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Andermann syndrome

What is Andermann syndrome?

Andermann syndrome is an inherited disease characterized by severe progressive damage to the nervous system and absence or malformation of the corpus callosum in the brain. Symptoms associated with Andermann syndrome are attributed to the abnormal function of a transporter protein that is involved in moving potassium (K) and chlorine (Cl) across the cell membrane. The altered transporter protein function is believed to interfere with the development and maintenance of nerve tissue.[1]

Other names for Andermann syndrome include hereditary motor and sensory neuropathy with agenesis of the corpus callosum (HMSN/ACC); agenesis of the corpus callosum with peripheral neuropathy (ACCPN); or Charlevoix disease.[1]

What are the symptoms of Andermann syndrome and what treatment is available?

Andermann syndrome is a progressive disease that affects the nervous system. Symptoms begin to appear in infancy and may include the following:[2]

- Hypotonia (weak muscle tone)
- Absent reflexes on clinical examination
- Muscle weakness, atrophy (wasting), and tremors
- Sensory abnormalities
- Delayed walking (usually by 3-4 years)
- Loss of ambulation (ability to walk) usually by early teens
- Ptosis (drooping eyelids)
- Facial weakness
- Scoliosis (abnormal curves in the spine) usually by the age of 10 years
- Intellectual disability and psychosis
- Absence or variable development of the corpus callosum (the structure that connects the right and left sides of the brain)

Currently there is no cure for Andermann syndrome. Treatment is supportive based on symptoms and may include walking aids, such as canes or walkers, early development and educational intervention, and corrective surgery for scoliosis. Most affected individuals are eventually wheelchair-bound and die before the age of 40.[2]

How is Andermann syndrome inherited?

Andermann syndrome is an autosomal recessive disease caused by mutations in the *SLC12A6* gene.[2] An individual who inherits one copy of an *SLC12A6* gene mutation is a carrier and is usually not expected to have related health problems. An individual who inherits two *SLC12A6* mutations, one from each parent, is expected to be affected with Andermann syndrome.

If both members of a couple are carriers of a mutation in the *SLC12A6* gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Andermann syndrome?

Andermann syndrome is rare, reported most frequently in individuals of French-Canadian ancestry, but occurring in other populations as well. In certain regions in Canada it occurs in 1 in 2117 live births, with an estimated carrier frequency of 1 in 23.[2]

Having a relative who is a carrier or is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

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What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/andermann-syndrome

References

1. Andermann syndrome. *Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/condition/andermann-syndrome. Accessed March 29, 2012.
2. Dupre N, Howard H, Rouleau G. Hereditary motor and sensory neuropathy with agenesis of the corpus callosum. *GeneReviews.* Available at: http://www.ncbi.nlm.nih.gov/books/NBK1372/. Accessed March 29, 2012.

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