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 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 degeneration 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 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 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 through 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 diseases like SCA6 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 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 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 find out what kind of ataxia a person or family has. Gene tests 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 or not 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.

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 causes it. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the diseases that cause ataxia.

DNA testing for SCA6 involves an 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 for those affected by ataxia and their families. The National Ataxia Foundation also provides and participates in many local and on-line support groups, as well as social networks. Visit our www.ataxia.org for a listing of these groups.

What type of research is being done on SCA6? The National Ataxia Foundation funds promising world-wide research in all the forms of ataxias including spinocerebellar ataxia type 6. Because of continued ataxia research, the pathogenic mechanisms of SCA6 are becoming increasingly clear. In 2009, a multidisciplinary network involving ten institutions in the United States was established to recruit patients and obtain longitudinal clinical data from patients with SCA6 (and other SCAs) for future clinical trials and to initiate a pilot study to determine genetic modifiers of SCA 6 (and other SCAs). This Natural History Study is on-going to better understand the progression of SCA6 and have information that will be useful for future clinical research and drug trials in SCA6.

What can be done to move research in SCA6 forward?
As ataxia research moves into the clinical phase, researchers will need to recruit patients to participate in clinical trials. Individuals with SCA6 or who are at-risk for SCA6 are encouraged to enroll in the CoRDS Ataxia Patient Registry. This can be done by going to the NAF website's homepage at www.ataxia.org and clicking on the "Ataxia Patient Registry" button. This is a secure site to complete the enrollment process in the patient registry. The National Ataxia Foundation funds research studies around the world. Supporting NAF's research funding efforts is another way that research in SCA6 and all the other forms of ataxia will move us closer to treatments and a cure for SCA6.

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