Cell Lines SACS/ARSACS/SPAX6 Publication

# 7078

## IDENTIFICATION

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|---|---|
| **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<br>https://pubmed.ncbi.nlm.nih.gov/27288452/<br>Altered organization of the intermediate filament cytoskeleton and relocalization of proteostasis modulators in cells lacking the ataxia protein sacsin - PubMed<br>https://pubmed.ncbi.nlm.nih.gov/28535259/<br>The ARSACS disease protein sacsin controls lysosomal positioning and reformation by regulating microtubule dynamics - PubMed<br>https://pubmed.ncbi.nlm.nih.gov/35933016/ |