While genetic testing can be beneficial, there may be emotional, social, or financial consequences as well. Genetic counseling can be helpful in walking families through the process. Also, genetic testing is not a perfect science and it does not always result in a diagnosis and sometimes, even with a diagnosis, there may be no current treatment options.

Genetic tests analyze the building blocks (nucleotides) of our genetic code (DNA). DNA is found in each of our cells and make up the genes that tell the cells what proteins to make, which then create structures in our body, such as brain, muscles, heart, and skin. DNA also helps create enzymes that are responsible for chemical reactions and biological functions.

Genetic variants may cause or contribute to disease, but also can be silent or benign. Genetic variants can be inherited from a parent, but also can be new and not inherited. Some common ways to test for genetic variants are:

- **Chromosomal Microarray Analysis (CMA):** Looks for missing (deletions) or extra (duplications) pieces of DNA.
- **DNA Sequencing:** These tests sequence the nucleotides that make up the genetic code to identify any changes compared to what is expected. Common sequencing tests include:
  - Gene Panels: sequence a select number of genes associated with a condition or of group of conditions, such as epilepsy.
  - Whole Exome Sequencing (WES): sequences all 21,000+ human genes, which is only about 2% of our overall DNA.
  - Whole Genome Sequencing (WGS): sequences all of our DNA, which is over 3 billion nucleotides, and includes all the data of WES and CMA, the remaining 98% of our DNA, including mitochondrial DNA. This test also has the ability to assess for DNA repeats of different sizes that might cause disease, such as myotonic dystrophy.

What are the benefits of genetic testing?

- Genetic testing can help determine if there is a genetic cause or risk factor for a neurologic disorder or condition.
- Genetic testing results can provide diagnostic information that eliminates the need for other invasive, unnecessary or expensive diagnostic tests, ultimately shortening the time to diagnosis.
- Both positive and negative results can provide information for treatments that target the causes of disease rather than just suppressing symptoms, change clinical management and improve outcomes.
- If a genetic diagnosis is found, families can connect with other families and advocacy groups specific to their condition.
- There may be increased opportunities to take part in clinical studies based on genetic diagnoses.
- Genetic information can identify risks for other health concerns and provide information about the chance of additional family members inheriting the condition to assist with family planning.

What are the types of genetic tests for child neurology?

Seek consultation from a medical specialist about choosing the appropriate genetic test. Most sequencing tests require only a saliva sample or cheek swab, but these tests can also be run on blood samples.
**What is the cost of genetic testing and how is it covered?**

As genetic testing technologies have become more sophisticated and accessible, costs have dropped dramatically. Depending on the type of genetic testing, self-pay costs can range from under $100 up to $3,000 per person. Often to get useful results, additional family members must be tested, increasing the total cost for genetic testing. Nevertheless, it is expected that costs will continue to drop further as technologies advance and competition grows.

Initially, many insurers and other payers were reluctant to cover the costs of advanced genetic testing, but as research and medical knowledge has proven the importance and cost effectiveness of such testing, it is increasingly common to obtain genetic testing as a covered service. Most patients are able to get genetic testing at a partially or fully covered rate. If not, labs often have policies that will help lower the cost to a patient and their family. There are also genetic testing companies that provide specific tests at no charge. Additionally, various research studies may include genetic testing at no charge.

**What role does genetic counseling play?**

This important counseling can help to:

- Advise families on the benefits and limitations of genetic testing.
- Order appropriate genetic tests.
- Help a person and their family understand the results and the implications for clinical management.
- Find advocacy groups, clinical research opportunities, and resources.

**Learn More**

Visit childneurologyfoundation.org/GeneticTesting

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