Cell Lines SCA7 Publication

1

IDENTIFICATION

Causal gene(s)	ATXN7
Repeat size or mutation	50 CAG
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	47
Age at collection (years)	52

SOURCE & PUBLICATIONS

Originating lab / institution	University of Califorania
Links to publications or public resources	Metabolic and Organelle Morphology Defects in Mice and Human Patients Define Spinocerebellar Ataxia Type 7 as a Mitochondrial Disease - PubMed https://pubmed.ncbi.nlm.nih.gov/30699348/ Increased nuclear import characterizes aberrant nucleocytoplasmic transport in neurons from patients with spinocerebellar ataxia type 7 - PubMed https://pubmed.ncbi.nlm.nih.gov/39649105/