Cell Lines SACS/ARSACS/SPAX6 Publication

Pt3

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.8289_8291delTTC/c.10462_1043delCT
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	18
Age at collection (years)	50

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/ Thickening of peripapillar retinal fibers for the diagnosis of autosomal recessive spastic ataxia of Charlevoix-Saguenay - PubMed https://pubmed.ncbi.nlm.nih.gov/21597885/