

Epilepsy Gene Testing

Genetic Technologies

Support Services

Many excellent support services exist to provide education, information and assistance to people with epilepsy and their families, such as:



www.epilepsy.org.au
Toll free 1300 EPILEPSY (37 45 37)



Australia's leading consumer led epilepsy organisation
... Bringing epilepsy out of the shadows ...

www.epilepsyaustralia.org
Toll free 1300 85 28 53

It is recommended that individuals thoroughly discuss issues surrounding any genetic testing with an appropriate medical professional before undergoing testing.

For further information, visit:
www.genetictechnologies.com.au

Disclaimer

This brochure provides general information about genetic testing for epilepsy. It does not provide specific advice. Recommendation for testing should be initiated by a doctor.



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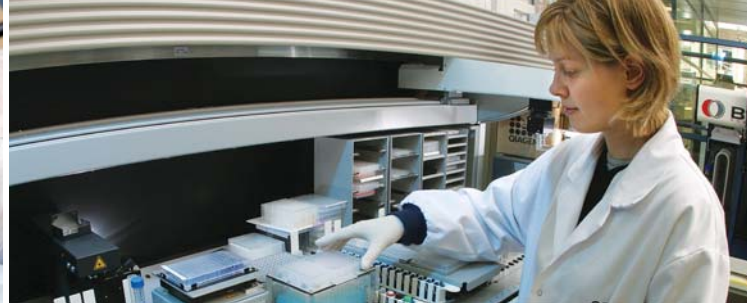
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What is Epilepsy?

Epilepsy is a neurological (nervous system) condition that presents with the occurrence of seizures (sometimes called 'fits', 'attacks', 'convulsions' or 'spasms'). Seizures can cause a person to lose consciousness or alter their awareness, behaviour or feelings.

Up to 3% of the population may suffer from one of the many forms of epilepsy. It affects people of all ages and ethnic backgrounds, but often develops in early childhood.

Sometimes the reason epilepsy develops is obvious, for example following brain injury or infections of the brain. Unfortunately in many instances the cause of epilepsy remains unknown.

Breakthroughs in genetic research have now identified particular genes responsible for certain types of epilepsy, for which the cause was previously unknown.

Diagnosing Epilepsy

A diagnosis of epilepsy is based upon what happens leading up to, during and after a seizure. Doctors frequently rely on eyewitness accounts of seizures which can be difficult to describe accurately. To support the diagnosis, doctors order additional tests for confirmation.

Genetic testing is now being used to identify changes in the gene known as *SCN1A* to assist doctors in diagnosing a number of epilepsy syndromes.

You, Your Genes and Epilepsy

Genes are the instructions that tell our bodies how to grow and develop. A change in a gene (sometimes called a mutation) can alter the instruction the gene sends to the cells of the body. The *SCN1A* gene plays an important role in controlling the way that chemical signals pass through the brain. Changes in the gene can affect this signaling process. This can contribute to the onset of a number of epilepsy syndromes which are included under the umbrella of the Generalised Epilepsy with Febrile Seizures Plus (GEFS+) spectrum:

- Severe Myoclonic Epilepsy of Infancy (SMEI) or Dravet Syndrome;
- Borderline Severe Myoclonic Epilepsy of Infancy (SMEB);
- Intractable Childhood Epilepsy with Generalized Tonic-Clonic Seizures (ICEGTC); and
- Febrile Seizures Plus (FS+).

Is Epilepsy Inherited?

Most changes in the *SCN1A* gene arise spontaneously during early embryonic development, which means that a change in the *SCN1A* gene is rarely of an inherited nature. However, in approximately 1 in 20 cases, a *SCN1A* gene change may be inherited from either parent. This parent may or may not experience epilepsy themselves.

Why Have the *SCN1A* Gene Test?

The *SCN1A* gene test assists doctors in making an accurate, early and definitive diagnosis for individuals experiencing seizures.

This helps doctors provide patients and families with information on the potential severity of the epilepsy and to select the optimal management options.

In addition, if a *SCN1A* gene change is identified in patients, parents and extended family members may access testing to determine if the change is of an inherited nature.

How is Testing Initiated?

All requests for genetic testing of children or other family members for epilepsy must be made through a medical practitioner. Tests results are confidential and will only be issued to a doctor, who will provide counselling, test interpretation and advise on management options.