**What is spinocerebellar Ataxia type 5?**

Spinocerebellar Ataxia type 5 (SCA5) is a specific form of Ataxia, which belongs to a group of inherited diseases that affects the central nervous system. As with the other inherited Ataxias, SCA5 is caused by the 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).

SCA5 is sometimes called “Lincoln’s Disease” because an 11th-generation SCA5 family is descended from the paternal grandparents of President Abraham Lincoln. However, SCA5 also is sometimes referred to as “Holmes Ataxia” after Dr. Gordon Holmes, who first described the condition in 1907.

**What are the symptoms of SCA5?**

The word Ataxia is defined as a loss of coordination and SCA5 is characterized by uncoordinated gait (impaired balance when walking), limb and eye movements, and slurred speech (dysarthria).

Onset of the disease typically is marked by stumbling, difficulty climbing stairs, and losing balance while standing on one foot (such as in the shower). Ataxia of the hands and arms also is common in SCA5, although this usually is not as disabling as the gait Ataxia. Affected individuals might notice deterioration in handwriting, fastening buttons, and other activities that require finger dexterity.

Slurred speech is also common, although this is typically not severe and the articulation problems generally are not significant enough to interfere with spoken communication. While some forms of Ataxia affect multiple parts of the brain, SCA5 is considered a “pure” cerebellar Ataxia, meaning only the cerebellum which coordinates movement is typically affected.

**What is the prognosis for SCA5?**

SCA5 tends to be a mild, slowly progressive form of Ataxia that generally does not shorten life span. It typically does not have the effects on breathing, swallowing, bowel and bladder control, thinking, and strength that results from some other forms of Ataxia.

While many patients with SCA5 eventually use a mobility aid, only rarely does the disease lead to wheelchair dependence. The onset of symptoms for SCA5 can vary from ages 10 to 70, with average age at onset in the early 30s. Symptoms of SCA5 are rarely reported in individuals under the age of 20, however, these individuals may develop more severe symptoms over time.

**How is SCA5 acquired?**

SCA5 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 SCA 5 occur when one of the body’s 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.)

SCA5 is an autosomal dominant disease and means that individuals of either sex are equally likely to inherit the gene and develop the disease, and the gene passes directly from one generation to the next without skipping generations. Each child of a person with SCA5 has a 50 percent chance of inheriting the SCA5 gene.
How common is SCA5?
SCA5 is a rare form of Ataxia.

How is the diagnosis made?
A neurologist is often the most helpful specialist in diagnosing spinocerebellar Ataxia type 5. A thorough neurologic examination can determine whether a person has symptoms typical of SCA5. If symptoms are consistent with this disease, a neurologist may order genetic testing to examine the DNA to determine the presence or absence of the abnormal gene that causes SCA5. A genetic counselor should be consulted prior to the genetic test. DNA is comprised of substances called nucleotides and the test involves examining a gene on chromosome 11 to determine if there are specific changes to the nucleotides. There are currently three known changes within this gene that have been shown to cause SCA5.

What kind of support is available after the
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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