SCN2A GENE

Some information about epilepsy caused by mutations in the SCN2A gene:

SCN2A is a sodium channel gene. Sodium channels are proteins in cells that allow sodium to pass to the inside. Sodium channels play a key role in a cell’s ability to generate and transmit electrical signals. The formal name of this gene is: Sodium Channel, Voltage-Gated, Type 2, Alpha Subunit.

What medical conditions are associated with mutations in this gene?

- Early Infantile Epileptic Encephalopathy Type 11 (EIEE11)
- Benign Familial Infantile Seizure’s Type 3 (BFIS3)

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

**Early Infantile Epileptic Encephalopathy Type 11 (EIEE11)**

- Seizures, tonic-clonic
- Delayed development
- Febrile (with fever) seizures may occur
- Status epilepticus
- Spastic quadriplegia
- EEG shows abundant slow waves and fast spike activity
- EEG shows suppression with burst activities during seizures
- Onset in infancy
- Seizures are resistant to treatment and there is variable seizure severity
- Mutations often occur de novo (not found in either parent)

**Benign familial infantile seizures Type 3 (BFIS3)**

- Seizures, partial, afebrile (not associated with fever)
- Secondary generalized tonic-clonic seizures may occur
- Seizures occur in clusters over 1 or several days
- Seizures often begin focally with head and eye deviation to one side
- Rigidity and staring episodes during seizures
- EEG during a seizures shows focal onset, often in the posterior region of brain
- Normal development and normal EEG between seizures
- Onset ranges from 2 days to 7 months (most at 2-3 months)
- Seizures are easily controlled by medications
- Spontaneous resolution by 12 months of age with no recurrence later in life
- Runs in families, passed down by one parent in an autosomal dominant manner
Who can I contact for more information about SCN2A epilepsy?

Please contact these groups for more information about SCN2A epilepsy (listed in alphabetical order):

- FAMILIEScn2a
- SCN2A Research Foundation
- Special Parent Connections

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

References


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