Cell Lines SACS/ARSACS/SPAX6 Publication

Pt4

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.6166_6167delTT/c.6166_6167delTT
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	6
Age at collection (years)	33

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/