Autosomal dominant epilepsy with auditory features

Description

Autosomal dominant epilepsy with auditory features (ADEAF) is an uncommon form of epilepsy that runs in families. People with this condition typically hear sounds (auditory features), such as buzzing, humming, or ringing, during seizures. Some people hear more complex sounds, like specific voices or music, or changes in the volume of sounds. Some people with ADEAF suddenly become unable to understand language before losing consciousness during a seizure. This inability to understand speech is known as receptive aphasia. Less commonly, seizures may cause visual hallucinations, a disturbance in the sense of smell, a feeling of dizziness or spinning (vertigo), or other symptoms that affect the senses.

ADEAF is called a focal epilepsy because the seizures start in one part of the brain, rather than involving the entire brain from the beginning. Most people with ADEAF have focal aware seizures, which do not cause a loss of consciousness. These seizures are thought to begin in a part of the brain called the lateral temporal lobe. In some people, seizure activity may spread from the lateral temporal lobe to affect other regions of the brain. If seizure activity spreads to the entire brain, it causes a loss of consciousness, muscle stiffening, and rhythmic jerking. Episodes that begin as focal seizures and spread throughout the brain are known as secondarily generalized seizures.

Seizures associated with ADEAF usually begin in adolescence or young adulthood. They may be triggered by specific sounds, such as a ringing telephone or speech, but in most cases the seizures do not have any recognized triggers. In most affected people, seizures are infrequent and effectively controlled with medication.

Frequency

ADEAF appears to be uncommon, although its prevalence is unknown.

Causes

Variants (also called mutations) in the *LGI1* gene or *RELN* gene are the most common cause of ADEAF. Variants in other genes have been identified in small numbers of affected families, and the genetic cause in many families is unknown.

The *LGI1* gene provides instructions for making a protein called Lgi1 or epitempin, which is found primarily in nerve cells (neurons) in the brain. Although the precise role

of epitempin remains uncertain, researchers suggest it may play a role at the junction of neurons (synapses) where cell-to-cell communication takes place.

The *RELN* gene provides instructions for making a protein called reelin. Reelin is produced in the brain both during brain development and after birth. It appears to play several roles at synapses, including controlling communication between neurons.

Variants in the *LGI1* or *RELN* gene may impair the formation or function of synapses. Abnormal communication between neurons can lead to seizure activity in the brain.

Learn more about the genes associated with Autosomal dominant epilepsy with auditory features

- DEPDC5
- LGI1
- RELN

Additional Information from NCBI Gene:

- MICAL1

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to raise the risk of developing epilepsy. About two-thirds of people who inherit an *LGI* gene variant will develop seizures. Some people who have the altered gene never develop the condition, which is known as reduced penetrance. The likelihood of developing seizures in people with a variant in *RELN* or other associated genes is unknown. Most people with ADEAF have one affected parent and other relatives with the condition.

Other Names for This Condition

- ADEAF
- ADLTE
- ADPEAF
- Autosomal dominant lateral temporal lobe epilepsy
- Autosomal dominant partial epilepsy with auditory features
- Epilepsy, partial, with auditory features
- ETL1

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Epilepsy, familial temporal lobe, 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551957/)

Genetic and Rare Diseases Information Center

- Autosomal dominant epilepsy with auditory features (https://rarediseases.info.nih.gov/diseases/2257/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- EPILEPSY, FAMILIAL TEMPORAL LOBE, 1; ETL1 (https://omim.org/entry/600512)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28autosomal+dominant+AND+partial+epilepsy+AND+auditory+features%5BTIAB%5D%29+OR+%28adlte%5BTIAB%5D%29+OR+%28adpeaf%5BTIAB%5D%29+OR+%28autosomal+dominant+lateral+temporal+lobe+epilepsy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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