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

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 one of the CAG repeat/ polyglutamine disorders. SCA6 is caused by a defect in a gene that makes a protein called a transcription factor, that regulates expression of other genes in the cerebellum. The genetic defect in this protein results in degenerations of primarily Purkinje cells of the cerebellum.

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 slurred speech will eventually be present in everyone with SCA6. Double vision or other visual disturbances occur in about 50 percent of the 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 to 71 years old. The severity of symptoms also varies considerably, even within families. Most often, it is a late onset disorder with symptoms first occurring between 43 and 52 years of age. It usually progresses slowly. 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 disease like SCA6 occur when one of the body's 20,000 genes does not work properly. (Gene 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 the gene 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.

What about genetic testing?

Gene tests can be performed for diagnostic purposes to determine what kind of Ataxia is within a person or family. Genetic testing also can be done, in some circumstances, even before there are symptoms to determine whether a person carries the abnormal gene or genes that cause Ataxia. This is called predictive or presymptomatic testing. A gene test also can be used to determine whether a fetus has an abnormal Ataxia gene. This is called prenatal testing. Anyone who is considering a predictive or prenatal test should consult with a genetic counselor to discuss the reasons for the test, the possible outcomes and how those outcomes might affect the person emotionally, medically, or socially.

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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 Italy, but it has been shown to represent as high as 31 percent in Japan. In the United States, SCA6 represents about 15 percent of all cases of dominant hereditary Ataxia. Overall, the prevalence of this disease is estimated at less than 1 in 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 of the abnormal gene that caused it. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that cause Ataxia.

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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