Cell Lines SACS/ARSACS/SPAX6 Publication

FARSM50-5

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.8844delT (homozygous)
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male
Age at collection (years)	50

SOURCE & PUBLICATIONS

Originating lab / institution	Obtained from: CRCHU de Quebec's induced pluripotency stem cells production platform
Links to publications or public resources	In Vitro Characterization of Motor Neurons and Purkinje Cells Differentiated from Induced Pluripotent Stem Cells Generated from Patients with Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay - PubMed https://pubmed.ncbi.nlm.nih.gov/37096129/