What does the test involve?

A blood sample is needed from your child and also from both biological parents (where possible). Whole genome sequencing will be performed on DNA from the blood samples provided.

Why do you need samples from parents?

Most of our genetic information is inherited from our biological parents. It is helpful to be able to compare the information from your child's sample with that of both parents. If this is not possible, we will still be able to perform rapid whole genome sequencing, although it may be more difficult for us to make a genetic diagnosis.

Who will give me the results?

The doctor who arranged the test will receive the results and will arrange to discuss them with you. You might also have a meeting with a genetic specialist to explain the results in more detail.

Will our data be stored somewhere?

See: https://www.england.nhs.uk/contact-us/privacy-notice/nhs- genomic-medicine-service/

For more information on genetic testing

https://www.nhs.uk/conditions/genetic-and-genomic-testing/

If you have any other questions, please speak to your child's clinician.

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NHS England

Rapid Whole Genome Sequencing (R14)

Information for parents and families

Version V1 November 2023

Why has my child been offered this test?

There may be a genetic condition affecting your child's health. A genetic diagnosis may be important for understanding more about your child's condition. It may help the clinicians to know how to treat your child. It may also give information about the risk of other family members or future children developing the condition.

What is Rapid Whole Genome Sequencing?

Our 'genome' holds all of the genetic information that our body needs to build it, and to keep it healthy. It includes over 20,000 genes which are made up of DNA – a form of chemical code.

We all have millions of gene changes or 'variants' in our DNA code. Most of these are just part of the common genetic variation that makes us all individual. However, some changes can cause some changes affecting health and development.

Whole Genome Sequencing (WGS) looks for changes or 'variants' in and around the genes. The test produces a huge list of these. Computers are used to filter this list down to those most likely to be causing your child's health problems. Genetic Scientists then review these to try to pinpoint a genetic diagnosis for your child.

If you have any other questions, please speak to your child's clinician.

What will the results of the test tell me?

There are 3 possible outcomes;

1. **A genetic diagnosis is found which explains your child's health problems.**

2. **No genetic cause for your child's health problems is found.** This does not rule out a genetic condition because not all genetic conditions are yet known about or detectable, even with this detailed testing.

3. **The test shows a variant "of uncertain significance" where we are not certain if it could cause a genetic condition or not.** Sometimes it can be difficult to decide if something is just part of normal variation or if it is something that could cause health problems. In this situation we can sometimes do extra tests to help decide. Your child's clinician would discuss this with you.

Will the test show anything else not related to my child's health problems?

- The test looks for the changes which will help us understand your child's current health problems. However, very occasionally an unrelated gene change or variant might be seen. If knowing this could make a difference to the future health of your child, yourself (as a parent) or other family members, then this will be explained to you.

- The test will show if a parent is not the child's biological parent. If this is a concern, please raise this with your clinician in advance.