

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

# AAR-437-014

## IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	Protein change: p.R272H (missense homozygous variant)
Cell type	Fibroblast

## DONOR INFORMATION

Age at disease onset (years)	1
Age at collection (years)	26

## SOURCE & PUBLICATIONS

Originating lab / institution	University of Bordeaux
Links to publications or public resources	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay - PubMed <a href="https://pubmed.ncbi.nlm.nih.gov/26288984/">https://pubmed.ncbi.nlm.nih.gov/26288984/</a>