Genetics Uncoded:

Facts about ARSACS

NxGen MDx

What Your Test Results Mean

Carriers typically show no symptoms of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

ARSACS Explained

ARSACS is an inherited neurodegenerative condition in which the variants in the *SACS* gene cause the resulting protein, sacsin, to be unstable. 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. Most people affected by ARSACS 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 thirties or forties.

How the Genetics Work

ARSACS is an autosomal recessive disorder caused by variants in the *SACS* gene. In general, individuals have two copies of the *SACS* gene. Carriers of ARSACS have a single variant in one copy of the *SACS* gene while individuals with ARSACS have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions?

Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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