

FREQUENTLY ASKED QUESTIONS ABOUT...

Spinocerebellar Ataxia Type 5 (SCA5)

What is spinocerebellar ataxia type 5?

Spinocerebellar ataxia type 5 is one specific type of ataxia among a group of inherited diseases of the central nervous system. As in other inherited ataxias, SCA5 is caused by 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 ataxia” because a 10-generation family with the condition has ancestries that trace to the paternal grandparents of President Abraham Lincoln. SCA5 also is sometimes called “Holmes ataxia” after Dr. Gordon Holmes, who first described the condition in 1907.

What are the symptoms of SCA5?

“Ataxia” means incoordination, and, as with many other forms of ataxia, the symptoms of SCA5 are usually incoordination of the hands, arms, and legs; gait ataxia (impaired balance when walking); and dysarthria (slurred speech).

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.

Dysarthric, slurred speech also is common, although this usually is not severe and the articula-

tion problems generally are not significant enough to interfere with spoken communication.

SCA5 tends mainly to affect the cerebellum, but not other areas of the brain. It is considered a “pure cerebellar ataxia.”

What is the prognosis for SCA5?

Life span generally is not shortened by SCA5. It tends to be a mild, slowly progressive form of ataxia and does not have the effects on breathing, swallowing, bowel and bladder control, thinking, and strength that some forms of ataxia may bring. Only rarely does SCA5 lead to wheelchair dependence. The onset of symptoms for SCA5 can vary from age 10 to 70, with average age at onset in the early 30s. Symptoms of SCA5 have, on rare occasions, been reported in individuals under age 20. Those individuals with early onset of SCA5 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 SCA5 occur when one of the body’s 30,000 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, which 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. Besides the neurologic exam, the neurologist will evaluate family history, patient history, and possibly electromyography (EMG) findings.

Because the gene or genes involved in SCA5 have not been identified, genetic testing is not yet available. It has been determined, however, that the genetic abnormality responsible for SCA5 is located on chromosome 11. Research continues. There is not currently a genetic test to diagnose SCA5.

What kind of support is available after the diagnosis?

Although there is no specific treatment to delay or halt the progression of SCA5, supportive therapy is available to help manage symptoms and there are resources to provide emotional support. *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. NAF also provides and participates in many support and chat groups on the Internet. Visit our Web site for a listing of these groups.

Contact the National Ataxia Foundation for a more complete listing of resources and of support groups affiliated with the NAF.

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