

Alternating hemiplegia of childhood (AHC)

is a rare, severe neurodevelopmental syndrome characterized by recurrent hemiplegic episodes and distinct neurological manifestations. The incidence of AHC is estimated at 1 in 1 million births with disease usually ensuing within the first six months of life.

AHC was first characterized as a distinct syndrome in 1971, with a report that described patients with episodes of intermittent hemiplegia on alternating sides of the body, development delay, dystonia and choreo-athetosis.

Provoking factors, or triggers, of AHC episodes have been reported in patient studies. They include water exposure, extreme temperatures, physical activity, lighting changes, foods, and environmental stress. Falling asleep leads to disappearance of all symptoms, although they may return after waking.

In 2012, three different research groups independently revealed that mutations in the ATP1A3 gene are the primary cause of disease for patients with AHC (at least 74%).

DIAGNOSTIC CRITERIA FOR AHC:

- (1) Paroxysmal episodes of hemiplegia
- (2) Episodes of bilateral hemiplegia or quadriplegia
- (3) Other paroxysmal manifestations, such as abnormal eye movements, dystonia, nystagmus, intermittent strabismus, tonic spells, or autonomic disturbance, which can occur during hemiplegia or as isolated events
- (4) Evidence of permanent neurological dysfunction, which can manifest as intellectual deficiencies, seizures, ataxia, choreoathetosis, developmental delay, or persistent motor deficits such as spastic diplegia or quadriplegia or hypotonia
- (5) Inducing sleep during a paroxysmal attack might relieve symptoms for a period of time after awakening
- (6) First signs of dysfunction occurring before age 18 months
- (7) Gene Testing.



www.AHCKids.org

ahcf

Alternating Hemiplegia
of Childhood Foundation

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For more information about AHCF,
including resources, genetic testing, research
and general information, contact us!
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ONE MISSION: END AHC

We Fund International Projects

The AHC Foundation is the largest sponsor of scientific workshops and symposiums on an international level. We enthusiastically support multicenter collaboration projects to facilitate trials in order to identify more effective therapies for AHC.

The complexity and severity of this disorder makes it imperative that new therapeutic options be explored immediately.

RESEARCH

There are now several neurologic disorders related to ATP1A3 gene mutations. In addition to Alternating Hemiplegia of Childhood (AHC), distinct mutations in the same gene cause Rapid Onset Dystonia Parkinsonism (RDP) and further distinct amino acid changes are also known to cause cerebellar ataxia, areflexia, pes cavus, optic atrophy and sensorineural hearing loss (CAPOS) syndrome.

These three disorders underlie the exquisite genotype/phenotype relationships in neurologic disorders and suggest commonalities of disease mechanisms as well as clear differences. Such findings are sure to be replicated in many aspects of brain disorders. Through the better exchange of clinical and genetic information amongst clinicians and scientists, we should see increased awareness, more accurate diagnoses and ultimately better therapies for patients afflicted with ATP1A3 associated diseases.



AHC FOUNDATION

The foundation was founded in 1993 by parents of children with AHC. Five years later, in collaboration with top AHC researchers, the AHCF established an international registry and database to help document clinical outcomes and promote worldwide research efforts.

AWARENESS

AHC is a rare and devastating disease that is difficult to diagnose and even more challenging to treat. Part of the foundation's mission is to raise awareness of AHC so that proper diagnosis may occur as early as possible.

FUNDING

Creative fund raising is our lifeline to supporting global research grants. After winning a \$250,000 PepsiRefresh grant in 2010, interest in AHC research was revitalized and the disease causing gene mutations were found in 2012.

SUPPORT

Family meetings are coordinated by the foundation approximately every other year to provide AHC families with a unique opportunity. Parents from the United States and around the world have clinical appointments with a variety of AHC specialists, attend seminars, network with families and socialize with AHC kids.

The foundation also coordinates genetic counseling services for families at **no cost**. By connecting parents and geneticists, mutation analysis of the ATP1A3 gene in patients who meet clinical criteria for AHC, allows for definite genetic diagnosis and sound genetic counselling.

Symptoms associated with AHC have a profound impact on affected patients and families. The AHC Foundation has

One Mission: End AHC!