

## **PALB2 gene alteration identified: Patient Information**

### **Why have I been given this information sheet?**

You have been given this information sheet because you, or someone in your family, has been found to have an alteration in a gene called *PALB2*. This sheet should accompany a letter from our Genetics Service which will have additional information and recommendations specific for your family.

### **What is *PALB2*?**

*PALB2* is a gene that is known to be connected with breast cancer risk.

### **What cancer risks are there if you have a *PALB2* gene alteration?**

The table below outlines the cancer risks associated with having *PALB2* gene alteration.

	<b>Breast cancer</b>	<b>Ovarian cancer</b>	<b>Pancreatic cancer</b>	<b>Male breast cancer</b>
<b>By age 50 with a <i>PALB2</i> gene alteration</b>	13-21%	<1%	<1%	<1%
<b>By age 80 with a <i>PALB2</i> gene alteration</b>	44-63%	Approximately 5%	2-5%	1%
<b>Population risk</b>	15.3%	2%*	Approximately 2%*	<1%*

\*Cancer Research UK estimated lifetime risk of being diagnosed with cancer for people born after 1960 in the UK.

If you have had breast cancer, the *PALB2* gene alteration is likely to be the main explanation for this. There may also have been other factors, both genetic and environmental, as well as an element of chance.

### **What cancer surveillance is available for individuals with a *PALB2* alteration?**

Women who have a *PALB2* gene alteration are offered high risk breast screening. This involves:

- Annual magnetic resonance imaging (MRI) scan from 25-39 years
- Annual MRI AND mammography from 40-50 years
- Annual mammography from 51-71 years (some women may continue to receive MRI too)

For women who have a 50% chance of inheriting a *PALB2* gene alteration but do not want a genetic test, their level of breast screening is worked out based on their personal family history.

Most NHS routine or high risk breast screening stops after the age of 70 or in some places 73. You can still have screening after this, and can arrange an appointment by contacting your local breast screening unit. This is something we would recommend if you were a carrier of a gene alteration. We also suggest that women carry out monthly self-examination of the breasts.

The aim of breast surveillance is to detect breast cancers at an earlier stage, when the cancer has a better prognosis and is easier to treat. However, surveillance is not perfect and may miss cancers. In addition, sometimes changes are seen on the scans that later turns out not to be a cancer, but do cause worry and can lead to unnecessary extra tests. We are not currently recommending surveillance for other cancers.

## **What preventative surgery options are available for individuals with a *PALB2* alterations?**

We know that your risk of cancer will vary according to your family history. If you have a strong family history of breast cancer you may wish to consider preventative surgery of the breasts (also called risk reducing mastectomy). If you have a close relative affected with ovarian cancer then preventative removal of the ovaries may also be an option to consider. Please talk to your Genetic Counsellor or Clinical Geneticist if you would like to look into this further.

## **How is the *PALB2* gene alteration inherited?**

We have two copies of every gene including *PALB2*. We give one copy of each gene to our children in the sperm and one copy in the egg. Someone with a *PALB2* gene alteration usually has this alteration in only one copy of *PALB2*. They also have a normal copy of the *PALB2* gene. Their children can inherit either the normal or the altered copy. This means they have a 50% (1 in 2) chance of having the same gene alteration.

## **Who should be offered a test for the *PALB2* gene alteration?**

Genetic testing for the *PALB2* alteration may be helpful for your relatives. Genetic testing is only offered through a Genetic Service. Your Genetic Counsellor or Clinical Geneticist can provide a letter for your relative to take to their GP to request a referral to Genetics.

## **What can I do to help my risk if I have a *PALB2* gene alteration?**

We recommend all women minimise risk of breast cancer by maintaining a healthy diet and taking plenty of exercise. You should also report any changes in your breasts to your GP. If you have a personal or family history of breast cancer you should discuss this with your GP before taking hormone replacement therapy.

## **Where can I get further information?**

The *PALB2* website [www.PALB2.org](http://www.PALB2.org) is run by the *PALB2* Interest Group and provides further information to families and health professionals.

## **Are there any research studies for *PALB2*?**

If you would like to know more about research please contact the *PALB2* Interest group through their website [www.PALB2.org](http://www.PALB2.org).

If you need more advice about any aspect of PALB2 genetic testing, you are welcome to contact:

Liverpool Centre for Genomic Medicine  
Liverpool Women's Hospital NHS Foundation Trust  
Crown Street  
Liverpool  
L8 7SS  
Telephone: 0151 802 5001 or 5008

Email: [lwft.clingen@nhs.net](mailto:lwft.clingen@nhs.net)

This leaflet can be made available in different formats on request. If you would like to make any suggestions or comments about the content of this leaflet, then please contact the Patient Experience Team on 0151 702 4353 or by email at [pals@lwh.nhs.uk](mailto:pals@lwh.nhs.uk)

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