Trio Whole Exome Sequencing

Information for families

What are genes?

Genes are stretches of DNA (genetic material) that direct how we grow and develop during pregnancy and control all aspects of how our bodies work after we are born.

Our bodies are made up of millions of cells which contain two copies of the human genome (a set of approximately 21,000 different genes). We inherit one copy of the genome from our mother and one copy from our father. Sometimes, one or both copies of a gene contain spelling mistakes which alter the function of the gene. Such changes can cause genetic disorders that significantly affect the development of around one in every 200 babies.

What is the exome?

The exome is the DNA that codes for proteins

Not all DNA found in the body acts as a set of instructions. Between genes, there are stretches of DNA whose function is not yet fully understood. Within a gene, there are also stretches of DNA that are not directly used. These stretches of DNA are called introns.

The sections of genes that act as instructions are called exons. Exons tell the body how to build proteins. Proteins act as building blocks for the human body and make up cells, tissues, and organs. The exome makes up about 1% of our DNA but is thought to contain approximately 85% of disease causing gene changes.

What is trio whole exome sequencing?

Trio whole exome sequencing involves looking at all the DNA that codes for proteins (the exome). Most commonly an affected individual's exome is compared to the exomes of their biological parents. This allows us to find new changes that have occurred in the affected individual, or inherited gene changes that can cause the genetic disorder.

With advances in genetic testing technology, we can now look at many different genes at the same time. Previously, we were only able to look at one gene at a time, which could make finding a genetic answer a very long and time consuming process. In a trio whole exome, we typically look at the exons of over 2000 genes that are known to be associated with developmental disorders.

Why is my family being offered this test?

This test is currently our best way of trying to find a genetic answer for a child's developmental difficulties or congenital problems.

Unfortunately, we do not always find an answer. Occasionally, we find a gene change and are uncertain whether it is a normal gene change seen in the general, healthy population or a disease causing change associated with a genetic condition.

We currently find a genetic answer from approximately 1 in 3 trio whole exome tests that are undertaken. Finding out the cause of a condition can be important for many families. Understanding the genetic basis of a condition can help health professionals give you information about the condition and its management. Sometimes, it can help with thinking about whether other family members could be at risk of the condition or the chance of its recurrence in a future pregnancy.

How is the test done and how long do the results take?

The test is done via a blood sample or saliva sample and the results usually take four to five months to come back.

The DNA is extracted from either a blood or saliva sample. We can arrange to take a blood sample in a clinic, or a saliva kit can be posted to your home address. The results from the trio whole exome usually take four to five months to come back, due to the amount of analysis involved.

Further information

Further information on whole exome sequencing can be found at https://rarechromo.org/media/information/Other%20Topics/DNA%20sequencing%20(whole%20genome%20and%20exome)%20FTNP.pdf

Further information on DNA, genes and genomes can be found at www.yourgenome.org

Local contacts

Your local genetics service:

South East of Scotland Clinical Genetic Services
Molecular Medicine Centre (MMC)
Western General Hospital
Crewe Road South
Edinburgh
EH4 2XU

Telephone: 0131 537 1116

Clinical Genetic Service leaflet reference: 084

v2.0 approved by NHS Lothian Patient Information Team, Feb 25, Review date: Feb 28