

FREQUENTLY ASKED QUESTIONS ABOUT...

Spinocerebellar Ataxia Type 2 (SCA2)

What is spinocerebellar ataxia type 2?

Spinocerebellar ataxia type 2 (SCA2) is one 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 his or her 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 like SCA2 occur when one of the body's 25,000 genes does not work properly. (Genes are microscopic structures within the cells of our bodies that contain instructions for features a person inherits from his or her parents.)

SCA2 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 SCA2 has a 50 percent chance of inheriting the SCA2 gene.

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.

How is the diagnosis made?

A neurologic examination can determine whether a person has symptoms typical of SCA2, and a blood test can accurately detect the presence or absence of the abnormal gene that causes it. A neurologist is often the most helpful specialist at 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?

Although there is no specific treatment to delay or halt the progression of SCA2, there is supportive therapy available to help manage symptoms, and there are resources to provide emotional support. *Living With Ataxia: An Information and Resource Guide* is a book published by the National Ataxia Foundation; it includes a range of practical information and lists additional resources. NAF also provides and participates in many support and chat groups on the Internet. Visit our Web site (see address below) for a

listing of these groups.

The National Ataxia Foundation (NAF) is committed to education about ataxia, service to individuals affected with the various forms of ataxia, and promoting and funding research to find the causes, better treatments, and a cure for ataxia. NAF can help by providing information for you, your family and your physician about ataxia. NAF does encourage you to visit our website at www.ataxia.org for additional information on ataxia, a listing of ataxia support groups, online chat groups, and more. For additional questions please contact NAF using the contact information listed below.

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