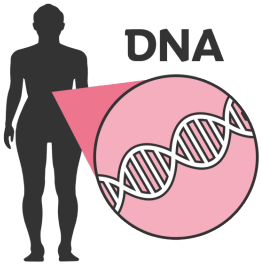
Whole genome sequencing for a rare disorder



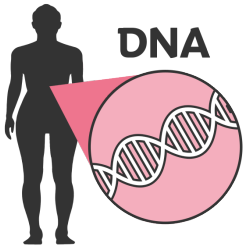
This leaflet is for patients who are offered a **whole genome sequencing** test for rare or inherited disorders.

Whole Genome Sequencing is when all the information that is needed to build and maintain your body is collected by a specialist doctor.

The NHS offers Whole Genome Sequencing for conditions where it can help make patient care better.

Your healthcare professional will give you more information before you decide whether to have the 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 code called **DNA**.

Your genome is made up of thousands of segments of DNA. These are called **genes**.

Many rare diseases are caused by changes in your genes and may be inherited.

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What can a Whole Genome Sequencing test tell you?

You may be referred for a Whole Genome Sequencing test to find out whether you have a health condition caused by a change in your genes.

Your Whole Genome Sequencing test result may help to:

- diagnose your condition
- suggest the best treatment
- tell you if you have a higher risk of getting a condition

- tell you whether members of your family may also have a higher risk of getting a condition or whether it could be passed on to your children.

If so, other family members may be offered testing. Your healthcare professional will talk about this with you.

What happens in a Whole Genome Sequencing test?

Your healthcare professional will explain the test.

Most patients will go through these stages:

Referral

You will be will passed on to another healthcare professional who will find out more about your condition.

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Consultation

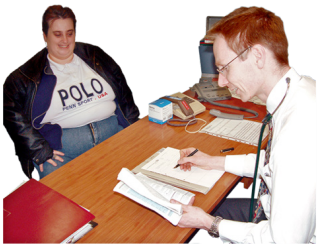
This will involve you meeting a specialist doctor.

If they recommend Whole Genome Sequencing, your healthcare professional will tell you about the test

Sample collection

If you decide to have the test, a blood sample will be taken and sent to a laboratory.

Results



You will find out what the result shows.

You can talk about the test with your healthcare professional.

You may meet a specialist who will talk to you about your results and can offer you support.

It is often useful to compare your genes with other family members. This can help tell the difference between changes that don't and do cause problems

Your healthcare professional can explain which family members should be tested.

If you decide **not** to have a Whole Genome Sequencing test, you will still receive the best health care.

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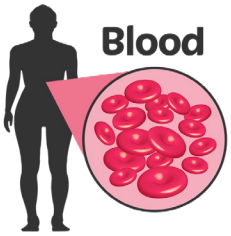

Getting your results

Your sample will be taken and checked. The results will be sent to your healthcare professional.

They will discuss with you whether your Whole Genome Sequencing test:

- Shows a change which explains your medical condition. This might affect your treatment and tell family members of their risk of developing problems.
- Shows a change which could explain your medical condition, but more tests are needed.

  This might mean testing other family members or comparing your alteration with other patients.

- Shows a change in your genes that we don't understand.

- Has not found the cause of your medical condition.
- Shows an unexpected change in your genome not related to your condition that may affect the health of you or family members.

How data about your genes is used and protected

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:
https://www.england.nhs.uk/contact-us/privacy-notice/

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Your test data and results are stored in a secure database for the NHS Genomic Medicine Service the part of the NHS that oversees Whole Genome Sequencing.

Only staff who have been approved see your data.

Data use for insurance

Data won't be shared with insurance companies without your agreeing. Insurance companies may ask you to provide medical information about you and your family.

Data use for research purposes

Health data donated by millions of other NHS patients has helped develop the treatments patients receive today.

All patients have the option to give their data to a Library so that approved researchers may use that data.

If you choose to do this then your data will help researchers to develop new treatments.

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

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Further information

Find out more about whole genome sequencing from these organisations:
http://www.nhs.uk/conditions/genetic-and-genomic-testing

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