

Your child's exome sequencing test

Information for patients, parents and guardians

We have written this factsheet as a guide to your child's exome sequencing test. Your child's doctor has recommended this test because it is possible that your child may have an underlying genetic disorder. The result of this test could help us to better understand your child's condition and decide whether we need to make changes to their current and/or future medical care.

What is exome sequencing?

Exome sequencing is a relatively new type of blood test. We use it to analyse large amounts of the body's genetic code at one time. The results of the test may show whether your child has a genetic disorder.

Our genetic code is embedded in our 'DNA', which can be taken from blood and other tissues. We can then read through the genetic code and check for changes that could suggest a genetic disorder. This process is known as 'sequencing'.

In 'exome' sequencing we are only looking at the parts of the genetic code known as 'exons'. Our genetic instructions (or 'genes'), have a complicated structure but the exons are essential because they contain the genetic code that is used to build important products known as 'proteins'.

What happens in the exome sequencing test?

For the exome sequencing test, we will take a sample of your child's blood (like in a normal blood test). If possible, we will also take blood samples from both parents, as this will help us to get the most information from the results. If only one parent is able to give a blood sample, this will still be helpful. Even if neither parent is able to give a blood sample, we can still perform the test.

We will send the blood samples to the laboratory for testing.

Before you agree to the test

Possibilities to consider:

It is important that you understand and consider the following points before agreeing for your child to have the test:

- It is possible that no diagnosis will be made. However, this would **not** mean that an underlying genetic disorder has been ruled out.
- The result of the test may be difficult to interpret. We may recommend further tests and/or assessments to try make a better interpretation.
- In rare cases, we may identify genetic findings that are important but completely unrelated to your child's current health problems. These are often referred to

as 'incidental findings' and could have implications for your child as well as other family members. We will not be actively looking for such findings and will only report them if the referring doctor and other specialists consider this appropriate.

- Because we will be sequencing the parents' samples as well as the child's, it is possible that genetic findings could have implications not only for the child but also for a parent and possibly wider family members as well.
- Exome sequencing of a 'family trio' detects biological relationships and can reveal whether a parent is biologically related to their child.

Please let your child's doctor know if:

- your child was conceived using a donor egg or donor sperm.
- you or your child have ever received a bone marrow transplant or peripheral blood stem cell transplant.

Results

You should receive your child's test results about four weeks after the test.

What happens to the samples, data and results?

Surplus DNA samples are stored in the Wessex Regional Genetics Laboratory (Salisbury). This is because they may be needed for further genetic investigations or as control samples in the future. The sequence data from the exome sequencing test is stored by the Exeter laboratory, where the test is performed. The test results, in the form of a laboratory report, are part of your child's medical record and are shared with relevant healthcare professionals.

Contact us

If you have any questions or worries, please contact your child's doctor.

If they are unable to help, you can seek advice from the Wessex Clinical Genetics Service:

Wessex Clinical Genetics Service
Mailpoint 627
Princess Anne Hospital
Coxford Road
Southampton
SO16 5YA

Telephone: **023 8120 6170**

If you need a translation of this document, an interpreter or a version in large print, Braille or on audiotape, please telephone **023 8120 4688** for help.

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