



**The Leeds
Teaching Hospitals**
NHS Trust

Yorkshire Regional Genetics Service

Genetic Testing

Information for patients



Yorkshire Regional
Genetics Service

Genetic testing finds changes in genes that can cause health problems.

Why have I been offered a genetic test?

You have been offered a genetic test because your health condition could be due to a change in one (or more) of your genes. Genetic testing may help to decide if this is the case.

What is a gene?

Human bodies are made up of billions of tiny “building blocks” called cells. Almost every single cell contains our entire genetic code.

Our genetic code contains over 20,000 genes. Genes are the instructions which tell our body how it should be made, grow, and develop.

These genes are stored on structures called chromosomes. We have 46 chromosomes which are arranged into 23 pairs. Each chromosome contains thousands of genes.

We all have two copies of each chromosome and gene. One copy has been inherited from our biological mother and the other from our biological father.

What testing can be done?

Genetic testing can look for either changes within a gene or changes to the chromosomes which may be associated with your condition.

Your doctor will explain which type(s) of genetic testing they recommend.

How may finding a gene change help me?

Genetic testing may:

- Help to confirm your diagnosis.
- Provide further information about your condition and whether you need to have more tests.
- Guide the doctors looking after you in deciding whether certain treatments are appropriate.
- Provide more options for you if you are planning a family and in pregnancy.

How may finding a gene change in me help my relatives?

Finding a gene change can help doctors to work out the inheritance pattern of your condition (how the condition passes through a family). This means we can tell you the chance of your family members having the same condition.

Where a gene change is identified, a genetic test may be available for your relatives, should they wish to know whether they have inherited the condition.

Depending on the gene and the condition, we may not offer gene testing to children until they are old enough to make the decision themselves.

Do you sometimes find uncertain gene results?

Yes - we all have changes in our genes which is part of normal variation in the general population. Most of these do not have any health implications.

It may not be clear whether these types of gene changes are causing your condition or not.

We cannot test family members for these types of changes.

Could there be any unexpected results from gene testing?

Yes - sometimes we find gene changes which we were not expecting. These are called 'incidental findings.' These findings are not the cause of your condition but may be important for you if they are associated with other health implications. Your doctor can discuss these further with you.

What does it mean if my test doesn't find any changes?

If your test does not find any gene changes, it makes it less likely that you have the condition that has been tested for.

A negative test does not completely exclude that you have the condition. This is because there can be a change in a gene that our current testing has not been able to identify, or a change in a gene that we have not tested for.

What will the testing involve for me?

A routine blood sample is required to do the testing – your doctor will arrange this.

Genetic material (DNA) from this sample will be stored in the NHS genetics laboratory.

Results and samples may be used for counselling other family members in relation to this condition.

How long will it take to get my results?

The length of time it takes to get a result will vary. Most genetic testing will be complete within 6-12 months, though some testing may take longer than this.

If you do not receive a result within 12 months, please contact the doctor who arranged the test.

Once a specific gene change is known in the family, further testing in your relatives is much quicker.

Who should I contact if I have questions about this?

Please contact the doctor who arranged your genetic test.



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