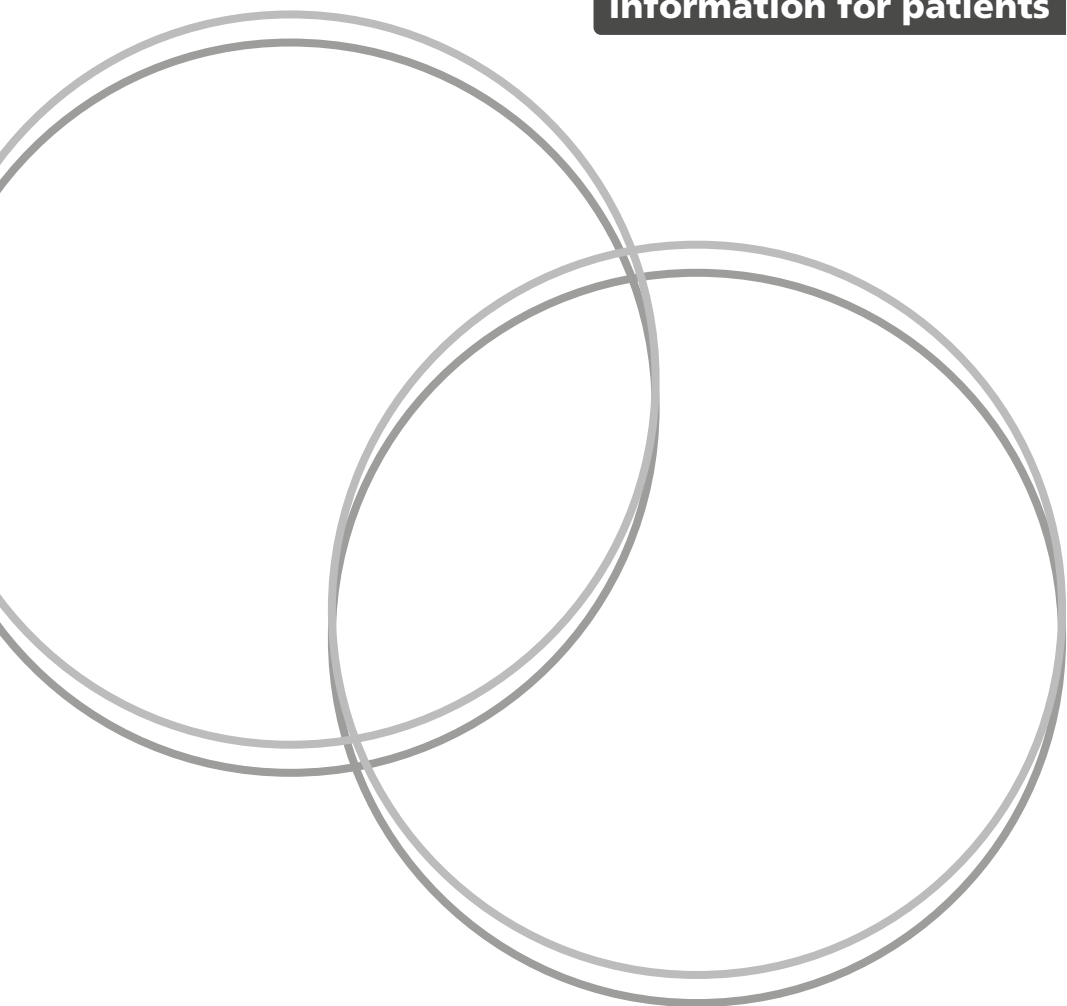


# **Rapid exome sequencing for acutely unwell adults <40 with COVID-19 with a suspected underlying genetic cause**

**Information for patients**



## Who is this leaflet for?

This leaflet is for young adult patients and their families who are being offered a test called rapid exome sequencing because they have unusually severe COVID-19 for someone of their age. It explains what the test is, how it is carried out, and the kind of information it might provide. You will have a chance to ask questions and discuss the test with a doctor or genetic specialist before deciding whether to go ahead.

It might be that the patient is too unwell to consent for him or herself. In this situation, their doctor, a family member or a friend can give consent on their behalf by taking on the role of consultee. More information about this is given at the end of this leaflet.

## Introduction

Rapid exome sequencing is a test which is used to find genetic changes which might cause health problems. Your doctor has suggested this test as having severe COVID-19 at a young age is unusual and may mean that you have a genetic change that is affecting your immune system. The result of this test may help to guide your treatment and future medical care.

## What is an 'exome'?

Our bodies are made up of cells. Inside most cells are 23 pairs of chromosomes which between them hold all of the genetic information needed to make us and for our bodies to work. This is known as our **genome**. Our genome is made of a chemical code called DNA which uses a combination of just four letters: A, C, G and T, which can be 'read' to produce our unique DNA sequence. **Genes** are the bits of the DNA code that give instructions for specific processes in our body. They make up only about 2% of our genome but have the most impact on our health. The DNA that makes up the genes is called the **exome**.

## **What is exome sequencing?**

DNA sequencing is the most detailed genetic test available. It reads the exact order of letters along the DNA. Exome sequencing reads all of the part of our DNA code that makes up our genes. Once the code has been 'read', the sequence of letters is compared to a standard code or that of close relatives to look for differences in genes that have been shown to cause health problems.

## **How is the test done?**

Your doctor or a genetic specialist will discuss the test with you and will ask you to sign a consent form if you are happy to go ahead. Testing involves taking a blood sample from you and also from both of your parents where possible. We know that this will not be possible in all cases and will be happy talk to you about other options.

DNA is purified from each of the blood samples and analysed by laboratory scientists. The scientists will look for genetic changes in you that might have increased the severity of COVID-19 and will share their findings with your doctors. If you have had genetic testing previously and DNA was stored, you may not need to give a new blood sample.

## **What are the advantages of the test?**

Exome sequencing may find a change in one of your genes which explains why you have had COVID-19 more severely than other people of a similar age. This would give your doctor more information about you and may help guide your care and treatment now and in the future. Testing may also provide important information if you are considering having children, and may help to access further testing in pregnancy.

## **Can I choose not to have the test?**

This test has been recommended by your doctor, but you can choose not to have it and you do not have to give a reason. This will not affect the rest of your treatment. If you are not sure, your specialist will be able to answer any questions and help you make the right decision for you. If you change your mind you might be able to have a genetic test at a later date.

## **What are the risks of the test?**

Some people worry about being identified from their genetic information. The chance of this happening is very small, and your data is not shared with anyone outside of the NHS.

Very rarely, a DNA sample can't be sequenced. If this happens, your doctor will explain why.

## **How do I get the results?**

Once the samples arrive in the testing laboratory, we hope the results will be available within three to four weeks. We will contact you once we have the results.

## Understanding the results

The test might find a genetic change that is affecting how well your immune system works which may have caused you to have COVID-19 more severely than usual. This will help your doctors to understand your condition better and may also be helpful for future care.

In many cases, the results of the test will be normal as far as we can tell. This does not necessarily mean there is no genetic cause, but there could be many other reasons why you have severe COVID-19 which are not genetic.

A third possibility is that the significance of the results is uncertain. As the test produces a lot of information and is relatively new, the results are sometimes difficult to interpret. This might leave you with some uncertainty over what the results mean for you and your family. As our knowledge of the causes of genetic conditions improves, we expect to be able to understand the information better. You can tell us if you would like to be contacted in the future should any new information be found that might be relevant to you.

Where one or both of your parents are also tested, results may reveal unexpected findings such as non-paternity or non-maternity. There is also a small chance that information about the risk of developing diseases in later life such as cancer and heart disease may be revealed. You can talk to your doctor or genetics specialist about unexpected findings before you have the test if you want more information.

It's possible that the results of the test may have implications for your wider family who may also carry the genetic change. If this is the case, your doctor will discuss ways of sharing your results with your family members. Sharing this information may be important for the future health of your relatives.

## **What will happen to the rest of my sample and my information?**

In line with hospital policy we will store any remaining DNA samples in the laboratory in case any further testing is needed later. We will handle your clinical information confidentially in line with the Data Protection Act and we will ask you for your consent before using your information for education and research purposes.

## **Will having the test affect my insurance?**

You don't have to tell an insurer that you are having genetic testing, or about your results. Insurance companies do not have access to your results and they have agreed not to ask about genetic testing. If the test leads to a diagnosis or any medical treatment, you should tell your insurer about these if they ask.

## **Information for nominated and personal consultees**

A personal consultee is a friend or relative of the patient who is not employed to look after the patient. A nominated consultee is someone who is a paid or professional carer. This can be someone in the patient's clinical team.

The consultee should set aside their own views and consider only the likely views and best interests of the patient. If you are taking on the role of personal consultee, you can talk to your friend or relative's doctor or a genetics specialist about this before signing the consent form.

## Contact us

### **Oxford Centre for Genomic Medicine**

Email: [orh-tr.churchill-clinicalgenetics@nhs.net](mailto:orh-tr.churchill-clinicalgenetics@nhs.net)

Tel: **01865 225931**

Website: [www.ouh.nhs.uk/clinical-genetics](http://www.ouh.nhs.uk/clinical-genetics)

## Further information

The following website offers useful information about genetic testing for patients and families:

Genetic Alliance: [www.geneticalliance.org](http://www.geneticalliance.org)

## Further information

Please speak to the department where you are being seen if you would like an interpreter. You will find their contact details on your appointment letter. Please also ask them if you would like this information leaflet in another format, such as:

- easy read
- large print
- braille
- audio
- electronically
- in another language.

We have tried to make this information meet your needs. If it does not meet your individual needs or situation, please speak to your healthcare team. They will be happy to help.

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[www.ouh.nhs.uk/information](http://www.ouh.nhs.uk/information)



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