CONDITION

Episodic ataxia

Episodic ataxia causes repeated attacks of poor balance and coordination, often because of an inherited gene change.

WHAT IT IS
Repeated attacks of impaired balance and coordination

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Unsteadiness, poor coordination, dizziness, slurred speech

WHAT CAUSES IT
Usually inherited gene changes

HOW IT IS DIAGNOSED
Neurologic assessment and genetic testing

MAIN TREATMENT
Trigger management, rehabilitation, and attack-reducing medicines

CAN IT BE CURED?
No - inherited gene changes remain

IS IT AN EMERGENCY?
Sometimes - sudden new ataxia may be stroke

LONG-TERM OUTLOOK
Attacks often persist; some types cause progressive imbalance

Overview

Episodic ataxia is a group of uncommon neurological conditions that cause repeated attacks of poor balance and coordination. Between attacks, you may have no symptoms or some continuing unsteadiness.

Most primary forms are caused by an inherited change in a gene involved in nerve-cell signalling. These genes often control ion channels, which help electrical signals move through nerves. When they do not work normally, communication in the cerebellum-the brain region that coordinates movement-can be disrupted.

Episodic ataxia type 1, usually linked to KCNA1 , often causes brief, frequent attacks. Type 2, usually linked to CACNA1A , tends to cause attacks lasting hours and may be associated with migraine or abnormal eye movements.

Migraine, inner-ear conditions, seizures, multiple sclerosis, medicines, and reduced blood flow to the brain can cause similar episodes. Careful assessment is important, particularly when symptoms begin suddenly for the first time.

Symptoms

An attack may begin without warning or after a trigger. Symptoms, frequency, and duration vary between people and genetic types.

Symptoms may include:

People usually remain awake and aware. Episodes may last seconds or minutes in some forms and several hours in others. Less often, they continue for a day or longer.

Type 1 often causes short attacks that may happen many times a day. Fine muscle rippling or twitching, called myokymia, may continue between attacks.

Type 2 generally causes longer attacks. Nystagmus may remain between episodes, migraine is common, and some people gradually develop persistent balance or coordination problems.

Possible triggers include physical effort, emotional stress, lack of sleep, fever, alcohol, caffeine, sudden movement, or missed meals. Attacks can also occur without an obvious trigger.

- Unsteady walking or difficulty standing
- Poor arm or hand coordination
- Dizziness or a spinning sensation
- Slurred or slowed speech
- Blurred or double vision
- Involuntary eye movements called nystagmus
- Nausea, headache, or movement sensitivity
- Muscle twitching, stiffness, or cramping

Causes and risk factors

Primary episodic ataxia is usually genetic. A gene change affects how nerve cells, particularly those in the cerebellum, send electrical signals.

Many forms are autosomal dominant. One altered copy of the gene can cause the condition,

and each child of an affected parent typically has a 1 in 2 chance of inheriting it.

Someone may be the first person diagnosed in their family because of a new gene change or mild, unrecognised symptoms in relatives. Genetic testing does not identify a cause in everyone.

The best-characterised forms are:

CACNA1A changes can also be associated with familial hemiplegic migraine and other neurological features. Symptoms may differ even among relatives with the same change.

Secondary episodic ataxia is caused by another problem, such as medicines, toxins, migraine, inflammation, metabolic disorders, seizures, or a structural brain condition.

- Type 1: Usually associated with KCNA1
- Type 2: Usually associated with CACNA1A

Diagnosis

Diagnosis starts with a detailed history of when episodes began, how long they last, what happens, possible triggers, and whether relatives have similar symptoms. A phone video recorded safely during an episode may help.

A neurological examination checks eye movements, speech, strength, reflexes, sensation, coordination, and walking. It may be normal between attacks, although some people have nystagmus, twitching, or continuing imbalance.

Genetic testing may examine one gene, a panel of ataxia-related genes, or a broader part of the genetic code. A negative result does not completely rule out episodic ataxia. A "variant of uncertain significance" means there is not enough evidence to know whether a detected change causes the condition.

Other tests may include:

Genetic counselling can explain testing choices, uncertain findings, and implications for relatives.

- Brain MRI to check for stroke, inflammation, structural changes, or cerebellar shrinkage
- Blood tests for metabolic, nutritional, immune, or thyroid problems
- Heart or blood-vessel assessment if reduced brain blood flow is possible
- Hearing or balance tests if an inner-ear condition is suspected
- An electroencephalogram, or EEG, if seizures are a concern

Treatment and management

There is no treatment that removes an inherited gene change. Management aims to reduce attacks, prevent injuries, and support balance between episodes.

A symptom diary can help identify relevant triggers by recording sleep, meals, activity, stress, caffeine, alcohol, illness, and episode duration. Avoiding every possible trigger is unnecessary; focus on patterns that consistently affect you.

Acetazolamide can reduce attacks for many people with type 2 and sometimes helps other forms. Side effects can include tingling, tiredness, altered taste, kidney stones, or changes in body salts, so monitoring may be needed.

Aminopyridine medicines, including 4-aminopyridine, may reduce attacks in some people with type 2 or related forms. They require clinical supervision and are not suitable for everyone because they can cause important adverse effects, including seizures in people at risk.

Some people with type 1 improve with medicines that reduce abnormal nerve firing, including certain antiseizure medicines. Evidence is limited, and responses vary. Migraine may need separate treatment.

Physical therapy can support balance, strength, and safer walking. Occupational therapy may suggest changes for work, school, bathing, or cooking. A walking aid may help during persistent unsteadiness.

Treatment should be reviewed if attacks continue, side effects develop, or the diagnosis becomes uncertain. Do not stop prescribed medicine suddenly without medical advice.

Complications

Unexpected attacks can cause falls and injuries, particularly on stairs or while swimming, cycling, driving, or operating machinery.

Frequent episodes may disrupt education, work, travel, and social activities. Uncertainty about attacks can also contribute to anxiety or activity avoidance.

Some forms, particularly type 2, can cause gradually worsening cerebellar problems, including persistent unsteadiness, unclear speech, abnormal eye movements, or reduced hand coordination. This does not happen to everyone.

Migraine, muscle stiffness or twitching, and medicine side effects may also require treatment.

Prevention

Inherited episodic ataxia cannot currently be prevented, and it is not caused by anything a parent did before or during pregnancy.

Attacks may be reduced by managing personal triggers, taking preventive treatment consistently, getting enough sleep, and eating regular meals. Alcohol or large amounts of caffeine trigger episodes in some people.

Genetic counselling can explain inheritance and reproductive options without directing personal decisions.

To reduce falls, keep walkways clear, use handrails, sit down when warning symptoms begin, and avoid hazardous activities during an episode.

Living with it and what to expect

Type 1 often begins in childhood and causes brief, frequent attacks. Type 2 commonly begins in childhood or adolescence, although later onset is possible, and attacks usually last longer.

Attack frequency, triggers, and treatment response can change over time. Long symptom-free periods are possible but do not necessarily mean the condition has resolved.

Many people are well between episodes and continue with education, work, relationships, exercise, and family life. Schools or employers may provide rest areas, flexible breaks, remote working, or extra time for tasks.

Regular follow-up is important if symptoms change, falls become more common, or treatment needs review. Care may also include physical therapy, migraine treatment, or genetic counselling.

Driving rules vary by location. If attacks could affect vehicle control, seek advice about licensing requirements and do not drive when symptoms or warning signs are present.

When to get medical help

Arrange a routine appointment for repeated unexplained episodes of poor balance, slurred speech, abnormal eye movements, or loss of coordination, especially if relatives have similar symptoms.

If you have a diagnosis, request follow-up when attacks become more frequent, last longer, change in character, or interfere more with daily life. Increasing falls, new symptoms between attacks, or possible medicine side effects also need review.

Sudden ataxia can be caused by stroke or another urgent condition, even if it resembles a previous episode. Call emergency services for sudden new loss of balance or coordination, especially with:

Do not drive yourself during a severe or unfamiliar episode.

- Facial drooping, one-sided weakness, or numbness
- New difficulty speaking or understanding speech
- Sudden loss of vision or severe double vision
- A sudden, unusually severe headache
- Collapse, loss of consciousness, or a seizure
- Inability to stand or walk safely
- Symptoms markedly different from previous attacks

Common questions

Is episodic ataxia the same as vertigo?

No. Vertigo is a spinning sensation. It can occur during episodic ataxia, but ataxia mainly affects coordination and balance. Inner-ear conditions may cause similar symptoms.

Can tests be normal between attacks?

Yes. Neurological examination and brain imaging may be normal, particularly early on. Diagnosis uses the episode pattern, examination, family history, exclusion of other causes, and genetic testing when appropriate.

Does episodic ataxia get worse with age?

Not always. Some people continue to have episodes without major problems between them. Others, particularly some people with type 2, gradually develop persistent imbalance or coordination difficulties.

Can my children inherit it?

Many primary forms are autosomal dominant. If you have a confirmed disease-causing change, each child usually has a 1 in 2 chance of inheriting it. A genetic counsellor can explain your result.

Can I exercise with episodic ataxia?

Many people can exercise, and activity can support strength and balance. Because strenuous effort may trigger attacks, build up gradually and avoid unsafe settings near traffic, heights, or

deep water.

Is episodic ataxia a form of multiple sclerosis?

No. Primary episodic ataxia is usually an inherited nerve-signalling condition. Multiple sclerosis is an immune condition that damages the protective covering of nerves, although it can cause similar symptoms.

Sources

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