NHS
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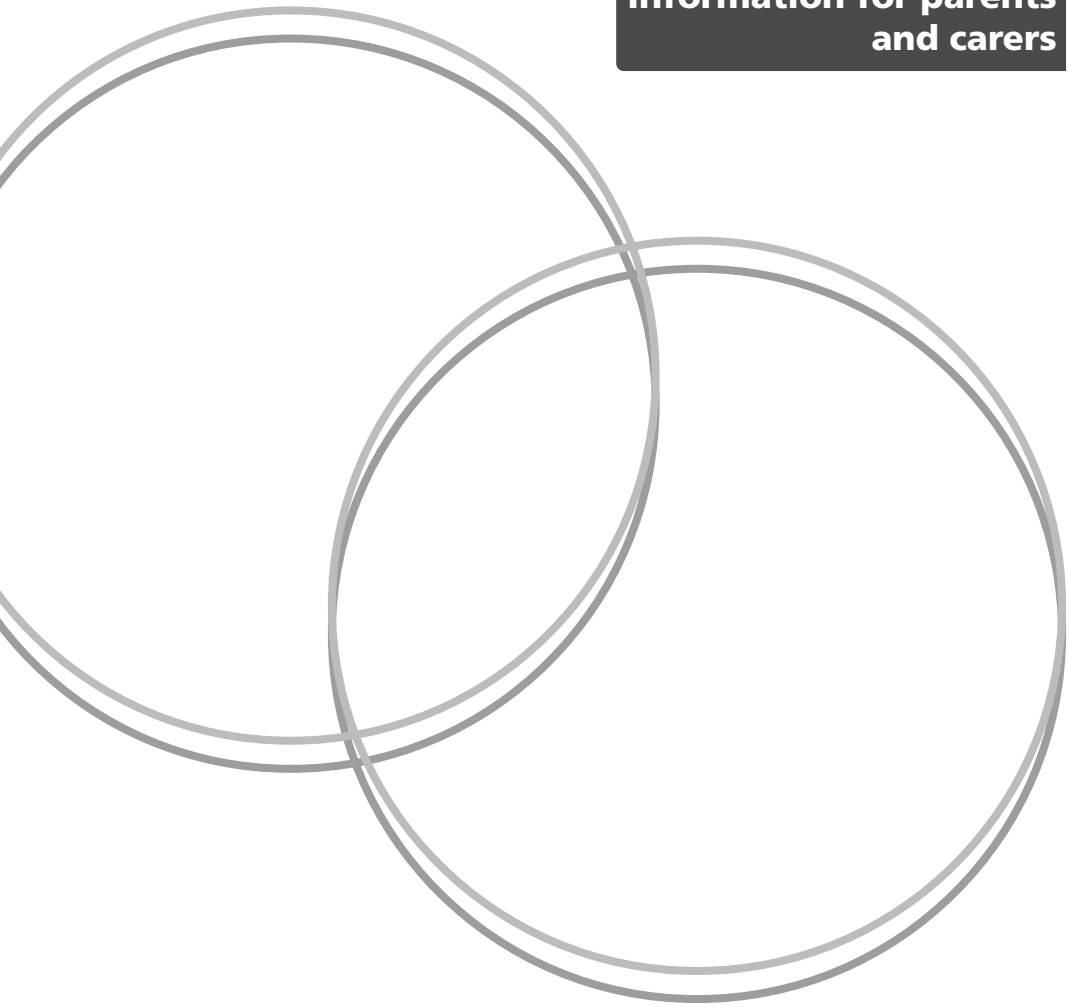
Rapid exome sequencing for acutely unwell children with COVID-19 with a suspected genetic cause

Information for parents and carers



Who is this leaflet for?

This leaflet is for the parents and carers of children who are being offered a test called rapid exome sequencing because they have unusually severe COVID-19 for someone 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.

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 your child has a genetic change that is affecting their immune system. The result of this test may help to guide their 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 your child and also from both 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 your child that might have increased the severity of COVID-19 and will share their findings with your child's doctors. If your family has 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 child's genes which explains why they have had COVID-19 more severely than others of a similar age. This would give their doctor more information and may help to guide their care and treatment now and in the future. Testing may also provide important information if you are thinking of having more children in the future for which further testing options may be offered.

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.

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 clinical team 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 child's immune system works which may have caused them to have COVID-19 more severely than usual. This will help their doctors to understand their 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 your child has 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 your child and you as a 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 your family.

If 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.

Contact us

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Tel: **01865 225931**

Website: **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**

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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