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Autosomal Dominant Partial Epilepsy with Auditory Features

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Perspective

Autosomal Dominant Partial Epilepsy with Auditory Features (ADPEAF) is an uncommon form of epilepsy that runs in families. This disorder causes seizures usually characterized by sound-related (auditory) symptoms such as humming, buzzing, or ringing. Some people experience more complex sounds during a seizure, such as specific voices or music, or changes in the volume of sounds. Some people with ADPEAF suddenly become unable to understand language before losing consciousness during a seizure. A rare, genetic, familial partial epilepsy disease characterized by focal seizures associated with prominent ictal auditory symptoms, receptive aphasia, presenting in two or more family members.

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 affect the entire brain, it causes a loss of consciousness, rhythmic jerking and muscle stiffening.

Seizures associated with ADPEAF 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.

Autosomal Dominant Partial Epilepsy with Auditory Features (ADPEAF) is a rare familial partial epilepsy syndrome with onset in the second or third decades of life characterized by recurrent auditory auras. Mutations in the leucine-rich, glioma inactivated 1 gene (*LGI1*) on chromosome 10q have been identified in approximately 50% of families with ADPEAF.

The disease heredity is autosomal dominant with varying penetration (about 70%). The diagnosis is based on personal and family history, normal MRI brain scan. Approximately 33% of patients show a pathogenic variant in the *LGI1* gene. In a smaller percentage of ADLTE cases, mutation in the reelin (*RELN*) gene is shown in heterozygous form. The *RELN* gene is primarily expressed in brain tissue. The protein product of the *RELN* gene is called reelin. Reelin regulates the correct formation of laminated structures during embryonic development and postnatally modulates dendritic growth and synaptic plasticity.

Among the various diagnostic techniques it is important to obtain an electroencephalogram and long-latency auditory evoked potentials. The prognosis is good and the treatment is based on carbamazepine, valproate and phenytoin.

Diagnosis is based on the presence of characteristic clinical manifestations, a family history suggesting autosomal dominant inheritance, and normal brain imaging studies. Epileptiform interictal EEG abnormalities can be present in up to 2/3 of cases. Molecular genetic testing identifying a causative mutation, confirms diagnosis.

Prenatal diagnosis is possible if a pathogenic variant has been previously identified in a family member. Variable penetrance is observed. ADEAF is treated with antiepileptic drugs that are routinely used in clinical practice, with seizure control attained in the majority of cases. Evaluation of relatives is important in order to identify those who may be at risk and might benefit from early treatment initiation and/or measures to reduce risk in the event of seizure onset.

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