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

Spinocerebellar Ataxia Type 2 (SCA2)

What is spinocerebellar Ataxia type 2?

Spinocerebellar Ataxia type 2 (SCA12) is one specific type of Ataxia among a group of inherited diseases of the central nervous system. In SCA2, 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 SCA2?

SCA2 is very similar to SCA1 and SCA3 in that the first symptom is usually Ataxia—incoordination of the hands and trouble with balance when walking. (The word ataxia means incoordination.) However, in addition to Ataxia, early symptoms of SCA2 often include neuropathy (loss of feeling and reflexes) and very slow eye movements. In some people with SCA2, muscle cramps and tremor also appear early in the disease. As SCA2 progresses over a period of several years, difficulty swallowing and indistinct speech are common. Other symptoms might include spasticity, weakness, or memory troubles. SCA2 may also cause a form of Parkinson's disease.

What is the prognosis for SCA2?

The onset of symptoms in SCA2 typically occurs when a person is in their thirties. In most cases, the duration of symptoms is from 10 to 15 years. However, when the onset of symptoms occurs before the age of 20, the disease tends to progress much more rapidly.

How is SCA2 acquired?

SCA2 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 occur when one the body's 20,000 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.)

SCA2 is an autosomal dominant disorder. This means that individuals of either sex are as equally likely to inherit the gene and develop the disease, and the gene passes directly from one generation to the next without skipping generations. Each child of a person with SCA2 has a 50 percent chance of inheriting the gene that causes it.

How common is SCA2?

SCA2 is about twice as common as SCA1 which appears in approximately 1 to 2 people in 100,000. (The ratio varies based on geographical location and ethnic background.) Overall, it accounts for about 13 percent of the autosomal dominant cerebellar ataxias.

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

How is the diagnosis made?

A neurologic examination can determine whether a person has symptoms typical of SCA2, and a genetic test can accurately detect the presence or absence of the abnormal gene that causes it. A neurologist is often the most knowledgeable specialist in determining the cause of symptoms and making the diagnosis of SCA2. It is important to rule out other causes of symptoms. When SCA2 is suspected, DNA-based testing is available to confirm the diagnosis and to help determine the severity of the disease. DNA tests involve analysis of a gene located on the 12th chromosome (each person 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 SCA2, a gene mutation on the 12th chromosome results in extra copies of a series of nucleotides identified by the letters C-A-G. The more extra copies there are of this series, the more severe the 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.

10/2008

NAF—01/2019