Top 5 Things to Know About Genetic Testing

REN Rare Epilepsy Network
rareepilepsynetwork.org

1. What is genetic testing?

Genetic tests analyze DNA, which contains the instructions for how a person's body develops and functions. Changes in DNA, called DNA variants, may impact growth, development or overall health. Genetic tests may provide valuable insights into a person's health as well as the health of family members.

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2. When should someone with epilepsy get genetic testing?

The National Society of Genetic Counselors (NSGC) and the American Epilepsy Society (AES) recommend exome, genome or multi-gene panel as first-line tests for anyone with unexplained epilepsy. Other professional organizations also recommend exome or genome for individual with other neurodevelopmental conditions.

Genetic testing may provide both clinical and personal benefits to people with epilepsy and their families including:

- Changes in a person's epilepsy treatment and management
- Better understanding of what to expect in the future
- Valuable information about inheritance to family members
- Emotional relief to families who have been searching for answers
- Specific resources for education and peer support
- Serve as an entry point into clinical trials or other important research studies

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3. Who orders testing? Where can I find genetic testing?

Genetic tests should be selected, ordered and interpreted by qualified healthcare providers who have expertise in genetics to ensure the appropriate tests are selected and the results are accurately interpreted. Involving a genetic counselor in this process can be crucial in providing guidance and support for families throughout their genetic testing journey.

Frequently, but not always, level 4 Epilepsy Centers (centers with the most experience in epilepsy care, tools and interventions) have access to and offer genetic testing.

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4. What kind of genetic testing is available?

There are many different genetic tests currently available. The specific test that your healthcare provider recommends may vary depending on your medical and family history. Although genome sequencing, exome sequencing and multi-gene panel tests are the recommended first line tests for people with epilepsy, there may be additional tests required in some situations.

Genome sequencing: The most comprehensive test that looks at a person's entire genomic sequence.

Exome sequencing: Focuses on sequencing the protein coding regions of a person's genes, called exons, which are where approximately 85% of disease-causing variants are found.

Multi-gene panel: Analyzes a subset of genes known to cause epilepsy. Typically, panels include 25 or more epilepsy-related genes. It is important to note that the genes included in a panel may vary among different laboratories.

These and other tests are described in more detail on the REN website [REN link].

5. What Does My Genetic Report Mean? What Kind of Results Can I Get from Genetic Testing?

The results that a person can get from a genetic test are classified into 3 categories below. For more in-depth information visit, [REN link].

Positive: A genetic change has been identified that explains the person's symptoms or is associated with a specific condition. The variant(s) identified may be classified by the laboratory as either pathogenic or likely pathogenic.

Negative: No disease-causing or potentially disease-causing changes associated with the person's symptoms have been identified in the analyzed genes. A negative result does not rule out the possibility of a genetic diagnosis.

Variant of Uncertain Significance (VUS): A genetic change has been identified, but there is not enough evidence to determine if the variant is potentially disease-causing or benign .

Learn more at rareepilepsynetwork.org/genetictesting | rareepilepsynetwork.org | info@rareepilepsynetwork.org

This GUIDE was developed by the Rare Epilepsy Network (REN) in collaboration with the University of Pennsylvania Orphan Disease Center JUMPStar program and Masters of Science in Genetic Counseling Program, as well as Beth Rosen Sheidley MS, CGC and Gillian Prinzing, MS, CGC of Boston Children's Hospital.