National Ataxia Foundation

Spastic Paraplegia Type 7 (SPG7)

What is SPG7?

Spastic Paraplegia type 7 (SPG7) is a rare neurodegenerative disorder. It is caused by mutations in the *SPG7* gene. We don't have a good understanding of how common SPG7 is. It is estimated that, if you count all forms of hereditary spastic paraplegia, the combined prevalence is around 2 to 6 in 100,000 people worldwide. However, SPG7 would only count for a small portion of this total number.

SPG7 Symptoms

There are two forms of SPG7: pure and complex. Patients with the pure form of SPG7 only experience spasticity and muscle weakness. However, a patient with the complex form of SPG7 will develop spasticity, muscle weakness, and ataxia.

Like many other forms of Ataxia, complex SPG7 is marked by poor balance and coordination. In fact, the word Ataxia means incoordination. There can also be problems coordinating muscles that control speech, swallowing, and vision.

People with SPG7 may also develop neuropathy, which is the progressive loss of feeling in the hands and/or feet. Less common symptoms of SPG7 may include high-arched feet, curves in the spine, urinary problems, mild hearing loss, and muscle wasting.

Prognosis

SPG7 symptoms usually begin between the ages of 30 and 45 years. However, symptoms can begin in childhood or late adulthood. The onset of SPG7 symptoms varies greatly among patients. The severity of SPG7 symptoms may also vary between patients, even within families.

SPG7 is a progressive disorder. Most patients will require the use of a wheelchair, either due to poor coordination or muscle weakness. Treatments such as physiotherapy, occupational therapy, and speech-language therapy can significantly improve the lives of people with SPG7.

Genetics

SPG7 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. Men and women are equally likely to inherit the genes that cause ARSACS.

Genetic diseases like SPG7 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.

SPG7 is an autosomal recessive condition. This means that an individual only develops symptoms of the disease if both copies of their *SPG7* gene are not working properly.

An individual who has one copy of an altered or nonfunctioning *SPG7* gene does not develop any neurological symptoms and is called a carrier. For people who are carriers, the normal *SPG7* gene compensates for the nonfunctioning copy of the gene. However, a child whose parents are both carriers can inherit a "double dose" of the altered *SPG7* gene and will therefore develop SPG7.

Most of the time, carriers have no idea that they have an abnormal *SPG7* gene. This is because they do not have any symptoms or medical problems. It is often only when a child is diagnosed with SPG7 that the parents learn that they are both carriers. When both parents are carriers, each of their children has a 25 percent chance of having SPG7 and a 50 percent chance of being a carrier.

Genetics (continued)

Gene tests can be performed for diagnostic purposes to determine what kind of Ataxia is within a person or family. Genetic testing can also be done, in some circumstances, even before there are symptoms, to determine whether a person carries the abnormal gene or genes that cause Ataxia. This is called predictive or presymptomatic testing. A gene test can also be used to determine whether a fetus has an abnormal Ataxia gene. This is called prenatal testing. Anyone who is considering a predictive or prenatal test should consult with a genetic counselor to discuss the reasons for the test, the possible outcomes, and how those outcomes might affect the person emotionally, medically, or socially.

Diagnosis

A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia. A neurologic examination can determine whether a person has symptoms typical of SPG7. There are several potential follow-up tests. MRI brain imaging may be used to confirm cerebellar atrophy. Electromyography (EMG) may be used to test for neuropathy.

A definitive diagnosis of SPG7 is established following genetic testing. This confirms that someone has a mutation that causes SPG7 in their *SPG7* gene.

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. 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.