Cell Lines SACS/ARSACS/SPAX6 Publication

AAR-519-001

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	Protein change: p.P2099L (missense homozygous variant)
Cell type	Fibroblast

DONOR INFORMATION

Age at disease onset (years)	8
Age at collection (years)	39

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 https://pubmed.ncbi.nlm.nih.gov/26288984/