



**Birmingham Women's
and Children's**
NHS Foundation Trust

Information for people who have a
personal and family history of cancer

Diagnostic Genetic Testing



By your side

What is a genetic test?

If you have a family history of cancer and have had the type of cancer which can be associated with inherited disease, we may have talked to you about the possibility of having a genetic test.

When we carry out the first genetic test in a family we usually test someone who has had cancer. Not only does this increase our chances of finding an alteration if there is one present, but the result is easier to interpret. This is called a diagnostic test because we are trying to diagnose the problem in a family.

A genetic test involves taking a small sample of blood. We would look at your sample to see if we can find an alteration in one of the genes which is linked with inherited cancer. Such an alteration may explain the cancer in your family. If we find that you have an altered gene, other members of your family can be tested to see if they have inherited the same altered gene as you.

If there is only a small chance that you have an altered gene, we may not test your sample now but suggest that we store a sample of your blood to test in the future when our understanding of the causes of cancer improves.

What is involved in genetic testing?

After you have had a chance to discuss what genetic testing means for you and your family, you may decide to give a blood sample. We will ask you to sign a consent form and we will take a small blood sample from you (about two teaspoons full). The laboratory team will then search through the relevant genes in your DNA from your blood sample to see if the code in these genes is different from what we would usually see.

When will you hear from us?

Looking for alterations in genes can take quite a long time and may be hard to find. If you would like to know the result, we will contact you, usually in several months, when we have finished our analysis.

The result is confidential to you but we would discuss how the information could help other members of your family to make decisions about the best ways to manage any risks.

What do the results mean if an altered gene is not found in your sample?

For you

It would confirm that your cancer was probably due to an altered gene that has been passed down to you from one of your parents. You may be at an increased risk of getting a second cancer so we will talk to you about the best ways to reduce any risks. Some people find it helpful to know why they got their cancer. Some may feel guilty about possibly passing it on to their children. This is a very normal response.

For your family

If you have any children each of them would have a 1 in 2 chance of inheriting the altered gene from you. Any of your brothers or sisters may also have the same 1 in 2 risk of having the gene.

We would be able to offer testing to other members of your family to see whether or not they have inherited the same genetic alteration as you.

Having an altered gene does not mean you will get cancer, but the risks may be high. Therefore we will talk to you about the best ways to manage the risks.

What do the results mean if an altered gene is not found in your sample?

Unfortunately, if we do not find an altered gene in your blood sample, we cannot give you a definite answer about whether or not your cancer is inherited. There are three reasons why we do not find gene alterations:

1. Your cancer may not have been inherited
2. You may have an alteration in another cancer gene that has not been discovered yet
3. You may have an alteration in one of the genes that we tested, but we have been unable to find the alteration.
4. Sometimes we find what we call a Variant of Unknown Significance (VUS). This may be a very small change and we don't know if it stops the gene from working or not.

We would store your sample unless you ask us not to. This is because if in the future we discover more cancer genes, we may be able to carry out further tests on your blood sample.

For your family

If we do not find an altered gene in your sample it means that other members of your family may still be at increased risk of getting cancer. However, we would not be able to offer them genetic testing.

It would be important for them to continue with any screening that has been arranged for them.

What can be done to detect cancer early?

Screening

If you have had cancer and are still seeing a specialist, he or she will help you to decide how often you should be screened if this is necessary. If you are no longer seeing a specialist but there is a chance that your cancer could have been inherited, we will be able to help to arrange regular screening for you if this is necessary.

Awareness

It is important to know what is normal for you and if you notice any unusual symptoms to report them to your GP or specialist promptly.

What can be done to reduce cancer risk?

In some cases surgery may be an option to reduce the risk of cancer. We would be able to talk to you about this, if appropriate, and refer you to a specialist surgeon. There is evidence that leading a healthy lifestyle can help to reduce the risk of cancer. In particular, a balanced diet combined with regular exercise is recommended, although the precise role of these factors in modifying the risk of cancer is not known. Not smoking and reducing alcohol is a good idea, in line with general health advice.

How can you help?

Research studies

There is still much we do not know about the genetics of familial cancer. There may be research studies which you could take part in to help with our understanding of inherited disease and in the early detection of cancer. Please ask your genetic professional for information if this is something you are interested in.

Where can I get further information?

Some information about cancer can be found on the following websites. You may wish to wait until after your appointment so that we can direct you to information most appropriate to you.

www.cancerresearchuk.org

www.macmillan.org.uk/Home.aspx

If you need more advice please contact:

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Reviewed and updated: June 2017

Next review: June 2020

Ref No: CG16/DT

