

Breast Cancer Genes

Information for patients



**Leeds Breast
Unit**

**Breast Family History and
Mainstream Genetics Service**

Information on breast cancer genes tested

In most people, cancer occurs by chance. Sometimes cancer occurs because of a genetic alteration (5 to 10% of cases) (Cancer Research UK, 2024). When genes are working properly, they protect us from getting cancer. When an alteration in a gene occurs, this increases the risk of getting cancers, including breast cancer.

If you carry one of these gene alterations, there is a 50% chance that the mutation is passed onto your children, if you have any. If you have one of these genetic mutations and you have already had a breast cancer, there is an increased risk of developing breast cancer in your other breast.

High Risk Gene Alterations

These include the **BRCA1**, **BRCA2** and **PALB2** genes and carry a 30-80% risk of breast and ovarian cancer.

A small percentage of people (around 1 in 400) (Breast Cancer Research UK, 2024) carry a mutation in the **BRCA1** or **BRCA2** genes. These are the most commonly inherited and heard of breast cancer genes.

A mutation in the **BRCA1** gene carries around an 80-85% lifetime risk of breast cancer, and around 40% lifetime risk of ovarian cancer for women. Men with the **BRCA1** gene have up to a 1% risk of breast cancer (Breast Cancer Now, 2022).

A mutation in the **BRCA2** gene carries around a 60% lifetime risk of breast cancer, and 17-20% lifetime risk of ovarian cancer for women. It also increases the risk of breast cancer in men up to 5-10% and prostate cancer up to 25% (Breast Cancer Now, 2022).

A mutation in the **PALB2** gene carries an increased risk (up to 50%) of breast cancer. It also carries a small increase in risk of ovarian and pancreatic cancer, and other cancers depending in family history (Cancer Research UK, 2024). PALB2 is a relatively new find in the world of genetics so there is not as much information about this gene compared to the BRCA genes.

Moderate Risk Gene Alterations

These carry a 1 in 6 or 17-30% lifetime risk and include the ATM, CHEK2, RAD51C and RAD51D genes (Breast Cancer Now, 2022).

A mutation in the **ATM** gene increases risk of developing breast cancer to a moderate level. An exception is the c.7271T>G specific alteration which gives an over 30% increased risk, tipping into the high risk category (Liverpool Women's NHS Foundation Trust, 2020).

A mutation in the **CHEK2** gene increases risk of developing breast cancer to a moderate level, specific percentages are family history dependent.

A mutation in the **RAD51C** and **RAD51D** genes increases risk of developing breast cancer by around 20%. It also increases the risk of ovarian cancer by around 11%. The risk level may be higher dependent on family history (UK Cancer Genetics Group, 2022).

Why am I being offered this test?

To look for genetic alterations that may explain the cause of your breast cancer diagnosis.

What are the benefits to me?

Knowing whether or not you carry a gene alteration gives your treating team more information about your cancer. This can help decisions about the treatments they recommend for you (eg – chemotherapy drugs or surgery options). It will also give you more information about your risk of developing cancer in the future.

What are the possible results of genetic testing?

There are three possible results.

1. **No gene alteration found** – this is the most likely outcome and means that a mutation in the BRCA1, BRCA2, PALB2, ATM, CHEK2, RAD51C and RAD51D genes has not been found. However, there may be other genes that increase breast/ovarian cancer risk that have not yet been identified.
2. **Cancer causing gene alteration found** – this confirms that there is a gene alteration present and there is a higher chance of developing breast and ovarian cancer. Your cancer team will use this information to inform your treatment options and further management.
3. **Variant of unknown significance** – very occasionally (<1%) we find a gene change, known as a variant, but the significance of this is unknown. The change may be harmless, and this is treated as a negative result.

What implications do the results have on family?

If no gene change is found, no-one else in your family will be offered a genetic test. However, your relatives may still be at an increased risk of developing cancer than the general population and may be eligible for further screening.

If a gene change is found, other members of your family will be eligible for a gene test to find out if they also carry the mutated gene.

If a variant of known significance is found, your family may still have an increased risk of breast/ovarian cancer due to family history.

Do I have to have the test?

No, this is completely optional. Your decision will not affect the standard of care that you receive. Some people decide that they do not want testing at the present time, but choose to wait until after their treatment is complete, or they do not want testing at all.

What if I can't decide?

You can be referred back to the to the Breast Family History and Mainstream Genetics Service at any point for further discussion.

What happens next if I say yes?

If you decide to go ahead with the test, you will be asked to sign two record of discussion forms. The test requires a blood sample which you can have taken in the Bexley Wing at St James's Hospital.

How do I receive the results?

The Breast Family History Team will give you the results and discuss management options if appropriate. This can be either over the phone, or a face to face appointment. You will then be referred to the Regional Genetics Team to discuss wider family implications.

If you have any further questions, do not hesitate to contact the Family History and Mainstream Genetics Secretary on:

0113 206 8890.

Meet the team

Kirtida Patel - Lead Breast Family History and Mainstream Genetics Specialist Practitioner

Siobhan Wilkinson - Breast Family History and Mainstream Genetics Specialist Practitioner

Lauren Burkill - Breast Family History and Mainstream Genetics Specialist Practitioner

Reference List

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