Cell Lines SACS/ARSACS/SPAX6 Publication

PN7

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.13132C > T, p.Arg4378X homozygous
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	14 months

SOURCE & PUBLICATIONS

Originating lab / institution	IRCCS Ospedale San Raffaele
Links to publications or public resources	Reduction of sacsin levels in peripheral blood mononuclear cells as a diagnostic tool for spastic ataxia of Charlevoix-Saguenay - PubMed https://pubmed.ncbi.nlm.nih.gov/39091421/