

What is genetics?

Genetics is the study of how traits such as hair and eye colour, and risk for disease are passed on or “inherited” from parents to their children. Genetics influences how these traits can be different from person to person.

Your genetic information is called your genetic code or “genome” which is made up of a chemical called deoxyribonucleic acid or DNA and is stored in almost every cell in your body.

What role do genetics play in epilepsy?

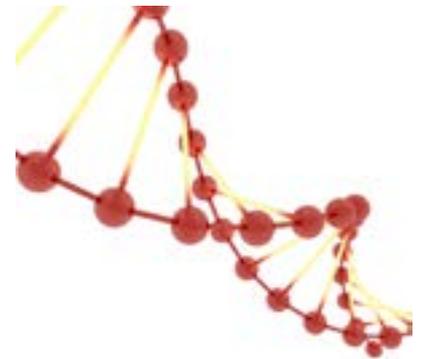
Genetics plays a noteworthy role in epilepsy, particularly in with drug resistant epilepsy.

While many people with epilepsy have no family relatives with epilepsy, there are many types of epilepsy that do run in families. Some epilepsy syndromes are completely determined by genetics, and genes are a major factor in other syndromes. Some inherited metabolic conditions also increase the risk of having seizures, as do some chromosomal disorders.

This means that genetic disorders can cause epilepsy as a single condition or can result in a syndrome or disorder where epilepsy is just one of a number of different effects on the body.

When epilepsy does arise from genetics, it can be:

- related to a specific gene,
- a combination of genetics and environmental factors,
- mutations in the DNA in mitochondria,
- missing or mutated chromosomes or
- changes in the activity of genes.



Genetic testing

Genetic research in epilepsy has led to the start of a range of clinically available genetic tests. Usually, genetic testing requires a blood or saliva sample to be taken and then sent to a laboratory for testing. The test looks at the DNA in the person’s blood or saliva. The sample is analysed for mutations or changes in a subset of genes that have a known association with different types of epilepsy.

By examining the changes in a person’s genes, researchers have been able to identify altered genes that lead to some peoples’ epilepsy.

Why have genetic testing?

Genetic testing may provide a more accurate diagnosis which can enable better management and prognosis. By using genetics to diagnose and treat each person individually, treatments can be tailored for different types of epilepsy. Specific precision therapies are emerging, and medications can be selected that are known to be effective for specific genetic epilepsies.

A specific genetic diagnosis avoids unnecessary testing with repeated blood tests, MRIs, invasive biopsies, pre-surgical workup, and even intracranial electrodes in the process of diagnosis. It also enables specific genetic counselling and the option for other family members to be tested.

A genetic diagnosis can be a relief for many families who have been searching for a cause of the epilepsy.

When genetic testing might be helpful

For people with epilepsy and families genetic testing:

- Allows doctors to estimate the risk to others in the family
- Is useful for reproductive planning
- Ends the search for a diagnosis
- May reduce parental guilt or shame
- Allows for improved knowledge about their condition and sourcing support



For treating medical practitioners genetic testing:

- Can sometimes enable changes in medical management
- Allows for prediction of epilepsy progression
- Enables genetic counselling
- Enables enrolment in clinical trials and research
- Can decrease the time and cost of diagnostic and treatment journey

Challenges

Many people expect that genetic testing will yield a diagnosis, specific prognostic information, and a specific treatment. This may not always be the case.

The testing might find positive results in genes that are not expected to be responsible for the persons epilepsy symptoms, as well as secondary positive findings in genes of relevance to other disorders.

Positive test results may be distressing for other family members who are possibly carrying the same gene. However, carrying a gene variation does not necessarily mean it will lead to developing the condition or disease.

Genetic testing may not always be affordable for everyone.

Who should have genetic testing

It is best to do genetic testing on a person who already has a definite diagnosis of epilepsy but has a suspected genetic cause of their epilepsy.

The chance of finding a genetic cause for epilepsy is higher when there are other family members who have similar symptoms. However, people with no previous family history of epilepsy may also have a genetic form of epilepsy.

A neurologist can recommend when genetic testing would be useful, choose the appropriate testing, explain the findings and refer to genetic counselling when appropriate.

What are the costs

The cost of genetic testing procedures varies, from less than \$100 to more than \$1000, depending on a number of factors and any health insurance rebates. It is best to speak with your neurologist about your specific situation.

Future outlook

The field of epilepsy genetics is expanding rapidly, and new genes associated with epilepsy are being identified. Identifying genes for epilepsy provides researchers with important information towards new treatments for the condition including the development of personalised and precision therapies for people with difficult and complex epilepsy. Finding drugs that directly target the genetic basis of epilepsy will ultimately lead to better seizure control and improved quality of life for people with epilepsy and their families.

Fact Sheet: Genetics in Epilepsy

Genetic research may also lead to a better understanding of why people with epilepsy experience other conditions such as depression and memory or learning difficulties, at a much higher rate than the general population.

Identifying the genes that cause epilepsy is particularly important when it is considered that a third of the 65 million people with epilepsy worldwide will not become seizure free using current treatment options.

For more detailed information on specific syndromes, please consult your doctor or a genetic counsellor.

For more information:

Introduction to Genetics <https://www.my46.org/intro/what-is-genetics>

Genetics and Epilepsy <https://www.aboutkidshealth.ca/Article?contentid=2059&language=English>

Genetic Testing <https://www.epilepsy.com/learn/diagnosis/genetic-testing>

Access to genetic testing Australia <https://www.alrc.gov.au/publications/10-genetic-testing/access-genetic-testing>

© Epilepsy Action Australia

This information is given to provide accurate, general information about epilepsy. Medical information and knowledge changes rapidly and you should consult your doctor for more detailed information. This is not medical advice and you should not make any medication or treatment changes without consulting your doctor.

