

[Cell Lines](#) [SACS/ARSACS/SPAX6](#) [Publication](#)**7168****IDENTIFICATION**

Causal gene(s)	SACS
Repeat size or mutation	K1715*/R4331Q (compound heterozygous)
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	Obtained from: Radboud University Nijmegen Medical Center
Links to publications or public resources	<p>A reduction in Drp1-mediated fission compromises mitochondrial health in autosomal recessive spastic ataxia of Charlevoix Saguenay - PubMed https://pubmed.ncbi.nlm.nih.gov/27288452/</p> <p>Altered organization of the intermediate filament cytoskeleton and relocalization of proteostasis modulators in cells lacking the ataxia protein saccsin - PubMed https://pubmed.ncbi.nlm.nih.gov/28535259/</p> <p>The ARSACS disease protein saccsin controls lysosomal positioning and reformation by regulating microtubule dynamics - PubMed https://pubmed.ncbi.nlm.nih.gov/35933016/</p>