Cell Lines SCA3 Publication

iHF SCA3

IDENTIFICATION

Causal gene(s)	ATXN3
Repeat size or mutation	23/67 CAG
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	50
Age at collection (years)	66

SOURCE & PUBLICATIONS

Originating lab / institution	University of Tübingen
Links to publications or public resources	Mitochondrial Dysfunction in Spinocerebellar Ataxia Type 3 Is Linked to VDAC1 Deubiquitination - PubMed https://pubmed.ncbi.nlm.nih.gov/35682609/ A combinatorial approach to identify calpain cleavage sites in the Machado-Joseph disease protein ataxin-3 - PubMed https://pubmed.ncbi.nlm.nih.gov/28334907/