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

Spinocerebellar Ataxia Type 8 (SCA8)

What is spinocerebellar Ataxia type 8?

Spinocerebellar Ataxia type 8 (SCA8) is one type of Ataxia among a group of inherited diseases of the central nervous system. As in other inherited Ataxia, SCA8 is the result of genetic defects that 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 SCA8?

Typically, balance and coordination problems (Ataxia) are noticed first. Often there is accompanying muscle spasticity, drawn-out slowness of speech, and reduced vibrations sense. As the disease progresses over a period of several years, difficulty swallowing and other symptoms are experienced. Although the disease onset is typically in adulthood, the age of onset can range from one year to more than 65 years. The progression is usually over decades regardless of the age of onset. Lifespan is typically not shortened.

How is SCA8 acquired?

SCA8 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 of the bodies 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.

Each person has 23 pairs of chromosomes with each chromosome containing two strands or chains of DNA. There are thousands of genes on each strand of DNA. Each gene is made up of substance known as nucleotides linked together in chains.

Each nucleotide is identified by a letter. The gene responsible for SCA8 is located on Chromosome 13. In SCA8, the gene mutation results in extra copies of a series of nucleotides identified by the letters C-T-G on the top strand of DNA and C-A-G on the bottom DNA strand.

SCA8 is more complex genetically than other SCAs in that it can appear to be dominant, recessive, or sporadic. A hereditary Ataxia is considered dominant if only one copy of the defective gene needs to be inherited in order to develop the disease. In a recessively inherited Ataxia, two copies of the defective gene (one from each parent) are required to develop the disease. In cases of sporadic hereditary Ataxia, there is no known family history of the disease. SCA8 acts like a dominant Ataxia in that a person needs to inherit only one copy of the defective gene in order to develop the disease. However, SCA8 is different from other dominant forms of Ataxia because the SCA8 mutation has “reduced penetrance”. This means that not everyone that inherits the C-T-G / C-A-G expansion mutation will go on to develop the disease. The reduced penetrance means that SCA8 patients may be the only member of their families to develop symptoms of the disease even though others in the family may also carry the gene.

How common is SCA8?

SCA8 is a relatively rare form of Ataxia; its occurrence is less than 1/100,000.

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How is a diagnosis made?

A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the diseases that cause Ataxia. Initially, a neurologic examination can determine whether a person has symptoms typical of one of the SCAs. DNA-based testing can determine the presence or absence of the abnormal gene that causes SCA8. However, the nature of the SCA8 gene makes the diagnosis more complex than in other SCAs. Research indicates that C-T-G/C-A-G repeat determines whether or not a person will develop Ataxia. People with fewer than 50 C-T-G/C-A-G repeats on the SCA8 gene tend not to develop the disease, while those with approximately 80 to 1300 or more C-T-G/C-A-G repeats are at risk of getting the disease.

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.

Ataxia research has moved into the clinical phase, and pharmaceutical companies have begun 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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