FREQUENTLY ASKED QUESTIONS ABOUT...

Spinocerebellar Ataxia Type 7 (SCA7)

What is spinocerebellar ataxia type 7?

Spinocerebellar ataxia type 7 (SCA7) has also been referred to as autosomal dominant cerebellar ataxia type 2 (ADCA2) or ataxia with pigmentary retinopathy. It is one type of ataxia among a group of inherited diseases of the central nervous system. As in many other inherited ataxias, SCA7 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 SCA7?

SCA7 differs from most other forms of spinocerebellar ataxia in that visual problems can occur in addition to poor coordination. When the disease manifests itself before age 40, visual problems rather than poor coordination are typically the earliest signs of disease, and begin as difficulty distinguishing colors and decreased central vision. These changes may progress until the person is legally blind. In addition, symptoms of ataxia (incoordination), slow eye movements, and mild changes in sensation or reflexes may be detectable. Loss of motor control, unclear speech (dysarthria), and difficulty swallowing (dysphagia) become prominent as the disease progresses. In children, failure to thrive and loss of motor milestones may be the earliest findings.

What is the prognosis for SCA7?

Initial signs of SCA7 most often appear in the late teens or early twenties, but the age of onset ranges from infancy to the fifties or sixties. The earlier the onset, the faster the disease progresses. For example, when symptoms appear in childhood, blindness can occur within a few years, while those who first show symptoms in their teens might not experience blindness until 10 years later. When symptoms first appear later in life, the disease progresses even more slowly and the degree of disability will vary accordingly.

How is SCA7 acquired?

SCA7 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 SCA7 occur when one of the body's 25,000 genes does not work properly. (Genes are submicroscopic chemical structures within the cells of our bodies that contain instructions for every feature we inherit from our parents).

SCA7 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 SCA7 has a 50 percent chance of inheriting the gene that causes it.

How common is SCA7?

SCA7 is less common than other forms of ataxia, occurring in less than one per 100,000 people. Some studies show that SCA7 represents 2% of all SCAs.

How is the diagnosis made?

A neurological examination can determine whether a person has symptoms typical of SCA7, 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, however, genetic counseling for families with the disease should be sought from a medical geneticist or genetics counselor. Retinal degeneration is the distinguishing feature of SCA7.

DNA tests for SCA7 involve analysis of a gene located on chromosome 3 (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 SCA7, a mutation in the ataxin-7 gene located on chromosome 3 results in extra copies of a series of nucleotides identified by the letters C-A-G.

What kind of support is available after the diagnosis?

Early identification of SCA7 can help individuals adapt better to changes in vision and mobility. Although there is no specific treatment to delay or halt the progression, there is supportive therapy available to help manage symptoms, and there are resources to provide emotional support.

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 SCA7 forward?

As ataxia research moves into the clinical phase, researchers will need to recruit patients to participate in clinical trials. Individuals with SCA7 or who are at risk for SCA7 are encouraged to enroll in the CoRDS Ataxia Patient Registry. This can be done by going to the NAF website, *www.Ataxia.org* and clicking on the Ataxia Patient Registry button that is on the homepage. You will be taken to a secure site to complete the enrollment process. The National Ataxia Foundation funds research studies around the world. Supporting NAF's research funding efforts is another way that research in SCA7 and all the other forms of ataxia will move us closer to treatments and a cure for SCA7.

How can the National Ataxia Foundation help?

Living With Ataxia: An Information and Resource Guide, published by the National Ataxia Foundation, includes a range of practical information and lists additional resources. NAF also provides and participates in social networks, support and chat groups on the Internet. Visit our Website (see address below) for a listing of these groups. For a more complete listing of resources and of support groups affiliated with the National Ataxia Foundation contact:

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