Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). Carriers are not expected to show symptoms. You and your partner would both have to be carriers of ARSACS for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for ARSACS, each of your children has a 1 in 4 (25%) chance to have the condition.

ARSACS Explained

What is ARSACS?

ARSACS is a progressive inherited neurodegenerative condition that primarily affects muscle movement. Affected individuals develop gait abnormalities in the first few years of life followed by abnormal muscle contraction, impaired coordination, involuntary eye movements, muscle wasting, and speech difficulties. Other signs and symptoms may include deformities of the fingers and feet, reduced sensation and weakness in the arms and legs, and yellow streaks of fatty tissue in the back of the eye that are sensitive to light (known as hypomyelination of the retina). Rarely, some individuals may develop leaks in one of the valves that control blood flow through the heart (mitral valve prolapse). It is important to note that in individuals diagnosed with ARSACS outside of Quebec, signs and symptoms may differ, such as age of onset of symptoms or a mild intellectual disability.

Prognosis

Prognosis for ARSACS is fair. Most individuals have normal intelligence and live to become independent adults, though eventually they lose their ability to walk and require a wheelchair by the time they reach their third or fourth decade of life. Individuals with ARSACS typically live until their fifth or sixth decade of life.

Treatment

Treatment for ARSACS is symptomatic as there currently is no cure. Individuals with ARSACS can benefit from physical therapy and antispasmodic oral medications that can help postpone major functional disabilities until severe muscle weakness. Additionally, individuals with ARSACS may benefit from speech therapy and psychological support to aid in academic performance.

Resources

ARSACS Foundation
http://arsacs.com/

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/autosomal-recessive-spastic-ataxia-of-charlevoix-saguenay

National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

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