Cell Lines SCA1 Publication

11-036

IDENTIFICATION

Causal gene(s)	ATXN1
Repeat size or mutation	30/41 CAG
Cell type	hiPSC

SOURCE & PUBLICATIONS

Originating lab / institution	University of Minnesota
Links to publications or public resources	ATXN1-CIC Complex Is the Primary Driver of Cerebellar Pathology in Spinocerebellar Ataxia Type 1 through a Gain-of-Function Mechanism - PubMed https://pubmed.ncbi.nlm.nih.gov/29526553/ Expanded ATXN1 alters transcription and calcium signaling in SCA1 human motor neurons differentiated from induced pluripotent stem cells - PubMed https://pubmed.ncbi.nlm.nih.gov/39307401/ Cas9 editing of ATXN1 in a spinocerebellar ataxia type 1 mice and human iPSC-derived neurons - PubMed https://pubmed.ncbi.nlm.nih.gov/39314800/