What is Genetic Testing?

Genetic testing is a kind of medical test that looks for changes in your DNA. Your DNA is the blueprint that your body uses to function properly. Certain changes in the DNA can cause this blueprint to be disrupted and can cause genetic disorders.

Why is Genetic Testing for the Epilepsies Important?

It is believed that the majority of epilepsy not associated with trauma, infection, or stroke, is caused by a genetic change. Finding a genetic cause for your epilepsy can result in a genetic diagnosis. Having a genetic diagnosis can:

**Impact Treatment**
Your doctor can recommend specific medications or treatments if they are available for your genetic condition.

**Predict longterm outcomes**
If you are diagnosed with a genetic condition, your doctor may be able to predict any medical concerns you may have and prepare for them.

**Inform risk to family members**
Because your genetic information is hereditary, having a diagnosis can help determine if any family members are at risk of having the same disorder and can help them get testing as well.

**Make it easier to obtain access to resources**
It can be easier to access school/government resources with a genetic diagnosis.

**Connect with patient advocacy groups, as well as community and support groups for your genetic diagnosis.**

**The Rare Epilepsy Network has a Members and Partners* page where you can access resources for specific diagnoses. These organizations offer education, information, research, support and more.**

Where Can I Go to Get Genetic Testing?

It is recommended that this genetic testing be ordered by your neurologist. If possible, find a neurogenetics clinic near you as they will have the best training to order and interpret your genetic testing.

Follow this link* to view a map of all the genetics clinics in the country. You can select neurogenetics to find a clinic close to you. Level 3 or 4 Epilepsy centers may also offer genetic testing. You can search for a center by zip code here*.
What Kind of Genetic Testing Is Available?

Genetic testing is recommended for all unexplained epilepsies*. There are two primary types of tests that your doctor may order. These test for the most common kind of genetic change that causes epilepsy:

**Gene panel**
Analysis of genes that are known to be most commonly associated with epilepsy. There are several different genetic testing companies that offer epilepsy panels and they may have slightly different genes on them. Your doctor/genetic counselor will order the most appropriate test depending on your presentation.

**Whole exome sequencing**
Analysis of the whole DNA code and looks for genes that may explain your epilepsy. Your doctor might order an exome if you had previous testing that did not provide an answer, or if there are other symptoms in addition to epilepsy that may suggest a more multisystemic disorder.

If these two tests do not give an answer for your epilepsy, your doctor may order the following tests:

**Chromosomal microarray**
Looks for areas of your genetic material that are missing or duplicated that, if they are in regions that contain genes, may cause genetic disease.

**Karyotype**
A visual representation of a full set of your chromosomes that can identify large structural changes in the chromosomes.

**Whole Genome Sequencing**
Analysis of the entire DNA sequence to find genes that may explain your epilepsy. Whole genome sequencing may be offered clinically in some centers, but this is dependent on the clinic and also your insurance coverage. This kind of testing may also be offered through research studies.

What kinds of results can I get from genetic testing?

There are three kinds of results that you can get from a genetic test:

**Positive**
This means that a genetic change was found that explains your symptoms. Having a positive result gives you a genetic diagnosis.

**Negative**
This means that a genetic change that explains your symptoms was not found. It is important to note that a negative result does not necessarily mean that there is not a genetic explanation for your epilepsy. It just means that with the current knowledge and technology, a genetic change was not found at this moment.

**Variant of Uncertain Significance (VUS)**
This means that a genetic change was found, but with the current knowledge, there is not enough evidence to conclude that this result is diagnostic.

A genetic change can be inherited from either parent, or can be a new change that occurred during conception. A new change is called a *de novo* variant.
Next steps after getting genetic testing

If your testing is diagnostic:
- Speak to your neurologist! This diagnosis may change your treatment and can help predict long term outcomes.
- Reach out to a patient advocacy organization or a community support organization for your genetic condition. You can find a list of organizations on REN’s Members and Partners* page.

If your testing is not diagnostic OR testing did not give a definitive answer:
- As mentioned previously, this testing is a snapshot of what is known about genetics at that point in time.
- You can ask for a reanalysis of your results in the future, which may result in a diagnosis at that time. It is recommended that reanalysis be done every year. If you had genetic testing done in the past that was not diagnostic, you may be able to get a reanalysis now.

How do I pay for genetic testing?

Some insurance companies may pay for genetic testing, including Medicare and Medicaid. This may be state dependent and differ state to state. If your insurance does not cover the genetic testing, most labs offer a $250 self-pay option for panel testing.

There are also industry and other sponsored genetic testing panels that may be available for free. An example is Invitae which offers a free epilepsy panel, called Behind the Seizure*, for anyone under the age of 8 who has a minimum of one unprovoked seizure. Some other are listed below.
- Probably Genetics* – requirements include symptoms suggestive of a pediatric epilepsy disorder
- Ambit Care* – free virtual genetic counseling
- TMA Precision Health* – Free DNA sequencing for patients with a rare disease.

There are certain considerations with sponsored/industry genetic testing. If you receive testing, it is important to understand how your genetic information is being used, how you will be able to access your genetic information, and if it is possible to speak with a genetic counselor about your results.

You can discuss all of these options with your doctor.

Additional Resources
https://www.rareepilepsynetwork.org/
https://www.epilepsy.com/causes/genetic/testing

Document Links*
NSGC Epilepsy Practice Guidelines (https://onlinelibrary.wiley.com/doi/10.1002/jgc4.1646) – you can share these guidelines for genetic testing with your doctor
https://www.rareepilepsynetwork.org/members-partners
https://clinics.acmg.net/
https://www.naec-epilepsy.org/about-epilepsy-centers/find-an-epilepsy-center/
https://ambitcare.com/genetic-counseling-and-testing/
https://www.tmaprecisionhealth.com/
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