



# **Pediatric Neurology Part I: Chapter 59. Genetics of idiopathic epilepsies (Handbook of Clinical Neurology)**

*Rima Nabbout, Ingrid E. Scheffer*

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One of the most exciting areas in epilepsy has been the explosion in our understanding of the genetics of the epilepsies over the last decade. Built on a long history of careful clinical genetic studies of the epilepsies, the relatively recent discovery of epilepsy genes has enabled insights into pathways causing seizure disorders. A variety of mutational mechanisms can cause epilepsy resulting from different, and sometimes surprising, molecular processes such as copy number variation within the genome. The majority of known epilepsy genes encode ion channel subunits leading many of the genetic epilepsies to be regarded as channelopathies. Understanding how dysfunction of a mutant protein leads to hyperexcitability is key to understanding the pathophysiology of this group of serious and common childhood disorders. The architecture of the common genetic epilepsies following complex inheritance, where multiple genes are involved, is also beginning to be unraveled. The clinical approach to understanding the genetics of the epilepsies has matured and requires a detailed family history of seizures together with delineation of the child's epilepsy syndrome. Recognition of specific genetic epilepsy syndromes enables optimal treatment and prognostic and genetic counseling.

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