

[Cell Lines](#) [SCA1](#) [Publication](#)

11-037

IDENTIFICATION

Causal gene(s)	ATXN1
Repeat size or mutation	33/46 CAG
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	University of Minnesota
Links to publications or public resources	<p>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/</p> <p>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/</p>