East Kent Hospitals University NHS Foundation Trust

Genetic testing for breast cancer susceptibility

Information for patients from the Breast Cancer Family History Clinic

This leaflet explains more about genetic testing for a breast cancer susceptibility gene. If you have any questions or concerns, please contact the Breast Cancer Family History Clinic.

Cancer in the general population

Cancer is a common condition. It will affect half of the people in the general population. For women in the general population:

- the lifetime risk (the chance of getting it at some point in life) of breast cancer is 1 in 8; and
- the lifetime risk of ovarian cancer is 1 in 50 women.

We know that the main thing that increases your risk (risk factor) is increasing age. The older we get the more likely we are to develop cancer, as our bodies become less effective at protecting us. Most cases (90 to 95%) happen by chance. A few (5 to 10%) happen when someone has inherited an increased risk of developing cancer.

What is inherited breast cancer?

In some families, we see more cases of breast cancer than we would expect to see by chance. We know that in some of these families, the cancer is due to an inherited (from our parents) susceptibility (risk of cancer). This can be caused by changes in genes such as BRCA1, BRCA2, PALB2 and CHEK2. These genes normally function to protect us from developing cancer.

What are genes?

Genes are coded messages which give instructions for how cells grow and function. Genes come in pairs. We all have two copies of each of these genes, as we inherit one copy from each of our parents.

The normal function of gene testing is to help protect us from developing cancer. A variant in one copy of a gene causes an increased cancer risk in people who carry it.

If someone has a variant in one copy of a gene, they have a one in two (50%) chance of passing on the variant to each child they have, whatever their gender or the child's gender.

What is a diagnostic genetic test?

A diagnostic genetic test involves taking a blood sample from you. We will examine your blood to see if you have a gene variant that we know is associated with inherited breast and possibly other cancers. The results will take up to 10 weeks to be reported. We can send them to you, call you with them, or you can come to the clinic to collect them.

We may suggest extra genetic tests to families based on their individual family history of cancer.

What are the possible results of breast cancer susceptibility gene testing?

- **Testing does not find a variant in one of the breast cancer susceptibility genes**

  If this happens, the chance of a breast cancer susceptibility gene variant being responsible for your history is small. However other genetic factors could be involved if you have a family history of cancer. We will not have a genetic test to offer your relatives who have not had cancer.

- **Testing finds an 'unclassified variant' in your breast cancer susceptibility genes**

  We have found a variant in one of your genes but we do not know if this is significant. This happens in about 1 in 20 cases. Future medical research may be able to tell us if these variants relate to a family history of cancer. We will not be able to use the test result to make decisions about cancer risk management for you or other family members.

- **A gene variant is found**

  This confirms an inherited susceptibility to breast and possibly other cancers.

BRCA1 and BRCA2

For women with a variant in the BRCA1 or BRCA2 genes, there is:

- an up to 70% (just over two thirds) lifetime risk of developing breast cancer, and
- an up to 45% (almost half) lifetime risk of developing ovarian cancer.

For women who have already been affected with breast cancer, we know there can be an up to 65% (two thirds) chance of them developing another breast cancer in the future. This can be influenced by the age of her first breast cancer diagnosis. Knowing that someone has a BRCA gene variant may help to direct potential treatment or surgical options.

Men with a variant in the BRCA2 gene may have:

- an up to 25% (quarter) lifetime risk of developing prostate cancer, and
- about an 8% (less than one in ten) lifetime risk of breast cancer.

Men and women with a BRCA2 gene variant have a small risk (three in 100) of pancreatic cancer.

PALB2

For women with a variant in the PALB2 gene there is an up to 55% (just over half) risk of breast cancer. Our knowledge about PALB2 is rapidly increasing, and advice could change over time. We recommend increased breast screening with mammography and / or MRI scans. The PALB2 gene also causes a possible increased risk of pancreatic cancer.

Specific risk figures and screening advice usually depends on family history. This should be discussed further with a genetic clinician.

CHEK2

Around one in 25 families with a strong family history of breast cancer are found to have a common variant in the CHEK2 gene. Women with a CHEK2 gene variant have about a 25% (one in four) risk of developing breast cancer in the absence of a close relative with breast cancer.

Specific risk figures and screening advice usually depends on family history. This should be discussed further with a genetic clinician.

ATM gene

The ATM gene is associated with an increased chance of developing:

- female breast cancer (around two to four times higher than the general population)
- pancreatic cancer, and
- prostate cancer in men.

Individuals with a variant in one copy of their ATM gene, would be at risk of these cancers, depending on their gender. We know that the risk of breast cancer in patients with a single ATM variant is also strongly influenced by a family history of breast cancer. This may lead to different breast cancer risk assessments in different ATM families.

You can get a clear risk assessment by assessing your full family history, and a calculation using a risk assessment tool.

RAD51C and RAD51D

These genes are associated with an increased risk of developing ovarian cancer, which is up to around 10% in females. If you have had a variant in this gene, it would be appropriate for you to consider surgery to remove your ovaries and fallopian tubes.

There may be a slight increased risk of breast cancer associated with a variant in these genes. Research is ongoing to try and quantify this risk.

What can you do if you have a gene variant?

Women who are carriers of a BRCA gene variant can have breast screening from the age of 30. The purpose of this screening is to help the early detection of breast cancer. In some families screening may start from the age of 25.

Unfortunately, we are not currently able to offer ovarian screening as it has not been proven to be effective. Some women who are carriers of a BRCA1 or BRCA2 gene variant may choose to have surgery to reduce their risk of developing a further cancer. This is done by removing as much as possible of the 'at risk' tissue, for example the breast and / or ovaries / fallopian tubes.

This is known as prophylactic (preventative) or risk-reducing surgery. This is obviously a very personal decision for any woman. Before making this decision, we would encourage you to discuss this with one of our breast or gynaecological surgeons. We want to make sure that this is the right decision for you. This can be accessed through the Hereditary Breast and Ovarian Cancer (HBOC) Family Service. See below for further information on the HBOC.

For the other genes in the panel test (such as PALB2 and CHEK2), screening is determined on a case-by-case basis. It is based on current national guidance and published information. We will discuss specific management options with you at your genetic counselling appointment.

If you have a gene variant you will be referred to the Cancer Genetics Service at Guy's Hospital.

What can be done if I decide not to have testing?

Not everyone who has been affected by cancer will choose to have genetic testing. If you decide not to have genetic testing, you and your family members may still be eligible for regular breast screening.

Things to think about

Deciding to have diagnostic genetic testing can be a difficult and complex process. You may wish to consider the points below before testing.

- **Timing**

  Why now? Will it change my immediate treatment? What other challenging events are happening in my life?

- **Psychological impact of knowing**

  How would I feel if a genetic variant was found? How would I share this information in my family?

- **Assess your own coping strategies**

  How have I dealt with difficult situations in the past? What strategies do I use to help deal with difficult news?

- **Next steps**

  What measures might I consider taking to address my risks of breast and ovarian cancer, if you find that I have a gene variant?

There are no right or wrong answers to these questions. Have a think about what your answers may be, so that when you get your result you have an idea of what you might do.

What support is available?

Undergoing diagnostic genetic testing and receiving the results can be a challenging time. Before making a decision or after receiving your results, you may wish to talk through:

- your feelings
- your coping strategies, and
- other any issues you may have.

Several support options are available to breast cancer gene carriers, including the following:

- your genetics clinician
- the HBOC Family Service
- BRCA support groups
- clinical psychologist
- your GP
- cancer charities, such as Breast Cancer Care (Someone Like Me service) and Macmillan.

Your genetics clinician will discuss these support options with you in more detail. Between you, you can work out what the best sources of support will be for you.

What is the HBOC Family Service?

We invite BRCA / PALB2 carriers to our HBOC Family Service clinic. At the clinic, you will be able to ask questions and discuss your management in detail with a team of specialists. If we find you have the variant, we will send you:

- an invitation to the clinic
- a leaflet about this service, and
- a psychological questionnaire to complete.

Can I take part in research studies?

Whatever your result is, there may be a chance for you to take part in a research study. Your nurse specialist can discuss this with you.

Contact details

If you have any questions or concerns, please contact the Breast Cancer Family History Clinic.

- **Breast Cancer Family History Clinic**
  Telephone: 01227 868666
  Email (ekh-tr.breastfamilyhistory@nhs.net)

Useful sources of information

- Breast Cancer Now (https://breastcancernow.org/)
  UK-wide charity providing care, information and support to people affected by breast cancer.
  Telephone: 0808 800 6000 (helpline, Monday to Friday 9am to 4pm, Saturday 9am to 1pm)
  Email (info@breastcancernow.org.uk)

- Someone Like Me (https://breastcancernow.org/information-support/support-you/someone-me)
  Service provided by Breast Cancer Now. Puts you in touch with a trained volunteer who has (or has had) primary breast cancer, or is undergoing genetic testing, for advice and support.
  Telephone: 0345 077 1893 (helpline, Monday to Thursday, 9 to 5pm, Friday 9 to 4pm)
  Email (someonelikeme@breastcancercare.org.uk)

- Macmillan Cancer Support (https://www.macmillan.org.uk/)
  Support and advice charity for people affected by cancer.
  Telephone: 0808 808 0000 (7 days a week, 8am to 8pm)

[Web sites last accessed 11 December 2025]

This leaflet has been produced with grateful acknowledgement to the Cancer Genetics Service at Guy's Hospital.

We have used the terms 'men' and ‘women’ throughout this leaflet. When we use these terms we also mean people with male or female reproductive organs who do not identify as a man or woman. East Kent Hospitals is committed to supporting people of all gender **identities**. Please tell your midwife or doctor how you would like them to address you, so we can be sure to get this right.

What do you think of this leaflet?

We welcome feedback, whether positive or negative, as it helps us to improve our care and services.

If you would like to give us feedback about this leaflet, please fill in our short online survey. Either scan the QR code below, or use the web link. We do not record your personal information, unless you provide contact details and would like to talk to us some more.

Giving feedback about this leaflet

https://www.smartsurvey.co.uk/s/MDOBU4/

If you would rather talk to someone instead of filling in a survey, please call the Patient Voice Team.

- **Patient Voice Team**
  Telephone: 01227 868605
  Email (ekhuft.patientvoice@nhs.net)

This leaflet has been produced with and for patients.

Please let us know:

- If you have any accessibility needs; this includes needing a hearing loop or wanting someone to come with you to your appointment.
- If you need an interpreter.
- If you need this information in another format (such as Braille, audio, large print or Easy Read).

You can let us know this by:

- Visiting the Trust web site (https://www.ekhuft.nhs.uk/ais).
- Calling the number at the top of your appointment letter.
- Adding this information to the Patient Portal (https://pp.ekhuft.nhs.uk/login).
- Telling a member of staff at your next appointment.

Any complaints, comments, concerns or compliments, please speak to a member of your healthcare team. Or contact the Patient Advice and Liaison Service on 01227 783145 or email (ekh-tr.pals@nhs.net).

Patients should not bring large sums of money or valuables into hospital. Please note that East Kent Hospitals accepts no responsibility for the loss or damage to personal property, unless the property has been handed into Trust staff for safe-keeping.

Further patient information leaflets are available via the East Kent Hospitals' web site (https://www.ekhuft.nhs.uk/patient-information).

Reference number: Web 709

First published:	Last reviewed:	Next review date:
July 2024	October 2025	November 2027

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