NHS
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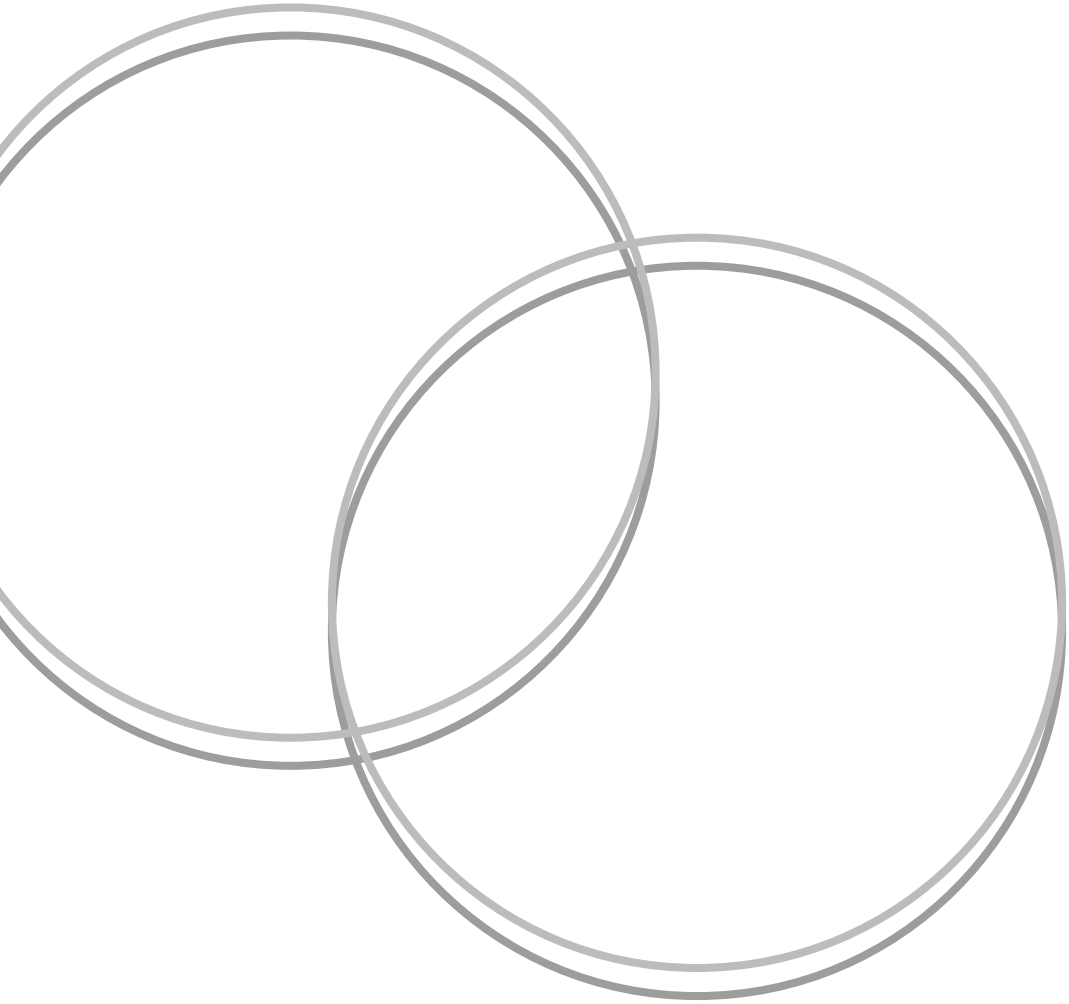
Rapid exome sequencing for acutely unwell children with a likely genetic condition

Information for parents and carers



Who is this leaflet for?

This leaflet is for the parents and carers of very unwell babies and children who are being offered a genetic test called exome sequencing. 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 other 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 your child has a condition with a combination of features which suggest that it may have a genetic cause. The result of this test may guide your child's 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 you and will ask you to sign a consent form if you agree 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 look for genetic changes in your child that might be causing their condition and share their findings with your 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 not typically detected through standard genetic tests. This would give your child's doctor more information about the cause of your child's condition and may help guide care and treatment. It might give you more information about the long-term outcome for your child. Testing may also provide important information for future pregnancies 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 child's treatment. If you are not sure, your specialist will be able to answer any questions you have and help you reach the right decision for you and your family. If you change your mind you could have the 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 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

We hope that the test will find the genetic change that is causing the condition in your child and that this will help your doctors to understand their condition better. This happens in about 40% of cases.

In other cases the results are normal as far as we can tell, but this does not mean the cause of your child's condition is not genetic.

A third possibility is that the significance of the results for your family 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 family. As our understanding 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.

Where the child and both parents are tested, results may reveal unexpected findings such as non-paternity or non-maternity. There is also a chance that information about the risk of developing diseases in adult 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 websites offer useful information for patients and families living with a genetic condition:

Unique: **www.rarechromo.org**

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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