms and the packaged saliva sample in the pre-paid plastic mailing bag
- Take the package to your local post office. You may find your nearest post office at: www.postoffice.co.uk/branch-finder
- This package may NOT be posted using a street post box

If you have any questions or would like to discuss further, please contact our genetics helpline at
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or email BRCADirect@rmh.nhs.uk