

FREQUENTLY ASKED QUESTIONS ABOUT ...

Spinocerebellar Ataxia Type 6 (SCA6)

What is spinocerebellar ataxia type 6?

Spinocerebellar ataxia type 6 (SCA6) is one type of ataxia among a group of inherited diseases of the central nervous system. It is caused by a genetic defect that inhibits the proper channeling of calcium in the body, which in turn results in degeneration of the cerebellum (the coordination center of the brain).

What are the symptoms of SCA6?

Like many other forms of ataxia, SCA6 is marked by incoordination. In fact, the word ataxia means incoordination. In most cases, the first symptoms of SCA6 are unsteady gait, stumbling, and imbalance. In about 10 percent of the cases the first symptom is unclear speech (dysarthria). As the disease progresses, incoordination of both upper and lower limbs, tremors, and dysarthria will eventually be present in everyone with SCA6. Double vision or other visual disturbances occur in about 50 percent of people with SCA6. In later stages, difficulty swallowing (dysphagia) is common.

(See note on similarities to episodic ataxia type 2 in the section discussing diagnosis.)

What is the prognosis for SCA6?

Age at onset of SCA6 symptoms ranges from 19 through 71. The severity of symptoms also varies considerably even within families. Most often, symptoms first occur from 43

through 52 years of age. Lifespan generally is not shortened by the disease.

How is SCA6 acquired?

SCA6 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 SCA6 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.)

SCA6 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 SCA6 has a 50 percent chance of inheriting the gene that causes SCA6.

How common is SCA6?

The frequency of SCA6 varies considerably according to geographical area. For instance, SCA6 represents only about 2 percent of the cases of dominant spinocerebellar ataxia in France, but it has been shown to represent as high as 31 percent in Japan. In the United States, SCA6 represents about 13 percent of all cases of dominant hereditary ataxia. Overall, the prevalence of this disease is estimated at less than 1/100,000. ►

How is the diagnosis made?

A neurologic examination can determine whether a person has symptoms typical of SCA6, and DNA-based testing can accurately detect the presence or absence of the abnormal gene that causes it. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the diseases that cause ataxia.

DNA tests for SCA6 involve analysis of a gene located on chromosome 19 (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 SCA6, a gene mutation located on chromosome 19 results in extra copies of a series of nucleotides identified by the letters C-A-G.

Note: A different mutation on chromosome 19 is responsible for episodic ataxia type 2, which means that symptoms of this disease and SCA6 may overlap. For instance, ataxia symptoms of SCA6 might be episodic (occurring in episodes lasting several hours), especially early in the disease.

What kind of support is available after the diagnosis?

Although there is no specific treatment to delay or halt the progression of SCA6, 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 contains practical information and a listing of 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.