KCNB1 encephalopathy

Description

KCNB1 encephalopathy is a condition characterized by abnormal brain function (encephalopathy), recurrent seizures (epilepsy), and developmental delay.

Most people who have *KCNB1* encephalopathy have more than one type of seizure. The seizure types that can occur in people with this condition include uncontrolled muscle twitches (myoclonic seizures), uncontrolled muscle stiffness (tonic seizures), loss of consciousness with muscle rigidity and convulsions (tonic-clonic seizures), sudden episodes of weak muscle tone (atonic seizures), sudden falls (drop attacks), or partial or complete loss of consciousness (absence seizures).

Some individuals with *KCNB1* encephalopathy do not develop seizures, but they do have an abnormal pattern of electrical activity in the brain called continuous spike and waves during slow-wave sleep (CSWS). This pattern occurs during sleep, specifically during deep (slow-wave) sleep.

Children with *KCNB1* encephalopathy have delayed development of speech and motor skills, such as sitting, crawling, and walking. Weak muscle tone (hypotonia) in some affected individuals can contribute to this delay. Many children with the condition eventually walk independently, but some individuals require assistance. Some affected individuals can communicate verbally using simple sentences, while others never develop the skill.

About half of individuals with *KCNB1* encephalopathy also have neurodevelopmental disorders, including attention-deficit/hyperactivity disorder (ADHD) and autism spectrum disorder (ASD). In *KCNB1* encephalopathy, problems with vision, digestion, and sleep can rarely occur.

Frequency

The prevalence of *KCNB1* encephalopathyis unknown. More than 65 cases have been reported in the scientific literature.

Causes

As its name indicates, *KCNB1* encephalopathy is caused by mutations in the *KCNB1* gene. The *KCNB1* gene provides instructions for making one part of a potassium

channel called Kv2.1. Potassium channels transport positively charged atoms (ions) of potassium in and out of cells. This activity plays a key role in a cell's ability to generate and transmit electrical signals. Kv2.1 channels are found primarily in nerve cells (neurons) in the brain where they are involved in regulating activity of neurons and sending electrical signals in the brain.

Most *KCNB1* gene mutations that cause *KCNB1* encephalopathy lead to an altered protein that results in impaired Kv2.1 channel function. As a result, the channels cannot regulate the flow of potassium ions in neurons, which disrupts normal communication between these cells. Impaired channel function disrupts normal brain development and leads to seizures, intellectual disability, and other features of encephalopathy that occur in this condition.

Learn more about the gene associated with KCNB1 encephalopathy

- KCNB1

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most cases result from a new mutation in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- Early infantile epileptic encephalopathy 26
- EIEE26
- Epileptic encephalopathy, early infantile, 26
- KCNB1-related epilepsy

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Developmental and epileptic encephalopathy, 26 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4015119/)

Genetic and Rare Diseases Information Center

- Developmental and epileptic encephalopathy 26 (https://rarediseases.info.nih.gov/diseases/12391/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 26; DEE26 (https://omim.org/entry/616056)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=KCNB1+AND+encephalopathy)

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