

# Genetic testing of the BRCA1 and BRCA2 cancer genes (tumour test)

We have given you this factsheet because you have been diagnosed with or are currently being investigated for ovarian, peritoneal or fallopian tube cancer. If you are diagnosed with one of these cancers, we would like to offer you a genetic test to identify any variants that can help to plan the best treatment for you, as each person's cancer has a unique combination of genetic variants (mutations).

We hope this factsheet will help to answer some of the questions you may have about genetic testing. If you have any further questions or concerns, please speak to a member of our team.

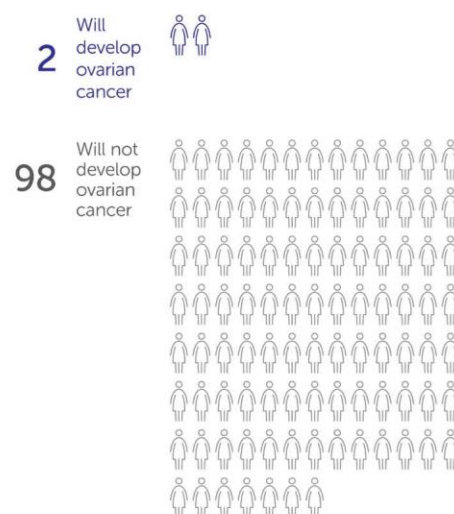
## What is cancer?

Cancer is a condition where cells in a specific part of the body grow and reproduce uncontrollably. This may cause a growth called a tumour. The cancerous cells can invade and destroy surrounding healthy tissue, including organs.

It is a common condition which will affect up to one in two people in the general population during their lifetime. In the UK, around two in 100 women will develop ovarian cancer.

### Estimated risk of developing ovarian cancer (ICD-10 C56-C57.4) in lifetime

Women born after 1960, UK



Some cancers start due to inherited faulty genes passed down in families (this is called hereditary cancer and is tested for separately using a blood sample). However, most cancers start due to gene changes that happen naturally as you get older.

### **What are genes?**

Genes are our cells' instruction manuals. We each have around 20,000 pairs of genes which are present in almost every cell of our bodies. Our genes tell our cells how to function normally.

### **What are the BRCA1 and BRCA2 genes?**

Everyone has two copies of the BRCA1 and BRCA2 genes (Breast Cancer genes). These genes are normally protective against cancer because they help repair breaks in DNA that can lead to cancer. However, variants can occur in these genes randomly during your lifetime and along with other factors such as your lifestyle, this can increase your risk of developing breast cancer or ovarian cancer.

### **How does genetic testing work?**

Most of the time, cancer-causing genetic variants are found only in the cancer cells (the tumour itself). These are called 'somatic variants' and are not present in the normal healthy cells of a person who has cancer. Somatic variants cannot be inherited.

The genetic test we are offering you looks for somatic variants in the BRCA1 and BRCA2 genes in your tumour. It also looks for other similar somatic variants in the tumour that are unable to repair damaged DNA.

### **Why am I being offered a genetic test?**

Genetic testing of your tumour will help your oncologist (cancer specialist) or surgeon plan the best treatment for your cancer.

### **What does the genetic test involve?**

We will take a sample of your tumour during a biopsy or surgery, which our pathology department will then send to a genetic laboratory so that it can be tested for somatic variants in the BRCA1 and BRCA2 genes. The result will be available in about four to six weeks and will be sent back to the doctor who organised the test.

### **What are the possible outcomes of testing?**

#### **A BRCA1 or BRCA2 variant is found in your tumour sample**

We will use this information to help guide your treatment. We will compare the findings with a genetic blood test to check whether this variant is inherited.

#### **A BRCA1 or BRCA2 variant is not found in your tumour sample**

We will use this information to help guide your treatment. We will compare the findings with a genetic blood test to check there is also no variant in your blood sample.

### **How will finding a BRCA1 or BRCA2 variant affect my treatment?**

We will use this information to help decide on the best treatment plan for your cancer. We may prescribe you a medication called a 'PARP inhibitor'. This medication has been shown in general to improve the response to cancer treatment in women with ovarian cancer who have a BRCA1 or BRCA2 variant. In certain circumstances, we may be able to offer this medication to women who do not have a BRCA1 or BRCA2 variant.

### **What will happen if I decide not to have the tumour test?**

If you decide not to have the tumour test (or the blood test) we will have less information to help us decide on the best treatment plan for you. However, we will offer you the most appropriate treatment for your type of cancer.

### **Contact us**

If you have any further questions or concerns, please contact us.

*Hilary Maxwell*

*Gynae-Oncology Clinical Nurse Specialist*

*07340 528902*

**For a translation of this document, or a version in another format such as easy read, large print, Braille or audio, please contact your Gynae-Oncology Specialist Nurse.**

### **Useful links**

[www.nhs.uk/conditions/ovarian-cancer](http://www.nhs.uk/conditions/ovarian-cancer)

[www.targetovariancancer.org.uk/about-ovarian-cancer/hereditary-ovarian-cancer](http://www.targetovariancancer.org.uk/about-ovarian-cancer/hereditary-ovarian-cancer)

<https://eveappeal.org.uk/our-research/our-research-programmes/brca-protect/>

<https://eveappeal.org.uk/inherited-risks/ask-eve-genetic-testing-in-ovarian-cancer/>

### **Wessex Clinical Genetics Service provided by:**

University Hospital Southampton NHS Foundation Trust