Gene testing after a diagnosis of breast cancer

You are being offered a genetic test to see if the breast cancer you have been diagnosed with has been caused by an error in one of your genes. This information leaflet has been written to help you decide whether having the test is the right option for you.

Background

Genes act as instructions that tell the body to make or do something. We inherit genes from our parents and sometimes they have an error that can be passed to us and increase our risk of diseases, such as cancer.

The test that you have been offered will look at a number of genes called BRCA1, BRCA2, PALB2, ATM and CHEK2. These genes help to protect us from developing certain cancers. If a person carries an error in one of these genes, some of that protection is lost, and it is more likely that a cancer will develop during that person's lifetime.

Why am I being offered this test?

You are being offered this test because the details of your cancer, and/or your family history, mean there is an increased chance that you carry an error in either the BRCA1, BRCA2, PALB2, ATM or CHEK2 genes. The test will check if this is the case, so that you can be given more accurate information about your current cancer and the chance of you developing a new breast or ovarian cancer in the future. The test will also help clarify the risks for your blood relatives.

Just because you are eligible for the test does not mean you will have an error in one of these genes. In fact, 9 out of 10 patients tested do not have a gene error.

What does the test involve?

If you decide to have the test, a blood sample will be taken from a vein in your arm. The laboratory will then test for any errors in your BRCA1, BRCA2, PALB2, ATM and CHEK2 genes. If you are aged 30 years or younger (or you are between 31 and 35 years and have a particular type of 'triple positive' breast cancer), we will also test for errors in a gene called TP53, which can cause cancer in very young women. The results will take up to six weeks and your breast team will inform you once the results are available.

What will the results mean?

1. **Testing may find an error in one of the three tested genes:**
   If we find an error in one of your genes, it would explain why you (and perhaps other family members) developed cancer. This result may also mean that there is an increased risk of developing another new breast or ovarian cancer in the future. If a gene error is found, your breast team will refer you to the specialist genetic service.

This specialist team will provide further information about the test results, and support you to make decisions to help protect your future health. The genetics team can also help you plan how to discuss this result with your family members.

2. **Testing may not find an error in any of the three tested genes:**
   If testing does not find an error in any of these genes, we will not have found an explanation for your cancer or the cancers in your family. This result would not completely exclude the chance of you or other family members developing future cancers, but it would mean it is less likely.

3. **Testing may find a change in one of the three genes but we are unsure whether it is an error:**
   Doctors and researchers are still learning about how genes affect cancer development and sometimes we find a change in a gene that we do not fully understand. This is known as a 'variant of unknown significance' because there is not enough information to say whether it causes a higher risk of cancer or not. For short, a variant of unknown significance is called a VUS. If a VUS is found, we do not change your treatment or test your family, but your breast team will ask the genetics laboratory to review your case in 5 years' time, to see if researchers have discovered any new information about the VUS found in your genes.

What are the possible benefits of having the test?

If a gene error is found soon after a diagnosis of breast cancer, your medical team may recommend different surgery or medication. You may also be given options to lower the chance of another cancer developing in the future, such as medication, or surgery to remove the breasts and sometimes, for females, the ovaries as well.

If we identify an error in your genes, we will suggest that your family members are referred to their local genetics service to discuss the option of genetic testing and, if appropriate, they may be offered regular monitoring, surgery or medication to reduce their risk of developing cancer.

If no errors are found in any of these high-risk genes, then your cancer is less likely to be due to an inherited genetic problem. This means the risk of you, or one of your blood relatives, developing a new cancer in the future would not be so high. Many patients feel reassured to know that the cancer they developed was not due to a gene error.

What are the possible negatives of having the test?

If a gene error is found, you may worry more about your own risk of developing a new cancer in the future. You may also worry about the risks to your family members and how best to share this information with them.

If no gene errors are found, then other family members will not be able to have a genetic test to help clarify their risk but may remain worried. Depending on the details of the cancers in your family, we may suggest relatives are referred to a family history clinic anyway, to assess if they are able to access extra screening.

Your blood sample will continue to be stored in case new scientific knowledge leads to the option of further genetic testing in the future.

As with all tests, there is a very small chance that the result of the test is wrong. However, lots of checks are in place to make this extremely unlikely.

Some people decide that having information regarding a possible genetic cause for their breast cancer is not helpful at an already difficult and emotional time. You do not have to have genetic testing and your treatment and care will not be affected by this decision.

Consent

- I agree to have testing of BRCA1, BRCA2, PALB2, ATM and CHEK2 genes. The TP53 gene will also be tested in women aged 30 years or younger and women between 31 and 35 years who have 'triple positive' breast cancer.
- I understand that this test is not intended to diagnose whether I have or will get a cancer in the future. It is intended to tell me about my inherited genetic risk.
- I understand I will have the opportunity to discuss the test, its results and consequences in more detail.
- I understand my sample will be stored for possible future testing.
- I understand that my result and/or sample may be shared with health care professionals across the UK to help interpret genetic results and clarify the risks for others including other family members.

Signature (patient)	
Name (print)	
Date	

Signature (clinician)	
Name (print)	
Date	

Please retain the original copy of the consent form in the patient's notes and provide a photocopy for the patient to take home

You can also watch a video about the testing using this QR code (open your phone/tablet camera, scan the code and click the link). Alternatively you can access the video here:

https://youtu.be/uzsyxl_6elw