ARSACS is the common name for Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. This condition was first seen in people of the Charlevoix-Saguenay region of Quebec, Canada. The majority of people with ARSACS live in Quebec or have recent ancestors from Quebec. People with ARSACS have also been identified in various countries such as Japan, Turkey, Tunisia, Spain, Brazil, Poland, Italy, and Belgium. It is a progressive disease that affects the body's ability to create a protein called sacsin, normally found in the brain, skin, and muscles. Over 170 SACS mutations have been reported worldwide and are thought to cause loss of function of sacsin, a poorly characterized and massive (520 kDa) protein.

Research suggests that sacsin might play a role in folding newly produced proteins into the proper 3-dimensional shape because it shares similar regions with other proteins that perform this function. Mutations in the SACS gene cause the production of an unstable sacsin protein that does not function normally. It is still unclear how the abnormal sacsin protein affects the brain and skeletal muscles and results in the signs and symptoms of ARSACS.

In existing in vitro (patients’ cell lines) and in vivo (Sacs(-/-) mouse) models, a disruption of mitochondrial transport is observed along with abnormal accumulation of non-phosphorylated neurofilament (NF) bundles in the somatodendritic regions of vulnerable neuronal populations, a significant reduction in mitochondrial motility and elongated mitochondria. The data points to alterations in the NF cytoskeleton and defects in mitochondrial dynamics as the underlying pathophysiological basis of ARSACS.

There are currently no treatments available to cure people with ARSACS.

The Ataxia Charlevoix-Saguenay Foundation, funded in 2006, is a charitable organization federally registered with no employees and is supported entirely by private donations and volunteers to fund scientific research on ARSACS. The Ataxia Charlevoix-Saguenay Foundation’s mission is to discover and develop a treatment for the disease.

The present call for proposals aims to funds projects that will clearly advance the understanding of the disease and lead the way to the development of a treatment for ARSACS patients.