Cell Lines SACS/ARSACS/SPAX6 Publication

BOR-090711-003

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	Protein change: p.L1180Lfs*7 (nonsense compound heterozygous variant)
Cell type	Fibroblast

DONOR INFORMATION

Age at disease onset (years)	25
Age at collection (years)	48

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/