Exome Sequencing

What Does It Do?
Exome sequencing (ES) is a broad genetic test which is used to figure out if your child's symptoms may have an underlying genetic cause.

What Is Genetic Material?
The human body is made up of billions of building blocks called cells. Inside each cell are thousands of genes, made of DNA, which tell the body how to grow and develop. It is estimated that humans have 20,000 genes. All of our genes grouped together make up our genome.

We get half of our genes from our mother and the other half from our father. We then pass half of our genes to each of our children. Sometimes there are new changes in a gene that were not inherited, or passed down, from a parent.

What Are Exons?
Each of our genes is made up of two types of sections - introns and exons. Exons are the coding sections of the DNA, while introns are edited out. Exons make up approximately 1-2% of our entire genome, but changes in exons cause up to 80-90% of genetic conditions.

What Can This Test Tell Me?
ES can identify small changes (missing, extra, or misspelled instructions) in your child's gene(s) that may explain their symptoms. In some cases, the results may change your child's medical care.

The results of ES may also show that other family members may be at-risk of having or passing along a genetic condition.

Limitations
ES is not 100% sensitive and cannot identify every change in a person's DNA. The thoroughness of the results depends on having accurate medical and family histories. The results are based on our current understanding of genetics.

Types of Results

Negative
No changes were found. This does not rule out all genetic conditions.

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Uncertain Variant
A change in the instructions of one or more genes was found, but it is unclear if it is causing your child's symptoms.

Positive
A change in the instructions of one or more genes was found that likely explains your child's symptoms.

How Is The Test Done?
ES is usually performed on a sample of the patient's blood sample, about 1 teaspoon. Sometimes the parents' blood will also be collected.

Parental Testing
Samples from the mother and father (parental samples) may or may not be collected at the same time as the child's sample. In some cases, parental samples may be needed after the results are back to help understand a genetic change found in the child.

Unique Results
Some of these tests can identify if the biological parents of a child are related to each other by blood or whether or not the parents are the child's biological parents.

When Will I Get The Results?
The test takes at least 3 months to be completed. We will call you when your results are available, and will either schedule an appointment for genetic counseling or discuss the results at your child's next appointment.

Reanalysis
In the case of a negative or uncertain result, your doctor may recommend rechecking your child's ES data after some time has passed.

Secondary Findings
Your family can choose if you would like the report to include certain genes chosen by the American College of Medical Genetics and Genomics (ACMG). These genes cause health conditions in adulthood and may change you or your child's medical care.

Data Sharing
Data from ES can be submitted anonymously to different databases in order to help other individuals with similar genetic changes or symptoms. You can choose to agree to data sharing or to not allow your child's data to be anonymously shared.

Thank you for allowing us to be a part of your child's care. If you have any questions or concerns, please call our office at (323)361-2178.

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