Some information about epilepsy caused by mutations in the KCNT1 gene:

KCNT1 is a potassium channel gene. Potassium channels are proteins that allow potassium to pass into and out of cells. Potassium channels play a key role in a cell’s ability to generate and transmit electrical signals. The formal name of this gene is: Potassium Channel, Subfamily T, Member 1.

What medical conditions are associated with mutations in this gene?

- Early Infantile Epileptic Encephalopathy Type 14 (EIEE14)
- Nocturnal Frontal Lobe Epilepsy Type 5 (NFLE5)

What are the some of the features of these medical conditions?

_Early Infantile Epileptic Encephalopathy Type 14 (EIEE14)_

- Seizures, partial with secondary generalization, focal at onset, usually motor
- Developmental regression, severe
- Hypotonia
- Lack of speech development
- Lack of motor development
- Quadriplegia
- Hyperreflexia
- Spasticity of the lower limbs
- Clonus
- Autonomic manifestations
- Multifocal discharges seen on EEG
- Migrating focal discharges from one cortical region to another seen on EEG
- Status epilepticus
- Delayed myelination seen on MRI
- Cortical atrophy
- Thin corpus callosum
- Neuronal loss in the hippocampus
- Reactive gliosis
- De novo mutations often seen (mutation not found in either parent)
- Onset of seizures in first 6 months of life
- Seizures become nearly continuous
- Normal development until onset of seizures
- Seizures are refractory to treatment
Nocturnal Frontal Lobe Epilepsy Type 5 (NFLE5)

- Seizures, focal, partial, motor
- Vocalizations
- Dystonic posturing
- Hypermotor automatisms
- Nocturnal occurrence
- Seizures occur in clusters
- Onset in childhood (mean 6 years)
- Seizures may be refractory
- Usually passed down from one parent in an autosomal dominant fashion

Who can I contact for more information about this type of epilepsy?

We do not know of any KCNT1 epilepsy groups. If you know any groups that can be added to this list, please email EGI@CUREepilepsy.org.

References


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