

## FREQUENTLY ASKED QUESTIONS ABOUT...

## Episodic Ataxia

### What is episodic ataxia?

Episodic ataxia is one type of ataxia among a group of inherited diseases of the central nervous system. Episodic ataxia is the result of genetic defects that lead to impairment of specific nerve fibers that carry messages to and from the brain to control movement of the body. Episodic ataxia includes:

- episodic ataxia type 1 (EA1)
- episodic ataxia type 2 (EA2)
- episodic ataxia with paroxysmal choreoathetosis and spasticity

There are now six recognized episodic ataxia syndromes, numbered 1-6. The genes are known for EA1, EA2, EA5, and EA6. The best characterized are EA 1 and EA2, the others are exceptionally rare.

Their symptoms and duration of attacks differ, but in each type the attacks occur sporadically, usually with periods of normal function in between.

Physicians may use different terms when diagnosing episodic ataxia. Some of those terms, or diagnoses, for EAI are:

- episodic ataxia with myokymia
- myokymia syndrome
- hereditary paroxysmal ataxia with neuromyotonia
- familial paroxysmal kinesigenic ataxia and continuous myokymia

For EA2:

- hereditary paroxysmal cerebellar ataxia I
- periodic vestibulocerebellar ataxia
- familial paroxysmal ataxia

- nystagmus-associated episodic ataxia For episodic ataxia with paroxysmal choreoathetosis and spasticity:
- dystonia-9
- DYT9

### What are the symptoms of episodic ataxia ?

Symptoms of episodic ataxia can vary considerably from family to family. The most common symptoms are episodes of ataxia (difficulty with balance and coordination) and unclear speech (dysarthria) interspersed with periods of normal or nearly normal neurological function. The attacks are usually brought on by exercise, excitement, rapid changes in posture or, in some cases, high- carbohydrate meals.

The attacks of EAI are usually associated with muscle twitching. Episodes are generally brief, lasting for only a few seconds or minutes. Symptoms of EAI may include incoordination and disturbed balance with involuntary movement or rippling of the muscles (myokymia) and/or muscle spasms (myotonia). There may be twitching or tremor in the face and hands. Myokymia may occur between attacks.

In EA2 the attacks last longer, ranging from 30 minutes to six hours. Symptoms often include muscle weakness, instability in the torso, and possibly dizziness and fatigue. Involuntary eye movement (nystagmus) is common between episodes. Muscle twitching generally is not a part of EA2.

Attacks of episodic ataxia with paroxysmal ►

choreoathetosis and spasticity generally last about 20 minutes and involve imbalance and incoordinated movement; stiffness or a writhing appearance (dystonia) in arms, legs, and/or toes; and a burning, tingling sensation in the legs and around the mouth.

Double vision and/or headache are possible. In some cases there may be involuntary muscle contractions and temporary paralysis in the lower body and legs persisting between episodes. In addition to stress, excitement, and exertion, attacks may be brought on by alcohol or fatigue.

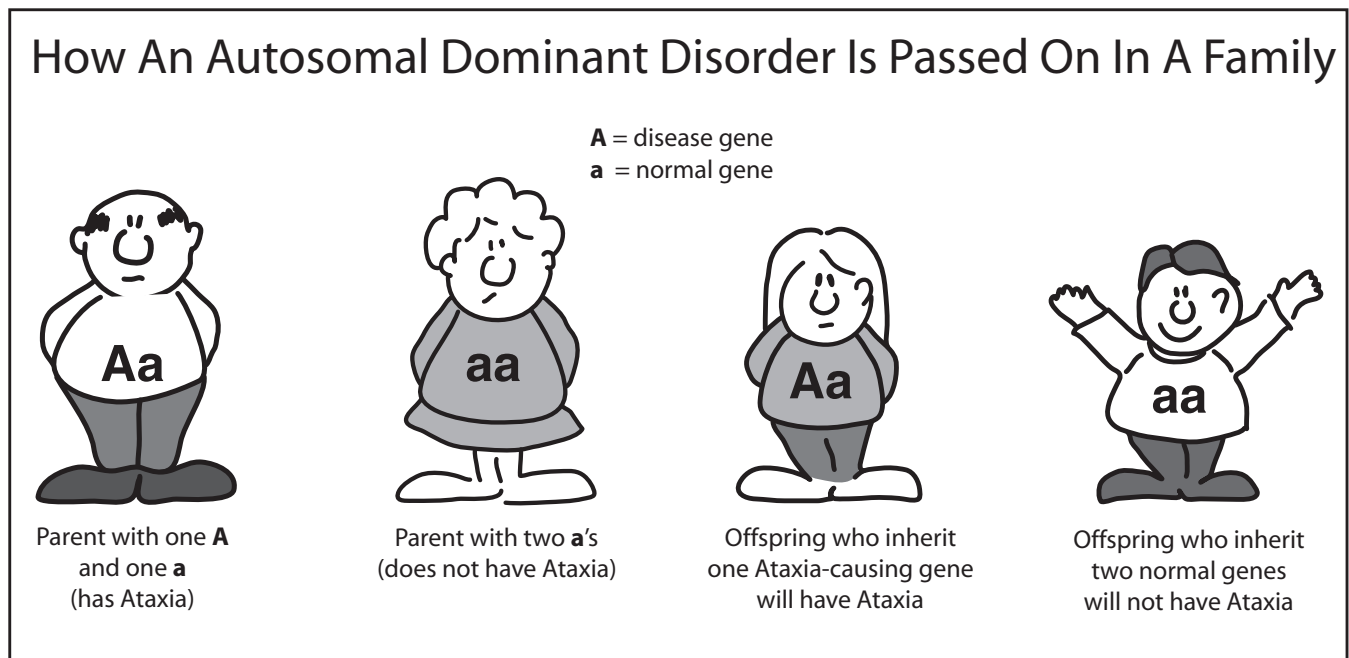
### What is the prognosis for episodic ataxia ?

Episodic ataxia most typically presents itself in the teen years, but time of onset can range from early childhood to early adulthood. Though there is no cure at this time, in some cases symptoms abate or disappear in later life, sometimes as early as young adulthood. In other cases symptoms continue into advanced years. The condition does not shorten lifespan, and symptoms often can be reduced or eliminated with medication.

### How is episodic ataxia acquired?

Episodic ataxia is a genetic disorder, which means that it is an inherited disease. The abnormal gene responsible for this disease is passed along from generation to generation by family members who carry it. Genetic diseases occur when one of the body's 50,000 genes does not work properly. (Genes are microscopic structures within the cells of our bodies that contain instructions for every feature a person inherits from his or her parents.)

Episodic ataxia is an autosomal dominant disease, which means that it is inherited from only one parent. Two copies of each gene are inherited, one copy from the mother and one from the father. Offspring who inherit one ataxia-causing gene will develop episodic ataxia. Offspring who inherit two normal genes will never develop episodic ataxia, and will pass normal genes on to their children. Each child of a parent with an autosomal dominant disease has a 50 percent chance of inheriting a defective gene and thus being afflicted with the disease. Males and females are equally likely to be afflicted.



EA1 is caused by a mutation (a mistake or variation in the gene that is significant enough to cause disease) in a potassium channel gene located on chromosome 12p. EA2 is caused by a mutation in a calcium channel gene located on chromosome 19p13. Episodic ataxia with paroxysmal choreoathetosis and spasticity is caused by a mutation in an as-yet-unknown gene located on chromosome 1p.

## How common is episodic ataxia ?

Data is not available specifically for the episodic ataxias, but have been collected on the combined number of cases of hereditary ataxias. Collectively, these forms of ataxia occur in 3 to 5 people per 100,000 in the population. Episodic ataxia is considered rare, with EA2 being reported more often than EA1.

## How is the diagnosis made?

A neurologist is often the most helpful specialist in diagnosing episodic ataxia. A thorough neurological examination can determine whether a person has symptoms typical of episodic ataxia. Besides the neurological exam, the neurologist will evaluate family history, patient history, and possibly electromyography (EMG) findings.

Mutations in the EA1 and EA2 genes are almost always found in those with early onset of discrete and recurrent attacks of ataxia. Genetic testing is available on a research basis at several laboratories around the world. There is also ongoing effort to identify defects in new genes that can cause episodic ataxia.

*Note:* A different mutation on chromosome 19 is responsible for spinocerebellar ataxia type 6 (SCA 6), and symptoms of EA2 and SCA 6 can be similar, especially in the early stages of SCA 6. Some people with EA2 develop a progressive ataxia in addition to their episodic attacks. DNA-based testing for SCA 6 is available and can accurately detect the genetic abnormality that causes SCA 6.

## What kind of support is available after Diagnosis?

Early identification of episodic ataxia can help patients and their families adapt to the condition. It can also provide opportunity for treatment of symptoms. For EA1, carbonic anhydrase inhibitors or phenytoin often can reduce or prevent attacks. For EA2 and episodic ataxia with paroxysmal choreoathetosis and spasticity, treatment with acetazolamide usually is effective.

Supportive therapy are available to help manage symptoms, and there are resources to provide emotional support. Practical information and a listing of additional resources are offered in the book, *Living With Ataxia: An Informational and Resource Guide*, published by the National Ataxia Foundation. NAF also provides and participates in many support and chat groups on the Internet. Visit our website at: [www.ataxia.org](http://www.ataxia.org) for a listing of these groups and for a more complete listing of resources affiliated with the National Ataxia Foundation.

CINCH (which NAF is a member of) is a multicenter consortium on the clinical investigation of neurological channelopathies—episodic ataxia is one of the three conditions under investigation. We aim to study the natural history of episodic ataxia and we genotype every participant to identify mutations in the known EA genes. Visit their web site: <http://rarediseasesnetwork.epi.usf.edu/cinch/index.htm>

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