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Epilepsy: Genetics

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The term "epilepsy" describes a heterogeneous group of disorders, and caused by genetic, molecular and environmental factors. And other side the genetic epilepsies are due to particular gene mutations or defined structural chromosomal aberrations, such as microdeletions.

Genetic factors are likely to play a role in most cases, either because the fundamental cause of epilepsy is mainly genetic or because genes modulate exposure to an epileptogenic insult. Primarily genetic epilepsies include conditions within which altered brain development or neurodegeneration are at the basis of seizures, but also conditions in which the brain is completely normal, and the main, if not only, medical feature is epilepsy. These are

called idiopathic epilepsies. A few idiopathic epilepsies are monogenic disorders due to mutations in a variety of genes affecting neuronal excitability, synaptic transmission, or network development. Most cases have a difficult etiology that combines prompting genetic variants with nongenetic factors. Few of these have been recognized so far and only in very few affected individuals, consisting mostly of deletions of critical chromosomal regions. Genetic factors conjointly play a role in the response to antiepileptic drugs, affecting both their effectiveness and their tolerability. There have been recent advances in discovering such factors, in particularly those underlying risk to medication toxicity.

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