

## Feature 1: Genetics in Epilepsy

### 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.” Your genome 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 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 several factors and any health insurance rebates. It is best to speak with your neurologist about your specific situation.

**After years of being told seizures were "just in her head", a 17-year-old Teresa finally learnt what was causing her mystery illness. Read her story here**

<https://www.abc.net.au/news/2017-12-26/dna-test-reveals-defective-gene-the-cause-of-teenagers-seizures/9224268>

### Future outlook

The field of epilepsy genetics is expanding rapidly, and new genes associated with epilepsy are being identified often. Identifying genes for epilepsy provides researchers with important information towards new treatments for the condition and the development of personalised and precision therapies for people with difficult and complex epilepsies. 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.

Genetics plays a major role in epilepsy, particularly in people with seizures resistant to medication. The neurologist can triage who most needs and could benefit from genetic testing, choose the appropriate testing, and explain the findings.

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.

For more detailed information on specific syndromes and genetic testing, 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>

## Feature 2: Focal Seizures

What type of seizures do you have? There are many terms used for focal seizures; some people use the term “petit mal” whilst others say partial or even absence seizures. There have been recent changes to seizure naming and organisation and these non-convulsive seizures are now called *focal seizures*. It can be confusing to say the least. Here we will discuss focal seizures.

### Old terms

Probably the oldest terms for seizures that are still incorrectly used are *grand mal* and *petit mal*. They are French terms meaning great sickness and little sickness and usually refer to convulsive seizure or non-convulsive seizure.

We then moved on to *partial seizures* which were sometimes used to refer to any minor or non-convulsive seizure. Partial seizures were split up into *simple partial seizure* and *complex partial seizure*. Simple partial seizures were also sometimes called an *aura* because there was complete awareness for the seizure. Complex partial meant the person lost or had impaired awareness and often became disorientated or confused and had behavioural changes.

### New terms

Seizures are now divided into groups depending on:

- where they start in the brain
- whether or not a person's awareness is impaired
- whether or not seizures involve movement

Depending on where they start, seizures are described as either focal onset, generalised onset or unknown onset. Focal onset seizures

The most recent changes have now called the petit mal or partial seizures, *focal onset seizures*.

*Focal onset seizures are seizures that start in one small area or one hemisphere of the brain.*

There are a few subgroups of focal onset seizures. They include:

### 1) Focal Aware Seizures

In these seizures the person is conscious (aware and alert) and will usually know that something is happening and will remember the seizure afterwards. Sometimes they may not be able to move or communicate during the seizure, but they are aware of what is happening.



- Focal aware seizures usually last less than two minutes and are distinguished by the symptoms that are seen or felt. They can be further divided into:
  - Focal sensory seizure– numbness, tingling or burning sensation in a region of the body, unusual smell or taste, dizziness, feeling hot or cold
  - Focal motor seizure – jerking of a limb, twitching of the face, weakness or paralysis of a muscle group
  - Focal autonomic seizure – blushing, pallor, heart-rate changes or palpitations, nausea
  - Focal emotional seizure – changes in mood or emotion such as fear, panic, laughing, crying
  - Focal cognitive seizure – unable to speak, speech difficulty, déjà vu, visual hallucinations, hearing sounds
  - Focal behaviour arrest seizure – the person just “freezes” and appear unable to move or talk

Remember the person know these focal aware seizures are happening and may not even regard them as a seizure, but as a warning of an impending seizure. What causes these symptoms though is seizure activity in a small region of the brain.

When a focal aware seizure ends, the person usually continues doing whatever activity they were doing before it started. This type of seizure can also develop into a stronger seizure with impaired awareness or loss of consciousness (focal impaired awareness and/or tonic clonic seizures)

Quite often people think they just have focal aware seizures, but there often can be a loss of awareness without the person

realising until an eyewitness describes to them what their behaviour was during the seizure.

2) **Focal Impaired Awareness Seizures** – these are often simply called focal seizures.

Awareness during a seizure is described as the person being fully aware of themselves and their environment throughout the seizure, even if they are immobile. If awareness is impaired at any point during the seizure, the seizure is then called a focal impaired awareness seizure. The degree of loss of awareness may vary.

In these seizures the person’s awareness is diminished, and they may be confused, may or may not be able to hear you, and will not fully understand what you say or be able to respond to you. Often, they lose memory for some or all the seizure.

These seizures typically last from 1 to 2 minutes but the post-seizure period can last several minutes. Symptoms vary but can include; blank staring or confusion, chewing or lip smacking, picking at clothes, fumbling, repetitive movements, unusual vocalisation, becoming unaware of surroundings, and wandering. Sometimes these seizures are very subtle and not recognised as a seizure, and other times they are mistaken for intoxication or psychiatric illness. This seizure type may be preceded by a focal aware seizure (previously referred to as an aura or warning)

Afterward, the person may be tired or confused for about 5 - 15 minutes and may need to rest and not be able to return to normal function for hours.

Sometimes these seizures can spread to the other side (hemisphere) of brain and develop into a tonic clonic seizure.

**Focal Onset Seizures**

Seizure Type	Older Terms	Seizure types seen
<b>Focal Aware Seizures</b>	Petit Mal, Simple Partial Seizure, Aura, Warning	Focal sensory, Focal motor, Focal emotional, Focal cognitive, Focal autonomic and Focal behaviour arrest

<b>Focal Impaired Awareness Seizures</b>	Petit Mal, Complex Partial Seizure, Focal Dyscognitive Seizure	Any focal seizure which involves impaired awareness
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The most common focal seizures arise from the temporal and frontal lobes. Sometimes people are told they have temporal lobe epilepsy or frontal lobe epilepsy. These are correct, more specific terms to describe the type of epilepsy.

Because focal seizures arise from a small part of the brain, if they are not successfully controlled by medication, then surgical removal of the seizure focus is possibly an option.

It’s not unusual for someone not to know the type of seizure or epilepsy they have. Often seizures are diagnosed based on descriptions of what an eyewitness has seen. These descriptions may not be fully complete, or one can’t tell where a seizure begins from this information.

Unfortunately, focal seizures can quite often be misunderstood or mistaken for other events. Diagnosis can take some time if this happens.

**In the News – The latest on epilepsy**



**How epilepsy may arise in a healthy brain**

Scientists at McGill University Canada have developed a new method to study how seizures arise in the healthy brain to better understand how seizures arise in humans.

<https://www.mcgill.ca/newsroom/channels/news/using-laser-light-study-how-epilepsy-arises-healthy-brain-295750>

**A new treatment approach is needed to improve prognosis of epilepsy.**

During the past two decades, more than two dozen new recognised anti-epileptic drugs have become available, however, the long-term prognosis of anti-epileptic drug treatment, however, hasn’t improved.

<https://lens.monash.edu/2019/03/25/1374040/epilepsy-urgent-need-to-improve-diagnosis>

### The link between concussions and epilepsy

Although concussions range in severity, and many people walk away relatively unscathed, the side effects that come after a concussion can be lengthy.

<https://www.robinspost.com/news/consumer-news/consumer-daily-reports/843418-consumer-news-new-study-shows-the-link-between-concussions-and-development-of-epilepsy.html>

### Andrew Johns' admission that concussions most likely contributed to his epilepsy

<https://www.stuff.co.nz/sport/league/111356655/rugby-league-legend-andrew-johns-shock-admission-on-concussions-contributing-to-his-epilepsy>

### Starstim – using mild electrical currents to treat drug-resistant epilepsy

Starstim, a device that uses transcranial current stimulation to deliver mild electric currents to the scalp to calm abnormal activity of the brain resulted in a reduction in seizure frequency of at least 40% from baseline in 75% of the patients.

<https://neuronewsinternational.com/starstim-medication-resistant-epilepsy/>

### Tweet goes viral after helping a stranger having a seizure

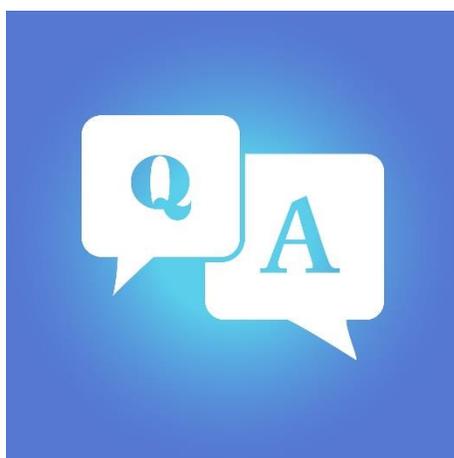
A media student in Canada writes about her encounter with a stranger on a train who has a seizure highlighting the reality of living with epilepsy <https://uk.style.yahoo.com/woman-goes-viral-twitter-encounter-stranger-highlights-reality-living-epilepsy-095815458.html>

### Study uses focussed ultrasound to treat epilepsy

Researchers at The Ohio State University College of Medicine are studying how well focused ultrasound surgery works in adults with a specific type of epilepsy whose seizures are not controlled by medication. [https://www.crescent-news.com/on\\_the\\_wire/health/study-uses-focused-ultrasound-to-treat-epilepsy/article\\_576739b2-2309-5600-afe9-31eb7e6acd25.html](https://www.crescent-news.com/on_the_wire/health/study-uses-focused-ultrasound-to-treat-epilepsy/article_576739b2-2309-5600-afe9-31eb7e6acd25.html)

[https://www.crescent-news.com/on\\_the\\_wire/health/study-uses-focused-ultrasound-to-treat-epilepsy/article\\_576739b2-2309-5600-afe9-31eb7e6acd25.html](https://www.crescent-news.com/on_the_wire/health/study-uses-focused-ultrasound-to-treat-epilepsy/article_576739b2-2309-5600-afe9-31eb7e6acd25.html)

**Q&As – Our service providers answer your questions**



### Q. I was diagnosed with epilepsy many years ago and I have a lot of side effects, particularly memory loss. Are there any online groups I can join to talk to?

A. There are many online support groups, too many to list here. We often recommend *Forum4e* or *MyEpilepsyTeam* and if you join one or both sometimes you can source a smaller group through them if that is what you prefer. These are the details of online support groups in this Factsheet (<https://www.epilepsy.org.au/wp-content/uploads/2019/02/Fact-Sheet-Online-Support-Groups.pdf>)

Memory difficulties are one of the biggest complaints for people with epilepsy. I've added this link with some videos from Neuropsychologist Dr Laurie Miller discussing different aspects of memory <https://www.epilepsy.org.au/psychosocial-wellbeing-for-adults-living-with-epilepsy/optimal-living-with-epilepsy/memory/> and this is a link to our web page specifically about memory <https://www.epilepsy.org.au/about-epilepsy/living-with-epilepsy/lifestyle-issues/memory/>

### Q. I have been seizure free for over 10 years and no longer taking medication for over 5 years. I recently went to reapply for my license and saw a new doctor. He has said I cannot get my license without recommencing medication. Is this right? I do not want to take medication if I am not having seizures.

A. I'm sorry to hear you have been told this. Certainly, there are different situations for individuals, but from what you described, it sounds like you are well within your rights to expect to return to driving.

The new classification and definition of epilepsy includes this: "Epilepsy is considered to be *resolved* for individuals who had an age-dependent epilepsy syndrome but are now past the applicable age or those who have remained seizure-free for the last 10 years, with no seizure medicines for the last 5 years." So your epilepsy would now be considered "resolved" according to this (ILAE) definition (<https://www.ilae.org/guidelines/definition-and-classification/the-2014-definition-of-epilepsy-a-perspective-for-patients-and-caregivers>)

Page 83 onwards of this Assessing Fitness to Drive document

([https://austroads.com.au/data/assets/pdf\\_file/0022/104197/AP-G56-](https://austroads.com.au/data/assets/pdf_file/0022/104197/AP-G56-17_Assessing_fitness_to_drive_2016_amended_Aug2017.pdf)

[17\\_Assessing\\_fitness\\_to\\_drive\\_2016\\_amended\\_Aug2017.pdf](https://austroads.com.au/data/assets/pdf_file/0022/104197/AP-G56-17_Assessing_fitness_to_drive_2016_amended_Aug2017.pdf)) explains many circumstances related to seizures and epilepsy where people can or cannot drive and how long they should expect to lose their license for.

I think it is unreasonable to expect you to recommence medication for this reason. My first advice is to see a different neurologist (maybe one that is an epilepsy specialist) or even contact your original neurologist if you can. Ideally, it's best to see someone who knows your medical history.

This is the position of the Epilepsy Society of Australia (run by neurologists) <https://www.epilepsy-society.org.au/resources/driving-guidelines.asp>

**Q. I'm 29 years old and have had epilepsy for 8 years. The doctor said it was "frontal epilepsy". Most of my seizures are during sleep and very mild. Occasionally I get really bad seizures where my whole body jerks and it takes a couple of hours to recover. I take carbamazepine 400mg a day. Is this the right medicine? Why aren't my seizures stopping? Am I at risk for sudden death?**

If you have frontal lobe epilepsy, then carbamazepine is one of the drugs that is often used to manage it. There are many epilepsy drugs, but carbamazepine has been used for a long time and has worked quite well for many people with focal epilepsies.

There are many different brands and formulations of carbamazepine, some last longer in the body than others, and the doses people take varies greatly also. The average dose for carbamazepine is up to 800-1200 mg/day. This obviously depends on side effects the medication causes you and effective seizure control. So, if your seizures are controlled with a lower dose, then there is no need to raise it. However, your seizures are not controlled, so it is worth discussing with your doctor about increasing your dose. This is an article explaining carbamazepine dosing

(<https://reference.medscape.com/drug/tegretol-xr-equetro-carbamazepine-343005>)

Here is more about carbamazepine. Your pharmacist should also be able to help with information.

(<https://www.healthdirect.gov.au/medicines/brand/amt,3827011000036102/carbamazepine-sandoz>)

You are right to be concerned about seizures during your sleep. There are more risks associated with the (tonic clonic) seizures you describe in your sleep than any other seizure type, so the best way to lessen your risk is to reduce your seizures. This information talks about reducing your risk.

<https://www.epilepsy.org.au/epilepsy-and-risk-taking-action-against-risk/>

It would be worth seeing your treating doctor to ask about your medication and dosages.

**Q. My seizures are very reactive to stress, so when I am under pressure, I have more seizures. I also have depression and taking medication for it. What can I do myself to help manage my stress and depression? I cannot drive so would prefer telephone or online support.**

There are many, many online services, information and apps that can help you. I will suggest a few here but have a look and see what you think so you can find something to best suit your needs.

**Call to Mind** is a telepsychiatry service that allows people across Australia to access quality mental health care. There is a team of psychiatrists and psychologists who consult via video-conferencing platform, so you can attend your appointment from anywhere. You need a referral from your GP and can access your consult from a computer, smart phone or tablet at home or maybe even at your GP's practice.

<https://calltomind.com.au/>

**The MindSpot Clinic 1800 614 434** MindSpot is a free service for Australian adults who are experiencing difficulties with anxiety, stress, depression and low mood. They provide assessment and treatment courses, or can help you find local services that can help. <https://mindspot.org.au/>

**Counselling at Your Place** is an Australian Skype and telephone counselling service open from 10am to 10pm, 7 days a week 365 days a year. They can confidentially debrief and provide therapeutic counselling support without the need to leave the safety and comfort of your own home. They can help with many issues which includes stress, depression and anxiety. <http://www.counsellingatyourplace.com.au/>

**Beyond Blue 1300 22 4636** has a wealth of information and online support for depression, anxiety and stress

<https://www.beyondblue.org.au/>

**SANE Helpline 1800 187 263** <https://www.sane.org/> Allows you to talk or chat online to a mental health professional (weekdays 10am-10pm AEST). They also have a lot of information and online support and forums.

**THIS WAY UP™** provides online learning programs and CBT, education and research in anxiety, depressive disorders and physical health. <https://thiswayup.org.au/>

**MoodGym** is an online self-help tool to help prevent and manage symptoms of depression and anxiety.

<https://moodgym.com.au/>

**Moodjuice Self Help Guide** is a free online book about stress and self help <https://www.moodjuice.scot.nhs.uk/Stress.asp>

Stress: There are endless apps to help manage stress. Some well-known ones include:

**Headspace** is a comprehensive meditation app, with guided and unguided meditations to help you through all phases of your life. It offers ten free sessions, with hundreds of hours of extra content if you love it and want to subscribe.

<https://www.headspace.com/>

**Stop Breathe Think** aims to give you the chance, and skills, to stop everything, just breathe, and think. The guided meditations cover a range of goals and are constructed to help you take some time out and concentrate on yourself. <https://www.stopbreathethink.com/>

**Calm** is designed to help you be calm. With stunning backgrounds that you can stare at for ages, and a range of meditations, Calm provides a space for you to get away from it all, without having to go anywhere. <https://www.calm.com/>

**Smiling Mind** is a free app meditation program developed by psychologists and educators to help bring mindfulness into your life. <https://www.smilingmind.com.au/>



include intricate illustrations, detailed portraits, enormous abstracts and even eye-catching photography.

This year we have pieces starting from just \$10, right up to many hundreds of dollars. It's an opportunity not to be missed! The Auction runs from 1<sup>st</sup> May the end of the month with various pieces being added every week and closing at different times. So be sure to keep checking the site - **check it out here:** <https://www.artforepilepsy.com.au/> See just some of the pieces below up for grabs!

## Taking Action – What's happening at Epilepsy Action

### ART FOR EPILEPSY 2019!

**Art for Epilepsy** is back for 2019! Don't miss this opportunity to get an amazing artwork while helping those impacted by epilepsy.

Now in its 4th year, Art for Epilepsy is an online art auction that sees artists from around the country donate original works that incorporate the colour purple – the internationally recognised colour for epilepsy awareness – with all proceeds going to support the work of Epilepsy Action Australia. Donated pieces come from a broad spectrum of visual art disciplines, and

### GLOBAL HEALTH INITIATIVE CONFERENCE & MASTERCLASS

Not-for-profit organisation Global Health Initiative, a foundation focused on medicinal cannabis education and research, is pleased to present a one-day conference and one-day masterclass on medicinal cannabis and treatment of mental health conditions in Melbourne on 20 & 21 July 2019. Our CEO, Carol Ireland is a Consumer Representative on the GHI Advisory Board.



The conference is open to all healthcare practitioners who wish to understand more about medicinal cannabis and other nutritional medicine approaches to treatment of common mental health conditions. We would love it if you could share this information with your doctors, as bookings are essential to attend. They can find out **more information here:** <https://www.epilepsy.org.au/global-health-initiative/>

To register or for more information on the Australian Pregnancy Register visit <https://www.epilepsy.org.au/research/research-projects/>



**THE AUSTRALIAN PREGNANCY REGISTER IS SEEKING WOMEN:**

- Planning a pregnancy, currently pregnant or recently given birth?
- On an antiepileptic medication with or without a diagnosis of epilepsy?
- Have a diagnosis of epilepsy and not on antiepileptic medication?

If your answer is yes to two of the three questions above,  
please register here for this important research.

**REGISTER NOW**