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IDENTIFICATION

Causal gene(s)	ATXN7
Repeat size or mutation	30 CAG
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	35
Age at collection (years)	51

SOURCE & PUBLICATIONS

Originating lab / institution	University of California
Links to publications or public resources	<p>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/</p> <p>Increased nuclear import characterizes aberrant nucleocytoplasmic transport in neurons from patients with spinocerebellar ataxia type 7 - PubMed https://pubmed.ncbi.nlm.nih.gov/39649105/</p>