FREQUENTLY ASKED QUESTIONS ABOUT...

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 ataxias, 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 vibration 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 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 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.

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 substances 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.

How is the 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?

Although there is no specific treatment to delay or halt the progression of SCA8, there is supportive therapy available to help manage symptoms. Feeding evaluations should be done to reduce risk of aspiration from dysphagia. Research has shown that physical therapy is able to improve ataxia even in cerebellar degeneration.

Resources available that can provide emotional support include:

Living With Ataxia: An Information and Resource Guide, a book published by the National Ataxia Foundation, includes a range of practical information and lists additional resources. Through its website, www.ataxia.org, NAF provides the latest research and medical information about SCA8. NAF also facilitates social networks, a chat room and local support groups. Information on these groups can be accessed from the website.

Each year the National Ataxia Foundation holds an annual conference where attendees can connect with others who have ataxia and hear presentations from leading ataxia researchers and clinicians. Check NAF's website for information about this conference which is usually held in March.

What can be done to move research in SCA8 forward?

As ataxia research moves into the clinical phase, researchers will need to access patients to participate in clinical trials. Individuals with SCA8 or who are at risk for SCA8 are encouraged to enroll in a patient registry for those with ataxia. The link to the registry is located on the home page of the National Ataxia Foundation's website at www.ataxia.org. The National Ataxia Foundation funds research studies around the world. Supporting NAF's research funding efforts is another way that research in SCA8 and all the other forms of ataxia will move us closer to treatments and a cure for SCA8.

Who can I contact for more information?

If you have other questions or would like to receive more information, you may send an email to naf@ataxia.org or call 763-553-0020. You may also join NAF's e-mail list by selecting *"Sign Up for NAF EBlasts"*found on the home page of the NAF website where you will be instructed to add your contact information.

August 2012

National Ataxia Foundation
2600 Fernbrook Lane, Suite 119 • Minneapolis, MN 55447-4752
Phone: (763) 553-0020 • Fax: (763) 553-0167
Email: naf@ataxia.org • Website: www.ataxia.org