Cell Lines SACS/ARSACS/SPAX6 Biobank

ESi063-A

IDENTIFICATION

Research Resource Identifier (RRID)	CVCL_VP83
Causal gene(s)	SACS
Repeat size or mutation	c.9938delC(p.G3313Qfs*11)/c.11374C>T(p.R3792*)
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male
Ethnicity	Caucasian
Age at collection (years)	14

SOURCE & PUBLICATIONS

Originating lab / institution	Research Center Principe Felipe
Links to publications or public resources	Generation of a human iPSC line from a patient with autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) caused by mutation in SACSIN gene - PubMed https://pubmed.ncbi.nlm.nih.gov/30144656/ ESi063-A u00b7 Cell Line u00b7 hPSCreg https://hpscreg.eu/cell-line/ESi063-A