Genetic Testing Overview

What Is Genetic Testing?
Genetic testing searches your child's gene(s) for any alteration (mutation) in the genetic material. Mutations in genetic material can cause symptoms related to a genetic condition.

Why Is Genetic Testing Recommended?
Genetic testing can be used to find out if your child's symptoms are being caused by an underlying genetic condition. If a genetic condition is identified, it may give your child's healthcare providers additional information about their current and future medical and developmental needs. Additionally, a genetic diagnosis can also help your family know if your other children (planned or present) could also have the condition.

Cell → Chromosome → DNA → Gene → Mutation

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, that tell the body how to grow and develop. It is estimated that humans have 20,000 genes.

We have 2 copies of all 20,000 genes. One copy comes from our mother and the other copy comes from our father. We then pass one copy of each gene to each of our children. Sometimes there are new changes in a gene that were not inherited from a parent.

Types of Genetic Testing
There are several types of genetic testing. The main differences between them are the number of genes that are reviewed and the type of differences that the test can find.

Chromosome Analysis
This test looks at the way the DNA is organized into chromosomes. It can find differences in the number of chromosomes and how they are organized. It does not look at the DNA instructions.

Chromosomal Microarray
This test looks for missing and extra pieces of genetic material. It does not look at the DNA instructions.

Single Gene Sequencing
In this test, the laboratory looks at the DNA instructions of one gene to find any differences in the instructions.

Multi-Gene Panels
In this type of test, a combination of 2 or more genes are reviewed for differences in the instructions. This test is sometimes referred to as a "focused exome".

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Approved by PFE 10/03/24

Exome Sequencing

In this test, the active sections of all 20,000 genes are reviewed for differences in the instructions.

Genome Sequencing

In this test, the entire sequence of all 20,000 genes will be reviewed for differences in the instructions.

***Your child's provider will decide which tests are appropriate and in what order they will be considered. Some of the above testing is not appropriate for all patients.

Types of Results

Normal/Negative

No changes were found in the genetic material that was reviewed. This does not rule out all genetic conditions.

Uncertain Variant

A change in the genetic material was found, but it is unclear if it is causing your child's symptoms.

Positive

A change in the genetic material was found that likely explains your child's symptoms.

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.

Parental Testing

Parental samples are not typically collected at the same time as your child's sample. In some cases, parental samples may be requested after the results are back to help understand the genetic change in your child.

How Long Will It Take To Get Results?

The time for the test to be completed varies by both the test and the genetic laboratory where it is performed. Please ask your provider for information about your child's specific test.

Your Child's Testing

At today's visit: _____ was recommended for your child.

This type of testing is called: _____.

We will submit the request for this testing to your insurance to find out if they will allow the test to be done.

Once the test is approved and the samples are collected, the test should be completed in _____ weeks/months.

We would like to see your child again in _____ weeks/months/years.

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.