

## Episodic ataxia

### Description

Episodic ataxia is a group of related conditions that affect the nervous system and cause problems with movement and coordination. People with episodic ataxia have episodes of poor coordination and balance (ataxia). During these episodes, many people also experience dizziness (vertigo), nausea and vomiting, migraines, blurred or double vision, slurred speech, and ringing in the ears (tinnitus). Seizures, muscle weakness, and paralysis that affect one side of the body (hemiplegia) may also occur during these episodes.

Additionally, a muscle abnormality called myokymia or an eye abnormality called nystagmus can occur during or between episodes. Myokymia causes muscle cramping; stiffness; or continuous, fine muscle twitching that appears as rippling under the skin. Nystagmus refers to rapid, involuntary eye movements.

Episodes of ataxia and other symptoms can begin anytime from early childhood to adulthood. They can be triggered by environmental factors such as stress, caffeine, alcohol, certain medications, physical activity, and illness. The duration of episodes may vary from seconds to days, and the frequency ranges from several episodes per day to one or two every few months. Between episodes, affected individuals may have no signs or symptoms. However, some continue to experience ataxia, which may worsen over time.

Some children with episodic ataxia have delayed development of speech or motor skills, such as standing and walking. They may also have learning difficulties.

Researchers have identified at least 11 types of episodic ataxia, distinguished by their pattern of signs and symptoms, age of onset, length of episodes, and genetic cause.

### Frequency

Episodic ataxia is uncommon, affecting less than 1 in 100,000 people. Only types 1, 2, and 6 have been identified in more than one family, and type 2 is by far the most common form of the condition.

### Causes

Episodic ataxia can be caused by variants (also called mutations) in several genes that

play important roles in the nervous system. Several of the genes provide instructions for making proteins that are involved in the transport of charged atoms (ions) across cell membranes. The protein produced from the *KCNA1* gene transports potassium ions, and the protein produced from the *CACNA1A* gene transports calcium ions. The movement of these ions is critical for normal signaling between nerve cells (neurons) in the brain and other parts of the nervous system. Variants in the *KCNA1* and *CACNA1A* genes cause episodic ataxia types 1 and 2, respectively.

Variants in the *SLC1A3* gene cause episodic ataxia type 6. This gene provides instructions for making a protein that transports chloride ions across cell membranes. The movement of chloride ions is thought to help maintain certain cellular conditions so the cells can survive and function. The protein also transports a brain chemical (neurotransmitter) called glutamate. Neurotransmitters allow neurons to communicate by relaying chemical signals from one neuron to another.

Researchers believe that variants in the *KCNA1*, *CACNA1A*, and *SLC1A3* genes alter the transport of ions in the brain. Changes in ion transport may cause certain neurons to become overexcited, disrupting normal communication between these cells. Although episodes of ataxia are caused by changes in the brain's chemical signals, it is unclear how variants in these genes cause the specific features of the disorder.

The genetic causes of other types of episodic ataxia have not been identified or are not well documented. Researchers are looking for variants in additional genes that can cause episodic ataxia.

[Learn more about the genes associated with Episodic ataxia](#)

- *CACNA1A*
- *KCNA1*
- *SLC1A3*

#### **Additional Information from NCBI Gene:**

- *FGF14*
- *SCN2A*

#### **Inheritance**

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

In some cases, an affected person inherits the variant from one affected parent. Other cases result from new variants in the gene and occur in people with no history of the disorder in their family.

## Other Names for This Condition

- EA

## Additional Information & Resources

### Genetic Testing Information

- Genetic Testing Registry: Episodic ataxia type 1 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1719788/>)
- Genetic Testing Registry: Episodic ataxia type 2 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1720416/>)
- Genetic Testing Registry: Episodic ataxia type 7 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2677843/>)

### Genetic and Rare Diseases Information Center

- Episodic ataxia (<https://rarediseases.info.nih.gov/diseases/9851/index>)

### Patient Support and Advocacy Resources

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

### Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Episodic ataxia%22](https://clinicaltrials.gov/search?cond=%22Episodic%20ataxia%22))

### Catalog of Genes and Diseases from OMIM

- EPISODIC ATAXIA, TYPE 2; EA2 (<https://omim.org/entry/108500>)
- EPISODIC ATAXIA, TYPE 1; EA1 (<https://omim.org/entry/160120>)
- CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-4 SUBUNIT; CACNB4 (<https://omim.org/entry/601949>)
- SOLUTE CARRIER FAMILY 1 (GLIAL HIGH AFFINITY GLUTAMATE TRANSPORTER), MEMBER 3; SLC1A3 (<https://omim.org/entry/600111>)
- EPISODIC ATAXIA, TYPE 4; EA4 (<https://omim.org/entry/606552>)
- EPISODIC ATAXIA, TYPE 3; EA3 (<https://omim.org/entry/606554>)
- EPISODIC ATAXIA, TYPE 7; EA7 (<https://omim.org/entry/611907>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Ataxia%5BMAJR%5D%29+AND+%28episodic+ataxia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

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