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### Ion channel genes mutation in epilepsy syndromes

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Epilepsy is a neurological disorder which is characterized by a long-term risk of recurrent seizures and affects up to 1% of the general population. Although the most of cases is unknown, but genetic factors play an important role in the development of this condition. Genetics is believed to be involved in the majority of cases, either directly or indirectly. Some epilepsy is due to a single gene defect; most are due to the interaction of multiple genes and environmental factors (multifactorial). More than 200 single gene defects contribute to epilepsy are described. All of these single gene forms of epilepsy display autosomal dominant inheritance. Mutations in cellular membrane ion channel genes impair Ion channels. Most genes contribute to single gene epilepsy syndromes are voltage-gated genes such as sodium channel *I*, calcium channel (*CACNA1A*, *CACNA1H*, *CACNB4*), potassium channel (*KCNA1*, *KCNQ2*, *KCNQ3*, *KCNMA1*), chloride channel (*CLCN2*); and ligand-gated such as acetylcholine receptor (*CHRNA2*, *CHRNA4*), gamma aminobutyric acid (*GABA*), GABA receptor (*GABRG2*, *GABRA1*, *GABRD*) and G protein-coupled receptors. These syndromes are distinguished by various affected organs and age of the onset. Epilepsy syndromes related to voltage-gated genes are: Generalized epilepsy with febrile seizures plus syndrome (GEFS+), severe myoclonic epilepsy of infancy (SMEI), intractable childhood epilepsy with generalized tonic-clonic seizures (ICEGTCs), infantile spasms (IS), episodic ataxia type 1,2 (EA1,2), spinocerebellar ataxia type 6 (SCA 6), etc. Epilepsy syndromes related to ligand-gated are autosomal dominant frontal lobe epilepsy (ADNFLE), juvenile myoclonic epilepsy (JME), etc. Based on the exact gene mutation, different type of epilepsy may occur. Understanding of the molecular pathogenesis of the epilepsy leads to targeted therapy.

### Biography

Mohsen Karimi is a Medical student at Tehran University of Medical Sciences. He holds both Iranian and British (IGCSE & A-level) High School Diplomas. He is a member of Universal Scientific Education and Research Network.

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