



Gene testing after a diagnosis of medullary thyroid cancer

You have been diagnosed with medullary thyroid cancer and we would like to offer you a **genetic test** to see if your cancer 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.

What are genes?

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.

Why am I being offered this test?

Some people who have medullary thyroid cancer have an error in a gene called **RET**. The test that you have been offered will look at the RET gene. If you have an error in this gene, it means you have a condition called **Multiple Endocrine Neoplasia type 2 (MEN 2)**. MEN 2 increases your lifetime risk of certain tumours (both cancerous and non-cancerous) of the endocrine system.

If we know that you have an error in the RET gene it means that we can give you **more accurate information** about your current cancer, and **the chance of you developing future tumours** in other parts of the body. For example, the adrenal glands (small glands just above each kidney) and parathyroid glands (small glands in the neck). We can also offer **monitoring** with an appropriate specialist to make sure that any abnormalities in the future are found quickly. The test will also help us **understand the risks for your blood relatives**.

Just because you are eligible for the test **does not** mean you will have an error in the RET gene.

How is the test done?



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 an error in your RET gene. The results will take up to twelve weeks and a member of your oncology team will tell you once the results are available.

What will the results mean?

1. Testing may find an error in the tested gene:

If we find an error in your RET gene, it will **explain why you developed cancer**. This result may also mean that you have a **higher risk** of developing a tumour of the adrenal glands or parathyroid glands. Tumours can be cancerous, or non-cancerous. If a gene error is found, your oncology team will refer you to the specialist genetics 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.

If you have advanced medullary thyroid cancer, finding an error in the RET gene may affect the treatment you are offered. Your oncology team will discuss this with you.

2. Testing may not find an error in the tested gene:

If testing does not find an error in the RET gene, we will not have found an explanation for your cancer and it is likely that **your cancer developed by chance**. This result would not completely rule out the risk of you developing other cancers in the future, but it would mean it is less likely.

3. Testing may find a change in the tested gene but we are unsure whether it is an error:

Occasionally the test may give a result that is difficult to interpret. If this happens, you may need to be referred to the specialist genetics service to discuss your results and decide whether further tests are needed.

What are the possible benefits of having the test?



If we identify a gene error, your oncology team will refer you to the specialist genetics service. They will talk to you about additional tests you may need to check for tumours in any other organs, and any monitoring tests that could be helpful. They will also talk to you about whether your family members could be referred to the specialist genetics service to see if they also have an error in the RET gene.

If no error is found in the RET gene, 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 as 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 need to have a genetic test but may still be worried.

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 medullary thyroid cancer is not helpful at an already difficult and emotional time. You do not have to have genetic testing and your ongoing thyroid oncology care will not be affected by this decision. If you change your mind in the future, the genetic test could be offered at a later date. The process may, or may not, be similar to what is described above depending upon updated guidance in the future.



Consent

- I agree to have testing of the RET gene.
- 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.

Signed (patient) _____

Name (PRINT) _____

Date _____

Signed (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