Cell Lines SACS/ARSACS/SPAX6 Publication

Pt5

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.9268_9271delGTTA/c.11319_11321delAAG
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
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/