Cell Lines SACS/ARSACS/SPAX6 Publication

7078

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.2094-2A > G/Q4054* (compound heterozygous)
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	Obtained from: Radboud University Nijmegen Medical Center
Links to publications or public resources	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/ Altered organization of the intermediate filament cytoskeleton and relocalization of proteostasis modulators in cells lacking the ataxia protein sacsin - PubMed https://pubmed.ncbi.nlm.nih.gov/28535259/ The ARSACS disease protein sacsin controls lysosomal positioning and reformation by regulating microtubule dynamics - PubMed https://pubmed.ncbi.nlm.nih.gov/35933016/