

Epilepsy and Genetics

What is genetics?

Genetics is the study of heredity or how different characteristics (also called traits) are passed from parent to child. A person inherits these traits, such as hair color or blood type, through the genes that they inherit from their parents. Human beings have many thousands of genes that are made up of DNA. Each gene results in the making of a protein, which is necessary for normal body function. Genes are packaged into larger structures called chromosomes, which are present in almost every cell (brain cell, muscle cell, skin cell) in the human body. Each cell holds 23 pairs of chromosomes (46 total). Half of the genes on the chromosomes are inherited from the mother and the other half from the father. As a result, genes are passed from both parents to the child.

What is the role of genetics in epilepsy?

Epilepsy can be caused by many different disorders, and genetic factors have been shown to play a role in many of these conditions. Only a few types of epilepsy syndromes are caused by changes (also called mutations) in single genes. These disorders may be passed on to future generations in a recognizable inheritance pattern or arise spontaneously through new mutations. In most epilepsies that have a genetic basis, the epilepsy is due to interactions between several genes and environmental factors. In these conditions, the epilepsy has a tendency to run in families, but the pattern of inheritance is usually difficult to identify.

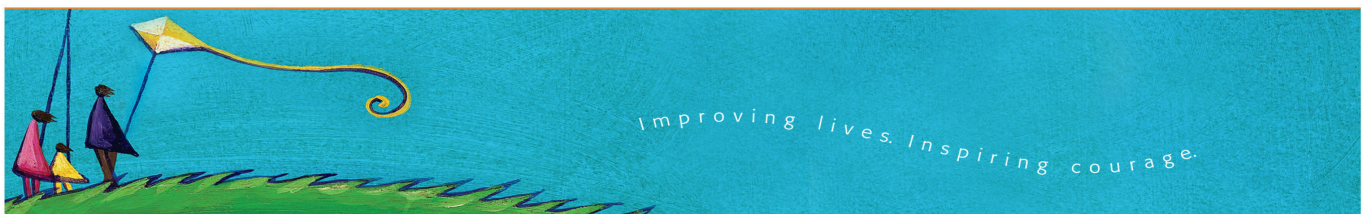
Other genetic disorders in which epilepsy occurs more commonly than in the general population includes some inherited metabolic conditions, some genetic syndromes, and some chromosomal disorders.

If a parent, brother or sister has epilepsy, how likely is it for other close family members to have seizures?

Certain types of epilepsy appear to run in families. Some studies have shown that the risk of epilepsy in siblings and children of people with a seizure disorder is about 5 % or about 1 in 20, but this will also depend on a number of factors described below. The risk of epilepsy in the general population is about 1% or 1 in 100. Although the risk is increased compared to the general population, most people with epilepsy will not have children or other family members with a seizure disorder.

What are some of the factors that seem to affect the risk of inheriting epilepsy?

- *Type of epilepsy.* People with generalized epilepsy (generalized means that the EEG pattern shows both sides of the brain being involved at beginning of a seizure) are slightly more likely to have other family members with seizures than those with a focal epilepsy (when an EEG pattern showing seizures beginning in a single area of the brain).



- *Cause of epilepsy.* The risk for developing epilepsy is not significantly increased in relatives of people with a seizure disorder caused by a brain injury that occurred after birth, e.g., stroke, brain tumor, brain infection or severe head trauma.
- *Age when epilepsy begins.* Relatives of people who develop epilepsy in childhood seem to have a higher risk of developing seizures than relatives of those with later onset of epilepsy.
- *Mothers and fathers with epilepsy.* The risk of epilepsy is about twice as high in children of women with epilepsy than in children of men with epilepsy. The reason for this is not yet known.

What kind of research is being done on the genetics of epilepsy?

There has been major research in the genetics of the epilepsies, including the discovering of epilepsy genes for some epilepsy syndromes. This discovery has helped in the diagnosis of some of these conditions. Further research into how these genes cause epilepsy could result in the development of new therapies, such as new antiepileptic drugs. Recent research in British Columbia has included a genetic study of severe myoclonic epilepsy of infancy. Further studies are currently underway in families with other epilepsy syndromes in infancy. A new technique that allows chromosome abnormalities to be seen in much greater detail is being pioneered in British Columbia.

Where can I find more information on epilepsy and genetics?

Information is available at the Epilepsy Foundation website at this webpage: www.epilepsy.com/learn/epilepsy-101/epilepsy-inherited and at Toronto's Hospital for Sick Kids Epilepsy website, www.aboutkidshealth.ca/Epilepsy (sections [An Overview of Genetics](#) and [Genetics of Epilepsy](#)). Additional questions, including specific risks of epilepsy in your family can also be discussed with your physician.

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