Cell Lines | SCA7 (unaffected family) | Publication

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IDENTIFICATION

Causal gene(s)	ATXN7
Repeat size or mutation	10/10 CAG; mother from 70 CAG SCA7)
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Female
Age at collection (years)	50

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/