National Ataxia Foundation

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)

What is ARSACS?

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) is a rare neurodegenerative disorder. It is caused by mutations in the *SACS* gene. In the Charlevoix-Saguenay region of Québec, the incidence of ARSACS is estimated to be in 1,500 to 2,000 individuals. The ARSACS incidence rate outside of Québec is less well known. However, ARSACS is known to be more common in areas around the world with French ancestry.

ARSACS Symptoms

Like many other forms of Ataxia, ARSACS 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 ARSACS also develop neuropathy, which is the progressive loss of feeling in the hands and/or feet. Spasticity, particularly stiffness in the legs, is common. Other symptoms of ARSACS may include high-arched feet, curves in the spine, urinary problems, intellectual disability, hearing loss, and seizures.

Prognosis

ARSACS symptoms usually begin between the ages of 2 and 5 years. However, symptoms can sometimes begin in the teenage or early adult years. The severity of ARSACS symptoms may vary between patients, even within families.

ARSACS is a progressive disorder. Most patients will require the use of a wheelchair by the time they reach 30 to 40 years old. Treatments such as physiotherapy, occupational therapy, and speech-language therapy can significantly improve the lives of people with ARSACS.

Genetics

ARSACS 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 ARSACS 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.

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

An individual who has one copy of an altered or nonfunctioning *SACS* gene does not develop any neurological symptoms and is called a carrier. For people who are carriers, the normal *SACS* 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 *SACS* gene and will therefore develop ARSAC.

Most of the time, carriers have no idea that they have an abnormal *SACS* gene. This is because they do not have any symptoms or medical problems. It is often only when a child is diagnosed with ARSACS 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 ARSACS and a 50 percent chance of being a carrier.

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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 ARSACS. MRI brain imaging may be used to confirm cerebellar atrophy.

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