Some information about epilepsy caused by mutations in the KCNQ2 gene:

KCNQ2 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, Voltage-Gated, KQT-Like Subfamily, Member 2.

What medical conditions are associated with mutations in this gene?

- Early Infantile Epileptic Encephalopathy Type 7 (EIEE7)
- Benign Familial Neonatal Seizures Type 1 (BFNS1)

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

**Early infantile epileptic encephalopathy-7 (EIEE7)**

- Seizures, tonic
- Seizures, clonic
- Generalized stiffening
- Automatisms (repetitive actions performed unconsciously)
- Delayed development
- Intellectual disability
- Hypotonia (low muscle tone)
- Dystonia (abnormal muscle tone)
- Spastic quadriplegia
- EEG shows burst suppression pattern
- EEG shows multifocal epileptic activity
- Hyperintensities in the basal ganglia and/or thalamus on MRI
- Thin corpus callosum (in some patients)
- Reduced posterior white matter volume (in some patients)
- Onset of seizures in infancy
- Multiple seizures daily at onset
- Seizure frequency decreases during early childhood
- Most patients become seizure-free by age 3 or 4 years
- Variable severity of seizures seen in family members and can be inherited
- Mutations may also occur de novo (not see in either parent)
- Seizures are often unresponsive to treatment

**Benign familial neonatal seizures-1 (BFNS1)**

- Seizures, afebrile (without fever)
- Focal clonic seizures
• Generalized tonic-clonic seizures
• Seizures start with tonic posturing
• Motor automatisms
• Febrile seizures may occur
• Increased risk of seizures in childhood or adulthood (11-16%)
• Normal development
• Finger twitching
• EMG with spontaneous discharge of normal motor unit potentials
• Onset of seizures at 2-8 days of life
• Most remit by 6 weeks (1-6 months)
• Variable severity of seizures seen
• Some patients may have isolated quivering of the muscles
• Runs in families and is inherited in an autosomal dominant manner

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

Please contact these groups for more information about KCNQ2 epilepsy (listed in alphabetical order):

• The Jack Pribaz Foundation
• KCNQ2 CURE

If you know any groups that can be added to this list, please email EGI@CUREepilepsy.org.

References


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