Epilepsy Action Australia

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EPILEPSY AND GENETICS

FACT SHEET

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EPILEPSY AND GENETICS

Genetics plays a notable role in epilepsy, particularly in drug resistant epilepsy.

While some epilepsies are caused by acquired factors like brain injury, many epilepsies are influenced by genetic factors. These genetic factors can be inherited or arise spontaneously.

What is genetics?

Genetics is the study of how traits, like hair colour, eye colour, or the risk for certain health conditions, are passed from parents to their children. It influences how these traits can be different for each person.

Many health conditions have a genetic link. A genetic disorder happens when there is a change (mutation) in the DNA, away from the normal sequence. This change can:

- Affect one gene or several genes
- Be a mix of genetic changes and environmental factors
- Happen because of damage to chromosomes (the structures that hold our genes)

Hereditary means a condition is passed down from a parent to a child and can continue through generations.

Genetic means it's caused by a change in DNA. It may be hereditary, but sometimes the change is new (not inherited from either parent).

Epilepsy and Genetics

Many types of epilepsy have a genetic basis.

- Some epilepsy syndromes are completely determined by genetics
- A genetic cause of epilepsy doesn't always mean it runs in the family.
- 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 one of many symptoms. 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 (part of the cell responsible for energy production),
- missing or mutated chromosomes or
- changes in the activity of genes.

Identifying the genetic basis of epilepsy can provide valuable information for individuals and families, including diagnosis, prognosis, and potential treatment options

GENETIC TESTING FOR EPILEPSY

Genetic research has helped create a range of tests that can look for changes in a person's DNA linked to epilepsy. Testing is usually done with a small sample of blood or saliva, which is sent to a laboratory. The lab examines specific genes known to be connected to different types of epilepsy, looking for changes (mutations) that may explain why seizures are happening.

By finding these changes, doctors can sometimes identify the exact genetic cause of a person's epilepsy.

Why have genetic testing?

Genetic testing can:

- Provide a more accurate diagnosis
- Avoid unnecessary tests during diagnosis
- Help doctors choose the most effective treatments for the specific type of epilepsy
- Offer access to new, targeted therapies and precision medicine
- Allow for genetic counselling and testing for other family members if needed
- Give families answers they have been searching for — which can bring relief and reassurance

NOTE:

Many people expect genetic testing will provide a specific diagnosis. This is not always the case.

The testing may find positive results in genes that are not expected to be responsible for the person's epilepsy, 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.

When can genetic testing be helpful?

For people with epilepsy and their families:

- Helps doctors estimate the risk for other family members
- Supports reproductive planning
- Ends the search for a clear diagnosis or cause
- Can reduce feelings of guilt or blame
- Improves understanding of the condition and helps connect with the right support

For doctors and healthcare teams:

- May lead to changes in treatment or management
- Helps predict how the epilepsy may progress
- Clarifies whether family-risk counselling is needed
- May identify eligibility for clinical trials or research studies
- May shorten and reduce the cost of the diagnostic process

! Genetic testing may not always be relevant or affordable for everyone

WHO SHOULD HAVE GENETIC TESTING?

Someone 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?

Costs for genetic testing for epilepsy in Australia varies considerably depending on the specific test and whether it is covered by Medicare or Private Health. It is best to speak with your neurologist about your situation.

Future outlook

Researchers are discovering many new genes linked to epilepsy. Identifying these genes can lead to new treatments, including personalised therapies designed for people with complex or hard-to-treat epilepsy. This means future medicines could directly target the genetic cause of epilepsy, giving people better seizure control and improving quality of life for them and their families.

Genetic research may also help explain why some people with epilepsy are more likely to experience other conditions such as depression, memory problems, or learning difficulties.

If you would like more detailed information about specific genetic syndromes, speak with your doctor or a genetic counsellor.

Contact us on:

Phone: 1300 37 45 37

Email: epilepsy@epilepsy.org.au

Further information

- Genetics for families
- National Health and Medical Research Council's advice on genetic testing
- NSW Health Centre for Genetics Education