Genetic Testing Basics

Understanding and managing your child's medical testing can be challenging. This fact sheet covers basic genetic testing terms and tips as you get started.

Genetic Testing Terms

There are many different types of genetic testing, each with a different diagnostic purpose. To learn more, visit our Fact Sheets for additional information on Genetic Testing. The terms below are common to most types of genetic testing.

Geneticist is a doctor that specializes in the study of genetics. The geneticist will work together with the genetics counselor as a team to help your family through the genetic testing process. The geneticist usually orders specific genetic tests and interprets the results to come up with a diagnosis. They will likely also manage any medications or treatments plans related to your child's genetic diagnosis, refer your child to other specialists if necessary, and follow-up with your family as needed.

Genetics Counselor is a healthcare provider with specialized training in both genetics and counseling. The genetics counselor will work together with the geneticist as a team to help your family through the genetic testing process. The genetics counselor will usually do pre-test counseling to discuss: your family history, the types of testing available, insurance and financial concerns, the benefits of testing, the risks and limitations of testing, and the possible results. They may also meet with you for post-test counseling to discuss: your child's diagnosis, your child's treatment plan, and what to expect with follow-up appointments. The genetics counselor will likely include your child in the conversations. They are trained to help explain these complex things to your family, even if you have no previous medical knowledge or experience.

Prenatal Testing is a form of genetic testing that is done during pregnancy. One of the most common types of prenatal genetic testing is called *amniocentesis*, which involves taking a small amount of your baby's amniotic fluid for testing.

Karyotype Testing is a form of genetic testing that looks at the number, size, and shape of a person's chromosomes in order to look for genetic changes. This type of testing can either be a prenatal test (*amniocentesis*) or can be completed any time after birth.

Chromosomal Microarray Testing is a form of genetic testing that looks very closely at a person's chromosomes in order to find missing or extra pieces of genetic material.

Gene Panel Testing is a form of genetic testing that looks at a specific set of genes that cause a known condition or illness. For example, this type of testing may test the set of known genes that may cause seizures or a specific type of cancer.

Whole Exome Sequencing is a form of genetic testing that is one of the most in-depth types of testing available to families. This test looks at all of the genes in a person's DNA.

Genetic Mutation is a change in a person's genetic material.

Spontaneous Mutation is an unexpected genetic mutation with no known cause. These may also be called "sporadic mutations" or "new mutations".

Hereditary Mutation is a genetic mutation that has been inherited, or passed on, from one or both parents. These mutations may cause illness or disease that runs in a family. These may also be called "inherited mutations".

For more information about genetics testing, contact Indiana Family to Family at 1-844-323-4636 or visit inf2f.org.

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Tips for getting started with Genetic Testing

Reasons to consider genetic testing for your child: There are many reasons for you and/or your medical provider to consider genetic testing for your child. These may include:

- A known illness or disease that runs in your family
- Multiple congenital (birth) defects or differences
- Developmental delays, including Autism Spectrum Disorder
- Developmental regression
- Facial differences
- Low muscle tone
- Seizures
- Unexplained symptoms or medical conditions

How genetics tests are done: Talk with your doctor about how your child's genetics testing will be done. A few types of genetic tests are done through a medical procedure, such as: fluid collection, bone marrow collection, or a blood draw; however, most genetic tests are a simple cheek swab that will likely not be traumatic or painful for children.

Finding answers and getting a diagnosis based on genetic testing: Some families will get answers and receive a specific diagnosis for their child based on the results of their genetic testing; however, most families will get uncertain results, benign (not harmful) results, or no diagnosis. Families should prepare for the possibility that they may not get a diagnosis for their child through the genetic testing process. This can be frustrating, and you can talk with your geneticist or genetics counselor about how you are feeling.

Communicating with your provider and advocating for your child during the genetic testing process: Your child's geneticist and genetics counselor are a team that should help you through your child's genetics testing process. Families should not be afraid to ask questions, ask for clarification, or ask for additional resources and information from their genetics providers. Many families have a wide range of emotions during the genetics testing process, including feelings of frustration and grief. Genetics counselors are trained to not only help families through the genetic testing process, but also through the feelings that come along with the process.

Where to find more information: For more information about genetics testing, see our other genetic testing fact sheets or the following resources:

- Prenatal Genetic Counseling (AAP, 2013)
- Congenital Abnormalities (AAP, 2015)
- Detecting Genetic Abnormalities (AAP, 2019)
- Genetic Testing 101: What Parents Need to Know (CHOP, 2019)
- The Journey Through Diagnosis (Midwest Genetics Network, 2022)

If you have additional questions about the genetic testing process or would like help finding more information, contact Indiana Family to Family at 1-844-323-4636 or inf2f.org

Programs and systems change often. It is important to ensure that you are using the most current information. This fact sheet was updated July 2022. Please check https://www.inf2f.org/fact-sheets.html for themost recent edition.
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