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Autosomal Epilepsy Syndrome with Auditory Features

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Abstract

A rare type of epilepsy that occurs in families is known as autosomal dominant partial epilepsy with auditory characteristics (ADPEAF). This condition results in seizures that are typically accompanied by auditory symptoms associated to sound, such as humming, buzzing, or ringing. During a seizure, some people hear more intricate sounds, such as particular voices or music, or variations in sound loudness. Some ADPEAF patients suddenly lose the ability to comprehend words before passing out during a seizure. An uncommon, genetic, familial partial epilepsy disorder that manifests in two or more family members and is characterised by focal seizures along with noticeable ictal auditory symptoms and receptive aphasia. The lateral temporal lobe, a region of the brain, is thought to be where seizures start. Seizure activity may expand from the lateral temporal lobe in some individuals.

Keywords: Seizures • Aphasia • Epileptiform interictal

Introduction

The first ADPEAF-related seizures appear in youth or early adulthood. While certain sounds, such a ringing phone or voice, may cause them to occur, this is not always the case with seizures. Most affected individuals have infrequent seizures that can be easily controlled with medication. A rare family partial epilepsy disease with recurring auditory auras, known as autosomal dominant partial epilepsy with auditory features (ADPEAF), develops in the second or third decades of life. In over 50% of families with ADPEAF, mutations in the leucine-rich, glioma inactivated 1 gene (LGI1) on chromosome 10q have been found. The prevalence of the disease's autosomal dominant inheritance varies (about 70%). The diagnosis is supported by a normal MRI brain scan and personal and family history [1].

Description

A long-latency auditory evoked potential and an electroencephalogram should be obtained among the other diagnostic methods. The prognosis is good andcarbamazepine, valproate andphenytoin are the mainstays of the therapy. The presence of distinctive clinical manifestations, a family history that suggests autosomal dominant inheritance andnormal brain imaging investigations all contribute to the diagnosis. In up to two thirds of instances, epileptiform interictal EEG abnormalities might be found. A causal mutation is found through molecular genetic testing, which validates the diagnosis. If a pathogenic variation has been previously detected in a family member, prenatal diagnosis may be achievable. Penetrance variation is seen. Antiepileptic medications are used to treat ADEAF on a regular basis in clinical practise andmost patients experience seizure control as a result.

The seizures that characterise ADNFLE typically happen in clusters and last anywhere from a few seconds to a few minutes. Some people just wake up from their sleep-induced minor seizures. Others experience more severe episodes that may involve abrupt, repetitive movements like arm flinging or tossing motions and leg cycling motions. The individual might get out of bed and start to move about, which is similar to sleepwalking. Additionally, the individual may scream or make sounds like groaning, gasping, or grunting. There are times when these events are incorrectly classified as nightmares, night terrors, or panic attacks.

A serine to phenylalanine transition at position 248 (S248F), which is found in the second transmembrane spanning region of the gene producing a nicotinic acetylcholine receptor 4 subunit, is the first mutation connected to ADNFLE. This mutation is known as S280F using the numbering system based on the human CHRNA4 protein. Although this mutant component is present in working receptors, it causes desensitisation to occur considerably more quickly than in receptors lacking it. Additionally, compared to receptors that solely include the wild-type, these mutant-containing receptors recover from desensitisation significantly more slowly.These mutant receptors also exhibit a reduced acetylcholine affinity and single channel conductance compared to wild-type receptors. Additionally significant, this mutation in CHRNA4 results in receptors that are less responsive to calcium.

Frequent short hypermotor seizures during sleep are a common feature of seizures in persons with SHE. An average of 8 seizures per person per night. The appearance of the seizures can be mistaken for a simple awakening from sleep or even a nightmare or night terror. Additionally, they may involve more complex motions like turning, turning around, pushing the pelvis, pedalling, grimacing, roaming, startling andvocalisations like yelling, groaning, or crying. While suffering uncontrollable arm and leg movements during a seizure, a person may occasionally be fully conscious. Some people feel as though they are unable to breathe when having a seizure. These convulsions typically last around 30 seconds but can last anywhere from a few seconds to several minutes. Finding the genetic and molecular causes of several epilepsies has made similar strides. In a number of inherited illnesses with escalating symptomatic epileptic episodes, direct molecular diagnosis is now feasible. Linkage analysis has been successful mostly in locating many mutations in uncommon monogenic epilepsy disorders. These findings only apply to a small number of families, but they have enormous potential because the found genes are linked to a variety of more complex common epileptic illnesses that are likely influenced by variance in numerous susceptibility genes [2-4].

Neurotransmitters, which are substances that are released from one neuron and absorbed by nearby neurons, are what allow neurons to communicate. The proper release and uptake of several neurotransmitters in the brain are thought to be affected by mutations in the CHRNA2, CHRNA4

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andCHRNB2 genes, according to research. The aberrant brain activity linked to seizures is most likely brought on by the resultant alterations in neuronal signalling [5].

Conclusion

The frontal lobes of the brain are where the seizures linked to ADNFLE start. The reasoning, planning, judgement andproblem-solving processes all take place in these parts of the brain. The reason why seizures in the frontal lobes rather than other parts of the brain are brought on by mutations in the CHRNA2, CHRNA4 andCHRNB2 genes.

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Conflict of Interest

There are no conflicts of interest by author.

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