

Whole genome sequencing for a rare disease

Information for patients
and family members



**Genomic
Medicine Service**

This booklet is for patients and their family members who may be offered a whole genome sequencing test for diagnosis of a rare or inherited condition.

Through the NHS Genomic Medicine Service, whole genome sequencing is now available for certain conditions where the scientific evidence shows it can help improve patient care.

Your healthcare professional will provide further information and you will be able to ask questions before you decide whether to have this test.



What is your genome?

Your genome is the information needed to build the human body and keep it healthy.

It is written in a chemical code called DNA. Your genome is made up of thousands of short sequences of DNA. These are called genes. Your genome is found inside all the cells in your body.

Many conditions are caused by changes in your genes. Some of these changes may be inherited.

What can a whole genome sequencing test tell you?

Unlike other genetic tests that only look at a few genes, whole genome sequencing looks at all your genes in one go. Even though this test looks at your whole genome, it is focused on changes in genes related to your condition; it is not looking for changes in genes that cause other conditions. The result of this test may help to:

- diagnose your condition
- suggest appropriate treatment options
- tell you if you have an increased chance of developing a condition
- tell you whether your condition could be inherited, which means other members of your family may have it, or that it could be passed on to your children. In this case other relatives may be offered testing.

Your healthcare professional will discuss this with you in more detail.



What happens in a whole genome sequencing test?

Your healthcare professional will explain the test and what will happen in more detail. Most patients will go through the following stages:

1. Referral

You will be referred to a healthcare professional who will find out more about your symptoms or condition, and take details of your family history to find out if any other relatives have a similar condition.

If whole genome sequencing is recommended, your healthcare professional will talk you through the test and what the results might show. You will have the opportunity to discuss the test and ask any questions that you or your family members might have.

If you decide not to have a whole genome sequencing test you will continue to receive the best possible care and support from healthcare professionals.

The whole genome sequencing test

The whole genome sequencing test is usually carried out on a sample of your blood, which is sent to a genomic testing laboratory to be analysed.

Testing the genome is complicated. We all carry changes in our genes that are harmless and have no impact on our health. It is often useful to compare your genes with other family members. This can help tell the difference between changes that do cause problems and those that are harmless. This means that in some situations, and often when testing children, we may need to take blood from other family members to help distinguish the harmless changes that run in families from changes that may be causing the condition.

In this case your healthcare professional will explain which family members should be tested and why.



2. Results

This is a complex test and the results will take several weeks, and may or may not find the cause of the condition. Your doctor may refer you to a Clinical Geneticist or Genetic Counsellor for further information and support.

Getting your results

Once your sample has been taken it will be analysed and the results will be sent to your healthcare professional who will discuss these with you.

The whole genome sequencing test result may:

- Show a change which explains your condition - this might affect your treatment. It may also show whether other family members are at risk of developing the condition.
- Show a change which could explain your condition, but more tests are needed before we can be sure - this might

mean testing other family members or comparing your genome changes with other patients who may have a similar condition.

- Show a change in your genes that we don't understand.
- Not find the cause of your condition.
- Show an unexpected change in your genome not related to your condition that may also affect the health of you or family members.

We are still learning about what some DNA changes mean, particularly some that are found more rarely. Your doctor may therefore discuss some changes found by the test that we don't yet fully understand but which may have implications for you and/or family members in the future.

Your healthcare professional will discuss this with you in more detail if it occurs.



Genomic data and data protection

All data is kept securely and confidentially. Your data is used in line with UK law and NHS policy. More information can be found at www.england.nhs.uk/contact-us/privacy-notice

The data from your genomic test is entered into a secure national database for the NHS Genomic Medicine Service. This system will store data about your test and the results, only staff with approved access can see your data.

Data use for insurance purposes

Insurance companies will not be given access to information about any predictive genetic test that you have had without your explicit consent.

When applying for an insurance policy, insurance companies may ask you to provide medical information about you and your family. The information that you need to share with an insurance company is regulated by a voluntary government code called the Code on Genetic Testing and Insurance: www.gov.uk/government/publications/code-on-genetic-testing-and-insurance



Data use for research purposes

Research is a vital part of healthcare. The health data donated by millions of other NHS patients has helped develop the medicines and treatments that patients receive today.

As part of the NHS Genomic Medicine Service all patients will be given the option to contribute their genomic data to a secure library so that approved researchers may access that data in a form that does not identify them. If you choose to do this then your data will be helping researchers and scientists to develop the treatments of tomorrow.

To find out more about how your genomic data can help research please see the Genomics England website www.genomicsengland.co.uk

If you choose to contribute to this research library, you may also be contacted in the future by someone in your clinical team (or occasionally by Genomics England) to give you more information if anything has been found which might be relevant to your health or that of your family, or if researchers identify a study that might be relevant to you and your condition. We would then explain the study and you would be able to say whether or not you would be prepared to take part.

Further information

You can find out more about whole genome sequencing from the following organisations:

www.nhs.uk/conditions/genetic-and-genomic-testing