Pairnomix

Genetic testing for people with epilepsy

Our DNA encodes thousands of genes, each of which plays an important role in how our bodies function.

Diseases can arise when a change in the DNA sequence that encodes a gene causes improper functioning. These are known as mutations, and they can occur in up to 80% of epilepsy cases.*

Knowing if there is a genetic explanation for your condition and which gene, if any, is mutated, can help your physician decide the best course of treatment for you — which is why he or she may ask you to undergo a genetic sequencing test. Using a small sample of your blood or saliva, these tests are able to read (or "sequence") your DNA and compare it to a healthy reference. If any changes in your DNA are identified, the specific change and the implicated gene will be reported back to your physician.

Described below are five common types of genetic tests that your doctor may order:

Test Type	Test Description
Single Gene	Only one gene is sequenced because a physician is reasonably sure a malfunction of that particular gene is causing their patient's condition.
Focused Panel	Anywhere from three to several thousand genes are sequenced because of known previous implications in seizures or epilepsy.
Whole Exome	The protein-encoding parts of all genes are sequenced.
Whole Genome	Almost all of the genome is sequenced, including regions that do not encode proteins.
Pharmacogenetic Panel	Anywhere from three to several thousand genes are sequenced to determine if you have mutations in genes that, when they are functioning properly, help you metabolize and receive the benefits of drugs used to treat epilepsy symptoms.

Whether your test results identify a responsible mutation or a variant of unknown significance (VUS, which means its contribution to disease is unclear) you may be wondering what caused your DNA to change. Most of the time, these changes are random events and we don't know why they occur. However, sometimes the change might have been inherited from your biological mother or father. If this is suspected, your doctor may also ask that you undergo what's known as "trio testing," in which your DNA and your parents' DNA are sequenced and compared.

Pairnomix empowers people living with rare disease by performing personalized genetic research that enables physicians to make better-informed healthcare decisions. We're here to answer your questions about genetic research.

Contact us to learn more.

Pairnomix helps people with epilepsy who have already received genetic sequencing and have successfully identified the gene mutation that is known or believed to cause their condition. We do not perform genetic sequencing tests, but we encourage you to speak with your physician about your options.

*Myers CT, Mefford HC. Advancing epilepsy genetics in the genomic area. *Genome Medicine*. 2015; 7-91.

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