Genetics and epilepsy

What is genetics?



Genetics is the scientific study of how certain features and characteristics are passed on, or 'inherited' from parents to their children before the children are born, and the effect a person's genes has on their health.

As research develops, we are learning more about our genes, and how they affect our chances of developing different medical conditions and how we respond to medication.

Researchers now believe that the chance of developing epilepsy is probably always genetic to some extent. This means that most people with epilepsy have a genetic tendency to have seizures.

This is the case even if a person starts having seizures after a brain injury or if they have damage to the brain left by a stroke or infection. This damage, combined with the underlying genetic tendency, causes seizures to start. This makes sense if we consider that many people might have a similar brain injury, but not all of them develop epilepsy afterwards.

Any of us could have a one-off single seizure under certain circumstances, but for most people, their natural resistance to having seizures is high enough to stop that happening. This natural resistance is part of our genetic makeup, and is sometimes called our 'seizure threshold'.

DNA and genomes

Our DNA (deoxyribonucleic acid) is made up of three billion letters to form a complete set of instructions that our bodies need to grow and develop. All of these instructions together make up our genome.

DNA is made up of four components, Adenine (A), Thymine (T), Cytosine (C) and Guanine (G). These components are repeated over and over in a different order, to make up several long strands. These strands are called chromosomes. When the letters in your DNA are rearranged, they give different characteristics, which is one reason why we look different from each other.

Although most of our genetic material is inherited, a person's DNA can go through small changes (or 'mutations') before they are born. These are called "de novo" changes. They are new genetic changes that are not passed down from a parent. Everyone has some genetic differences, which is why we are not exactly the same as our parents.

For many people, their epilepsy may be partly due to a mutation in one or more of their genes rather than from their parents.

Is there a 'gene for epilepsy'?

There is no one single gene for epilepsy. So far, more than 1500 genes associated with epilepsy have been identified. Some epilepsies have several genes that may contribute in some way to the person's chance of developing that epilepsy.

Does 'genetic' mean my epilepsy is inherited?

Genetic conditions can happen when you inherit an altered or changed gene from your parents. This can increase your chance of developing a genetic health condition. However, as changes to genes can happen randomly before we are born, but after we are conceived, not all genetic conditions are passed down from our parents.

However, as changes to genes can happen randomly, hereditary, or 'inherited', conditions or diseases are caused by changes to all our genetic material, sometimes called our genome. These are passed on by our parents. Epilepsy can sometimes be a symptom of rare hereditary conditions, such as tuberous sclerosis and neurofibromatosis.

Visit genomicseducation.hee.nhs.uk/genotes/knowledge-hub/tuberous-sclerosis-complex/
Visit nhs.uk/conditions/neurofibromatosis-type-1/

Epilepsy is not just one condition, but is a term used to group together many conditions ('the epilepsies') which share the common symptom of seizures.

Helpline 0300 102 0024
Confidential, information, and emotional support.
Visit epilepsysociety.org.uk/helpline for opening hours.

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Some types of epilepsy called 'familial epilepsies' run in families. These include Benign familial neonatal seizures and Generalised epilepsy with febrile seizures plus (GEFS+).

Could my child inherit my epilepsy?
Most parents with epilepsy do not have children with epilepsy, and the chances of inheriting epilepsy are generally low. The risk for any child to develop epilepsy by the age of 20 is around 1 in 100 (1%), and the risk may increase to around 2 to 10 in 100 (2 – 10%) for most children of parents with epilepsy.

However, the chances of your child inheriting epilepsy will vary, due to different factors. These include what type of epilepsy you have, and whether it may be due to a genetic change that can be identified, at what age it started, and whether your partner or another child of yours has epilepsy.

All these factors, plus others which may be unique to your child's own genetic makeup, contribute to the chance of your child developing seizures. Your neurologist can give you more information about your epilepsy.

What is genetic testing?

Genetic or genomic testing can help to identify changes in genes that can cause certain health conditions. It usually involves taking a sample of your blood or saliva and sending it to be analysed at a laboratory.
Visit nhs.uk/conditions/genetic-and-genomic-testing/

Whole genome sequencing
This test looks at all your genes at the same time. It focuses on changes in genes that are related to your epilepsy, rather than changes in genes that cause other conditions. This is because there is so much information to process. The results may help to:

- diagnose your condition;
- suggest the best treatment options;
- monitor other parts of the body that may be affected; and
- tell you whether your condition could be inherited, to help you with family planning.

This could mean other members of your family may be affected, or that it could be passed on to your children. If this is the case your relatives may be offered testing.
Visit epilepsysociety.org.uk/genomic-clinics
Visit epilepsysociety.org.uk/research/genomics

Can I have genetic testing?
Guidance published by the National Institute for Health and Care Excellence (NICE) recommends whole genome testing specifically for people whose epilepsy:

- started before two years of age; and
- is associated with developmental disorders or certain clinical features that are more likely to have epilepsy as a genetic cause.

Visit nice.org.uk/guidance/ng217/chapter/1-Diagnosis-and-assessment-of-epilepsy#genetic-testing for more information.

Genetic counselling
Before having genetic testing, you may be referred to a genetic counsellor or a genetics clinic. You can discuss how much information you want to know, and whether it would be helpful to you. You may also want to think about what any results might mean for you and your family.

Further information

NICE (National Institute for Health and Care Excellence)
nice.org.uk/Guidance/ng217
NICE provides guidance on the diagnosis and treatment of epilepsy.

Epilepsy Society is grateful to Dr F J Rugg-Gunn, Consultant Neurologist & Honorary Associate Professor, Clinical Lead, Chalfont Centre for Epilepsy, who reviewed this information.

For a printed copy of this information contact our helpline.

Visit epilepsysociety.org.uk/comments-compliments-and-complaints to give us feedback on this information.

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