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Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)

What is ARSACS?

ARSACS is an inherited disease characterized by progressive muscle damage, speech and vision problems. Individuals with ARSACS have defects in sacsin, a protein that may be involved in organizing proteins within cells, especially those of the nervous system and muscles.[1] ARSACS is also known as spastic ataxia of Charlevoix-Saguenay.

What are the symptoms of ARSACS and what treatment is available?

Affected individuals appear normal at birth. Signs of ARSACS usually become apparent during the second year of life and become progressively worse. Symptoms of ARSACS may include:[1,2]

- Difficulty and unsteadiness while walking
- Spasticity (abnormally tight muscles) leading to ataxia (difficulty coordinating movements)
- Amyotrophy (muscle wasting)
- Nystagmus (involuntary side-to-side eye movements)
- Loss of sensation due to nerve damage, especially in the legs
- Deformities of the fingers and feet
- Dysarthria (problems with speech)
- Yellow streaks in the retina (the back of the eye)

There is no cure for ARSACS. Treatment is supportive and may include muscle relaxants, leg supports, physical, and occupational and speech therapies. Individuals with ARSACS will often require a wheelchair by the fifth decade of life and often do not survive past the sixth decade.[2]

How is ARSACS inherited?

ARSACS is an autosomal recessive disease caused by mutations in the *SACS* gene. An individual who inherits one copy of an *SACS* gene mutation is a carrier and is not expected to have related health problems.[2] An individual who inherits two mutations in the *SACS* gene, one from each parent, is expected to be affected with ARSACS.

If both members of a couple are carriers, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.[1]

Who is at risk for ARSACS?

ARSACS is a disorder most frequently reported in individuals of French Canadian ancestry, but occurring in other populations as well.[2] The carrier frequency of ARSACS in the French Canadian population is approximately 1 in 21.[3]

Having a relative who is a carrier or is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Ataxia of Charlevoix-Saguenay Foundation: http://www.arsacs.com/

National Ataxia Foundation: http://www.ataxia.org/

References

1. Genetics Home Reference, Autosomal recessive spastic ataxia of Charlevoix-Saguenay. Available at http://ghr.nlm.nih.gov/condition/autosomal-recessive-spastic-ataxia-of-charlevoix-saguenay. Accessed: Mar 24, 2012.
2. Robitaille Y *et al*. ARSACS. *GeneReveiws* Available at http://www.ncbi.nlm.nih.gov/books/NBK1255/. Accessed: Mar 15, 2012.
3. De Braekeleer M *et al.* Genetic epidemiology of autosomal recessive spastic ataxia of Charlevoix-Saguenay in northeasern Quebec. *Genet Epidmiol* 1993; 10(1):17-25.