

What is Spinocerebellar Ataxia Type 1?

Spinocerebellar Ataxia type 1 (SCA1) is one specific type of Ataxia among a group of inherited diseases of the central nervous system. In SCA1, genetic defects lead to impairment of specific nerve fibers carrying messages to and from the brain resulting in degeneration of the cerebellum (the motor coordination center of the brain).

What are the symptoms of SCA1?

The first symptoms are usually incoordination of the hands and trouble with balance when walking. In fact, the word *Ataxia* means incoordination. As SCA1 progresses over a period of several years, difficulty swallowing and slurred speech are common. In some cases, individuals develop additional symptoms such as neuropathy (loss of feeling and reflexes in the feet or legs), spasticity, weakness, eye movement disorders or memory problems.

What is the prognosis for SCA1?

In most cases, from the onset of symptoms the duration of the disease varies from 10-30 years. The onset of symptoms in SCA1 is usually in adulthood, with average age being in the mid-30's. When the onset of symptoms is before age 20, symptoms in addition to Ataxia occur more frequently. In cases of very early onset (before the age of 13), the disease tends to be more severe and progresses much more rapidly.

How is SCA1 acquired?

SCA1 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 like SCA1 occur when one of the body's 20,000 genes does not work properly. (Genes are stretches of DNA, the genetic material, within the cells of our bodies that contain instructions for every feature a person inherits from his or her parents.) SCA1 is an autosomal dominant disease which means that individuals of either sex are as equally likely to inherit the gene and develop the disease. Each child of a person with SCA1 has a 50 percent chance of inheriting the SCA1 gene. The SCA1 gene passes directly from one generation to the next without skipping generations.

How common is SCA1?

Approximately 1-2 in 100,000 people will develop SCA1, but the frequency varies considerably based on geographical location and ethnic background.

How is the diagnosis made?

A neurologic examination can determine whether a person has symptoms typical of SCA1. A genetic test can accurately detect the presence of the abnormal gene that causes SCA1. A neurologist is often the most helpful specialist at determining the cause of symptoms that might be indicative of SCA1. It is important to rule out other diseases and to consider other forms of Ataxia.

How is the diagnosis made? (Continued)

When SCA1 is suspected, DNA-based testing is now available to confirm the diagnosis as well as to predict the severity of the disease. DNA tests involve analysis of a gene located on the 6th chromosome (each individual has 23 pairs of chromosomes). Genes are made up of substances known as nucleotides linked together in chains. Each nucleotide is identified by a letter. In SCA1, a gene mutation of the sixth chromosome results in extra copies of a series of nucleotides designated by the letter C-A-G. The more extra copies of this nucleotide series a person has, the more severe his or his disease is likely to be.

What kind of support is available after the diagnosis?

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 questions contact the NAF directly at 763/553-0020 or naf@ataxia.org.

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