What is genetic testing?

Genetic testing looks for changes in the structure of an individual's genes. Genes are the DNA instructions that are inherited, or “passed down,” from parents. Each person has one copy of genes inherited from his or her mother and another copy inherited from his or her father. Genes are located on the chromosomes, which are like libraries, in the cells in our bodies. Genes tell the body how to make the proteins to maintain good health.

Why is genetic testing done?

Genetic testing is often used to identify the cause of a health disorder or problem. In some instances, information from genetic testing can be used to determine the best treatment for a specific health disorder. For some families known to have a health problem that is an inherited condition, genetic testing is recommended to determine if an individual has an increased risk of developing this condition.

How is genetic testing done?

Your child’s doctor may recommend performing genetic testing. Testing is usually performed using a blood sample, but, sometimes, saliva, urine, amniotic fluid, or other tissues are used. The sample is then sent to the genetics laboratory.

At the genetics laboratory, the testing involves looking at the pattern of DNA in the individual’s sample. The major goal is to determine if a difference or change is present in the DNA sample that can affect health. Genetic testing includes several different methods. One method is to analyze one specific gene. Another method, called microarray testing, looks for missing or extra portions of DNA. Another method is whole exome sequencing, which looks at the pattern of most genes. Lastly, in some instances, the chromosomes are examined, which is called a karyotype. Your doctor will decide which method is best for your child. This process may include a consultation with a specialized genetics physician (a health care professional who is specially trained in genetics). For most genetic tests, pre-authorization from your insurance company will be necessary.

When testing is complete, the results are sent to your child’s doctor, who can share the results with you. A genetics counselor or genetics physician may be asked to help explain the results and provide recommendations for medical management and prevention.

Who has access to your child’s genetic test results?

Similar to other medical test results, genetic test results are usually part of your child’s medical record. However, genetic test results are a type of health information that is covered by the privacy rules of the Health Information Portability and Accountability Act of 1996, or HIPAA. Another legal protection is the Genetic Information Nondiscrimination Act of 2008, or GINA, which makes it illegal for a health insurance company or employer to discriminate against someone because of his or her genetic information.

What are the limitations of genetic testing?

Genetic testing cannot provide information on all inherited diseases. All of us have small differences, known as genetic variations, in our genes. Some gene differences cause disease, and some do not seem to affect our health. Unfortunately, in some cases, genetic disorders can be identified for which no treatment currently exists.

Although there are many advantages to genetic testing, there are some ethical issues related to the risk of finding unexpected results. In some instances, family members may become depressed or anxious when they learn of the results of their genetic testing. Some insurance companies refuse to cover the costs of genetic testing. Parents should discuss the benefits, costs, and potential risks of genetic testing with their child’s doctor.