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 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 begins as a difficulty in distinguishing colors and decrease 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 the disease is passed along from generation to generation by family members who carry it. Genetic diseases like SCA7 occurs when one of the body’s 20,000 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 a diagnosis made?
A neurological examination can determine whether a person has symptoms typical of SCA, 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 causes Ataxia, however, genetic counseling for families with the disease should be sought from a medical geneticist or genetic counselor. Retinal degeneration is the distinguishing feature of SCA7.

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.