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Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay

National Cancer Institute

Source

National Cancer Institute. *Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay*.
NCI Thesaurus. Code C154614.

An autosomal recessive condition caused by mutation(s) in the SACS gene, encoding saccin. It is characterized by early onset cerebellar ataxia, pyramidal tract signs and peripheral neuropathy.