Some information about epilepsy caused by mutations in the SCN1A gene:

SCN1A 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 1, Alpha Subunit.

What medical conditions are associated with mutations in this gene?

- Generalized Epilepsy with Febrile Seizures Plus Type 2 (GEFS+2)
- Dravet Syndrome. This syndrome is also known as Early Infantile Epileptic Encephalopathy Type (EIEE6) or Severe Myoclonic Epilepsy of Infancy (SMEI)
- Familial Hemiplegic Migraine Type 3 (FHM3)

What are the some of the features these medical conditions?

*Generalized Epilepsy with Febrile Seizures Plus Type 2 (GESF+2)*

- Seizures, generalized, associated with fever (febrile seizures)
- Febrile seizures
- Generalized tonic-clonic seizures
- Absence seizures
- Myoclonic seizures
- Hemiclonic seizures
- Partial seizures
- Patients show normal development
- Onset of febrile seizures typically between 6 months and 6 years of age
- Simple febrile seizures usually remit by age 6 years
- Runs in families (passed down from one parent usually in an autosomal dominant manner)
- Variable severity of epilepsy seen in families
- Some patients have a more severe phenotype and have febrile and afebrile seizures after childhood (GEFS+)
- Seizures recur in 33% of patients
- Between 2 and 7% of children will develop afebrile seizure disorders later in life
Dravet Syndrome

- Generalized clonic or tonic-clonic seizures
- One sided or unilateral clonic seizures
- Absence seizures
- Complex partial seizures
- Myoclonic seizures
- Delayed development
- Developmental delay after second year
- Mental deterioration
- Ataxia
- Status epilepticus
- EEG may be normal at first
- EEG later shows generalized spike or polyspike waves and focal spikes
- EEG may show migrating focal or multifocal origin (in severe cases)
- Cerebral degeneration (in severe cases)
- Most mutations occur de novo (not found in either parent)
- Onset of seizures in the first year of life
- Developmental delay may already be apparent at onset of seizures
- May be induced by fever or hot bath
- Often not treatable with medical therapy
- May be found in families with generalized epilepsy with febrile seizures plus (GEFS+)
- Mutations are often de novo (not found in either parent)

Familial Hemiplegic Migraine 3 (FHM3)

- Migraine
- Migraine with aura
- Hemiparesis
- Hemiplegia
- Seizure (in a subset of patients in infancy)
- Onset in first 2 decades (range 6 to 15 years)
- Variable frequency and duration of migraines and episodes
- Blindness episodes are not associated with FHM
- Runs in families

Who can I contact for more information about SCN1A epilepsy?

Please contact these groups for more information about SCN1A epilepsy (listed in alphabetical order):

- Dravet Syndrome Foundation

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

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