

CHD2 myoclonic encephalopathy

Description

CHD2 myoclonic encephalopathy is a condition characterized by recurrent seizures (epilepsy), abnormal brain function (encephalopathy), and intellectual disability. Epilepsy begins in childhood, typically between ages 6 months and 4 years. Each individual may experience a variety of seizure types. The most common are myoclonic seizures, which involve involuntary muscle twitches. Other seizure types include sudden episodes of weak muscle tone (atonic seizures); partial or complete loss of consciousness (absence seizures); seizures brought on by high body temperature (febrile seizure); or tonic-clonic seizures, which involve loss of consciousness, muscle rigidity, and convulsions. Some people with *CHD2* myoclonic encephalopathy have photosensitive epilepsy, in which seizures are triggered by flashing lights. Some people with *CHD2* myoclonic encephalopathy experience a type of seizure called atonic-myoclonic-absence seizure, which begins with a drop of the head, followed by loss of consciousness, then rigid movements of the arms. Epilepsy can worsen, causing prolonged episodes of seizure activity that last several minutes, known as status epilepticus. The seizures associated with *CHD2* myoclonic encephalopathy are called refractory because they usually do not respond to therapy with anti-epileptic medications.

Other signs and symptoms of *CHD2* myoclonic encephalopathy include intellectual disability that ranges from mild to severe and delayed development of speech. Rarely, individuals can have a loss of acquired skills (developmental regression) following the onset of epilepsy. Some people with *CHD2* myoclonic encephalopathy have autism spectrum disorders, which are conditions characterized by impaired communication and social interaction. In some instances, areas with a loss of brain tissue (atrophy) have been found with medical imaging.

Frequency

The prevalence of *CHD2* myoclonic encephalopathy is unknown; at least 32 cases have been described in the scientific literature.

Causes

As its name suggests, *CHD2* myoclonic encephalopathy is caused by mutations in the *CHD2* gene. This gene provides instructions for making a protein called chromodomain DNA helicase protein 2. This protein is found in cells throughout the body and regulates

gene activity (expression) through a process known as chromatin remodeling. Chromatin is the complex of DNA and proteins that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. The role of chromodomain DNA helicase protein 2 in the brain is unknown. Researchers suspect that the protein may be involved in regulating the development and functioning of nerve cells.

CHD2 gene mutations either prevent the production of any chromodomain DNA helicase protein 2 or lead to the production of a nonfunctional version of the protein. As a result, chromatin remodeling and gene expression normally regulated by chromodomain DNA helicase protein 2 are disrupted. It is unclear why *CHD2* gene mutations seem to only affect nerve cells in the brain or how they lead to the signs and symptoms of *CHD2* myoclonic encephalopathy.

[Learn more about the gene associated with CHD2 myoclonic encephalopathy](#)

- CHD2

Inheritance

This condition is considered autosomal dominant, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

This condition results from new (de novo) mutations in the gene that occur during the formation of reproductive cells (eggs or sperm) or in early embryonic development. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- CHD2 encephalopathy
- CHD2-related neurodevelopmental disorders

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Developmental and epileptic encephalopathy 94 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3809278/>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22CHD2 myoclonic encephalopathy%22>)

Catalog of Genes and Diseases from OMIM

- DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 94; DEE94 (<https://omim.org/entry/615369>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CHD2%5BTIAB%5D%29+AND+%28epilepsy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

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