

FREQUENTLY ASKED QUESTIONS ABOUT...

Spinocerebellar Ataxia Type 1 (SCA1)

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 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 indistinct 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, or memory troubles.

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 progress 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 100,000 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.)

SCA1 is an autosomal dominant disease, which means that individuals of either sex are equally likely to inherit the gene and develop the disease, and that it passes directly from one generation to the next without skipping generations. Each child of a person with SCA1 has a 50 percent chance of inheriting the SCA1 gene.

How common is SCA1?

Approximately 1-2 in 100,000 people will develop SCA1, but the ratio 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, and a blood test can accurately detect the presence of the abnormal gene that causes it. 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. When SCA1 is suspected, DNA-based testing is now available to confirm the diagnosis as well as to determine the severity of the disease.

DNA tests involve analysis of a gene located on the 12th 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 on the sixth chromosome results in extra copies of a series of nucleotides identified by the letters C-A-G. The more extra copies of this nucleotide series, the more severe the disease is likely to be.

What kind of support is available after the diagnosis?

Although there is no specific treatment to delay or halt the progression of SCA1, there is supportive therapy available to help

manage symptoms and resources to provide emotional support. *Living With Ataxia: An Information and Resource Guide* is a book published by the National Ataxia Foundation, includes a range of practical information and lists additional resources. Through the World Wide Web, NAF provides and participates in many support and chat groups. For a listing of these, see our homepage at www.ataxia.org.