Current research suggests that for women who carry a PALB2 gene alteration, the breast cancer risks are relatively high, in the order of 30-60% over a lifetime.

Gene carriers can be offered extra breast screening and may wish to discuss breast surgery to reduce their cancer risk, dependent on their family history. Your clinician will discuss this with you further.

Men who carry a PALB2 gene alteration may have a small increase in the risk of male breast cancer but overall this risk remains low.

Men and women may have a small increase in their risk of pancreatic cancer. We do not currently recommend screening for pancreatic cancer outside of a research study as this screening is invasive and can be difficult.

Further information
PALB2 Interest Group
This research group aims to gather more information to help improve our understanding of the effects of the PALB2 gene.
add-tr.palb2@nhs.net
www.palb2.org

Macmillan Cancer Support
Tel: 0808 808 0000
www.macmillan.org.uk

Breast Cancer Care
www.breastcancercare.org.uk

Feedback
We appreciate and encourage feedback. If you need advice or are concerned about any aspect of care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR
www.nuh.nhs.uk

If you require a full list of references for this leaflet please email patientinformation@nuh.nhs.uk or phone 0115 924 9924 ext. 67184.

The Trust endeavours to ensure that the information given here is accurate and impartial.

Testing for the PALB2 gene for familial breast cancer
Information for patients
Clinical Genetics

This document can be provided in different languages and formats. For more information please contact:

Clinical Genetics Service
Nottingham City Hospital
The Gables, Gate 3
Hucknall Road
Nottingham NG5 1PB

Tel: 0115 962 7728
Email: nuhnt.clinicalgenetics@nhs.net
How are genes involved in inherited breast cancer?
We know that of all women who develop a breast cancer, only about 5% may have an inherited genetic cause.

For some years we have been able to offer genetic testing for two high risk genes known as BRCA1 and BRCA2 (BReast CAncer 1 and BReast CAncer 2) in some families with a history of breast and/or ovarian cancer. You may have had this testing yourself in the past to try to find out if there could be a genetic (inherited) explanation for your own cancer diagnosis.

We know that there will also be other genes which could provide the explanation for some cases of familial breast cancer. Some of these genes we have yet to learn about and some have been discovered over the last few years.

What is the PALB2 gene?
Research has shown that changes in the PALB2 gene can be an explanation for some cases of familial breast cancer.

PALB2 gene works together with BRCA1 and BRCA2 genes in the same DNA-damage response pathway, normally helping to protect from breast and other cancers. Current research does not suggest that PALB2 is linked to ovarian cancer risk.

Who can have genetic testing for PALB2?
Your clinician will discuss whether it might be appropriate to test you for alterations in this gene, based upon your family history. We may be able to use a stored DNA sample we already have to complete this test.

How is the PALB2 gene inherited?
Alterations (mutations) in the PALB2 gene are inherited in the same way as the BRCA1/2 gene (dominant inheritance). Therefore, if you are found to have an alteration in PALB2, your children would have a 50% (1 in 2) chance of also inheriting this. If a gene change is found in the family, other family members can then seek genetic testing to clarify their own risks (predictive genetic testing).

What are the possible results of PALB2 testing?
Testing may take up to three months and there are 3 possible results from this type of genetic testing

1. No gene changes are found
This means that it is unlikely that the cancer is caused by a mutation in the PALB2 gene. No predictive testing is therefore available to unaffected relatives. In this situation, there may still be other genes that might increase the risk of breast cancer.

Your clinician will review whether any other testing would be appropriate for you or your family and discuss ongoing breast screening recommendations.

2. A PALB2 gene change (mutation) is found
This confirms that an inherited tendency to breast cancer is present in the family. Women with breast cancer who carry this change will have a higher chance of developing a new breast cancer.

Other members of the family can have a predictive genetic test to see if they have inherited the same gene change (mutation).

3. An ‘unclassified’ gene change is found
This is a gene change of unknown significance which may be the cause of the increased cancer in the family or may be entirely harmless. This type of result cannot be used for predictive testing for relatives.

Relatives may still qualify for additional breast screening, based upon the family history.

How might the test result affect me and my family?
We do not know as much about the PALB2 gene as we do about other genes such as BRCA1 and BRCA2 and so we are still learning about the cancer risks involved.

Currently there are no clear NHS guidelines on the best way to screen and manage the cancer risks for carriers of PALB2 gene changes. Your clinician will discuss this with you further based upon your family history.