Cell Lines SACS/ARSACS/SPAX6 Publication

PN6

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.4593dupA, p.Ser1531fs*9 homozygous
Cell type	Fibroblast

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/