Pyridoxine-dependent epilepsy

Description

Pyridoxine-dependent epilepsy is a condition that involves seizures beginning in infancy or, in some cases, before birth. Those affected typically experience prolonged seizures lasting several minutes (status epilepticus). These seizures involve muscle rigidity, convulsions, and loss of consciousness (tonic-clonic seizures). Additional features of pyridoxine-dependent epilepsy include low body temperature (hypothermia), poor muscle tone (dystonia) soon after birth, and irritability before a seizure episode. In rare instances, children with this condition do not have seizures until they are 1 to 3 years old.

Anticonvulsant drugs, which are usually given to control seizures, are ineffective in people with pyridoxine-dependent epilepsy. Instead, people with this type of seizure are medically treated with large daily doses of pyridoxine (a type of vitamin B6 found in food) . If left untreated, people with this condition can develop severe brain dysfunction (encephalopathy). Even though seizures can be controlled with pyridoxine, neurological problems such as developmental delay and learning disorders may still occur.

Frequency

Pyridoxine-dependent epilepsy occurs in 1 in 100,000 to 700,000 individuals. At least 100 cases have been reported worldwide.

Causes

Mutations in the *ALDH7A1* gene cause pyridoxine-dependent epilepsy. The *ALDH7A1* gene provides instructions for making an enzyme called α-aminoadipic semialdehyde (α-AASA) dehydrogenase, also known as antiquitin. This enzyme is involved in the breakdown of the protein building block (amino acid) lysine in the brain.

When antiquitin is deficient, a molecule that interferes with vitamin B6 function builds up in various tissues. Pyridoxine plays a role in many processes in the body, such as the breakdown of amino acids and the productions of chemicals that transmit signals in the brain (neurotransmitters). It is unclear how a lack of pyridoxine causes the seizures that are characteristic of this condition.

Some individuals with pyridoxine-dependent epilepsy do not have identified mutations in the *ALDH7A1* gene. In these cases, the cause of the condition is unknown.

Learn more about the gene associated with Pyridoxine-dependent epilepsy

- ALDH7A1

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- AASA dehydrogenase deficiency
- EPD
- Epilepsy, pyridoxine-dependent
- PDE
- Pyridoxine dependency
- Pyridoxine dependency with seizures
- Pyridoxine-dependent seizures
- Vitamin B6-dependent seizures

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Pyridoxine-dependent epilepsy (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849508/)

Genetic and Rare Diseases Information Center

- Pyridoxine-dependent epilepsy (https://rarediseases.info.nih.gov/diseases/9298/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Pyridoxine-dependent epilepsy%22)

Catalog of Genes and Diseases from OMIM

- EPILEPSY, EARLY-ONSET, 4, VITAMIN B6-DEPENDENT; EPEO4 (https://omim.org/entry/266100)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28pyridoxine-dependent+epilepsy%5BTIAB%5D%29+OR+%28pyridoxine-dependent+seizures%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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