Breast Cancer Genomic Testing (R208/R444.1)

Patient Information sheet

Cancer occurs by chance in most people. Around 5-10% of people with breast cancer carry a variant in a cancer gene (also sometimes called a gene change or gene alteration) that gives an increased chance of developing of breast cancer, but may also be associated with an increased chance of other cancer.

Why am I being offered this test?
Your specialist has offered to arrange testing for you because you have been diagnosed with breast cancer. This test will look for variants in a group of genes that are associated with a significantly increased risk of breast cancer. If you are found to have a variant in one of these genes, there may be different treatment options available to you now or in the future. The result of this test may also give information about your chance of developing another cancer in the future.

How is testing arranged?
Your specialist will discuss the process with you, including the possible outcomes, which are outlined below. You will be asked to sign a consent form and a blood sample will be required for the test. The result of your blood test will be ready in around 6-8 weeks, possibly sooner. Your specialist will contact you with the results.

What are the possible results from having this blood test?
This test can reveal one of three results:

- **A gene variant is found**. If a gene variant that stops the gene working is found, this will confirm an inherited tendency to breast and possibly other cancers, such as ovarian cancer. This result may alter the oncology treatment options available to you now or in the future and your specialist will discuss these with you.

  It may also mean that you have an increased chance of developing another cancer. The main risk with these genes is for breast cancer, but there can sometimes be an increased risk of ovarian or other cancers, depending on the gene involved.

  If testing shows that you have a gene variant your specialist will refer you to the Clinical Genetics service. You will be offered an appointment within a few weeks to discuss what this result means regarding your future risk and the options that you may want to consider to manage this risk. At an appointment you will be asked for details of your family history, which in combination with your test result helps determine what options may be appropriate for you.

- **No gene variants are found.** This means it is unlikely that your cancer was caused by a variant in one of the genes tested. You should review with your specialist whether you require a referral to Clinical Genetics.

  If you have relatives who have been diagnosed with cancer, depending on the type of cancers, and the ages at which they were diagnosed, your specialist may discuss a

referral to the Clinical Genetics service, or suggest that your relatives seek a referral. Your result will be reported in line with current genetic knowledge and guidelines. Further testing may be an option for you in the future. This can be reviewed if you are referred to the Clinical Genetics Service.

- **A 'variant of unknown significance' is found**. This is an uncertain/inconclusive result. In this situation your specialist will review with you what implications this may have for your treatment options.

  Your specialist will discuss the result with the Clinical Genetics service. You may be offered an appointment to discuss your result and whether any further investigations are possible to clarify the implications for you and your relatives. This is more likely if you have relatives who have been diagnosed with cancer. Alternatively, the Clinical Genetics service may give your specialist advice about the implications for you and your family.

Does this testing have implications for my family?
If you are found to carry a gene variant it is possible that some of your relatives may carry the same gene variant and they will have the option of being tested. Relatives who carry the same gene variant will be offered personalised information about what risk management options, such as screening or risk-reducing surgery, are available to them.

If your test does not identify any gene variants, this will be reassuring for your relatives. If you do not have a family history, and are not under 40 years old, there will not usually be an increased risk for close relatives and a referral to Clinical Genetics will not be needed. However, if there are any changes in your family history in the future a referral can always be considered at that time.

Breast awareness
It is important for all women to be breast aware, and to discuss any concerns with their GP. It is also important to take part in the National Screening Programmes as they become available.

Where can I find more information?
If you have questions regarding genetic testing you can discuss them with your specialist or you can contact the All Wales Medical Genomics Service.

South East Wales (Cardiff) – 02921 834000
South West Wales (Swansea) – 01792 285972
North Wales (Wrexham) - 03000 858 477

For more information please visit our website:

https://medicalgenomicswales.co.uk/