Some information about epilepsy caused by mutations in the KCNQ3 gene:

KCNQ3 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 3.

What medical conditions are associated with mutations in this gene?

- Benign Familial Neonatal Seizures Type 2 (BFNS2)

What are the some of the features of BFNS2?

- Seizures, afebrile (without fever)
- Focal clonic seizures
- Generalized tonic-clonic seizures
- Increased risk of seizures in childhood or adulthood (11-16%)
- Normal development
- Onset of seizures at 2-8 days of life
- Most seizures remit by 2 months
- Usually runs in families and is inherited in an autosomal dominant fashion

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

We do not know of any KCNQ3 epilepsy groups. If you know any groups that can be added to this list, please email EGI@CUREepilepsy.org.

References


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