National Ataxia Foundation
Connecting Ataxia families, researchers, clinicians and the community

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 may be 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 is clinically characterized by attacks of Ataxia with a clear onset of resolution.

Episodic Ataxia includes:

- Episodic Ataxia Type 1 (EA1) often associated with muscle twitching or stiffness
- Episodic Ataxia Type 2 (EA2) often associated with involuntary jerky eye movement.
- Episodic Ataxia Type 3 (EA3)
- Episodic Ataxia Type 4 (EA4) with the onset between 30 & 60 years old
- Episodic Ataxia Type 5 (EA5) with seizures
- Episodic Ataxia Type 6 (EA6) associated with seizures, hemiplegia, migraine
- Episodic Ataxia Type 7 (EA7) of adult onset
- Episodic Ataxia Type 8 (EA8) of infantile onset
- Episodic Ataxia with paroxysmal choreoathetosis and spasticity
- Episodic Ataxia of late onset after 60 years old typically with no family history, slow progression, and poor responsiveness.

There are now eight recognized episodic Ataxia syndromes, numbered 1-8, in addition to late-onset episodic Ataxia. The genes are known for EA1, EA2, ES5, and EA6. The best characterized are EA1 and EA2, the others are exceptionally rare and largely defined by single families. Episodic Ataxia is considered a rare disease.

Physicians may use different terms when diagnosing episodic Ataxia. The symptoms, duration, severity, and triggers of ataxic attacks differ, usually with periods of normal function in between.

What are the symptoms of episodic Ataxia?

Symptoms of episodic Ataxia can vary considerably from family to family and from individual to individual within the same family. The most common symptoms are episodes of Ataxia (difficulty with balance and coordination) and unclear speech (dysathria) 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 EA1 are usually associated with muscle twitching and are generally brief, lasting for only a few seconds or minutes.

Symptoms of EA1 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, stiffness or dystonia may be a feature.

Attacks of episodic Ataxia with paroxysmal choreoathetosis and spasticity generally last about 20 minutes and involve imbalance and uncoordinated movement; stiffness or a withering 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 exertions, attacks may be brought on by alcohol or fatigue.

What is the prognosis for episodic Ataxia?

Episodic Ataxia most typically presents itself 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 or heritable disease. The abnormal gene responsible for this disease is passed along from generation to generation by family members who carry it. The genetic defect may also arise spontaneously. Genetic disease occur when one of the body's 20,000 genes does not work properly.

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 copies of the gene 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 affected with the disease. Males and females are equally likely to be affected.

How is a 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 in the EA2 gene on chromosome 19 is responsible for spinocerebellar Ataxia type 6 (SCA6), and symptoms of EA2 and SCA6 can be similar, especially in the early stages of SCA6. Defects in EA2 gene may also cause familial hemiplegic migraine type 1 (FHM1).

Some people with EA2 develop a progressive Ataxia in addition to their episodic attacks. DNA testing for SCA6 is available and can accurately detect the genetic abnormality that causes SCA6.

What kind of support is available after the diagnosis of episodic Ataxia?

Early identification of episodic Ataxia can help patients and their families adapt to the condition. A physician can also provide opportunity for treatment of symptoms. For EA1, carbonic anhydrase inhibitors or phenytoin often can reduce or prevent attacks. For EA2, treatment with acetazolamide, or 4-aminopyidind anecdotally may be effective.

The National Ataxia Foundation (NAF) is committed to providing information and education about Ataxia, support groups for those affected by Ataxia, and promoting and funding research to find the cause for the various forms of Ataxia, better treatments, and, hopefully someday, a cure. NAF has been at the forefront funding promising worldwide research to find answers.

As Ataxia research moves into the clinical phase, pharmaceutical companies will begin recruiting participants for clinical trials. Individuals with Ataxia or who are at-risk for Ataxia are encouraged to enroll in the CoRDS Ataxia Patient Registry. To access the Registry, go to NAF's website www.ataxia.org and click on the "Enroll in the Patient Registry" tab and follow the directions on the CoRDS website.

NAF provides accurate information for you, your family, and your physician about Ataxia. Please visit the NAF website at www.ataxia.org for additional information, including a listing of ataxia support groups, physicians who treat Ataxia, social networks, and more. For additional questions please contact the NAF directly at www.ataxia.org or 763/553-0020.

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