Cell Lines SACS/ARSACS/SPAX6 Publication

Pt1

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.5719C > T/c.12630_12634delTGAAA
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	2
Age at collection (years)	38

SOURCE & PUBLICATIONS

Originating lab / institution	IRCCS Stella Maris / Federico II University
Links to publications or public resources	Powerhouse failure and oxidative damage in autosomal recessive spastic ataxia of Charlevoix-Saguenay - PubMed https://pubmed.ncbi.nlm.nih.gov/26530509/