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AX-27

IDENTIFICATION

Causal gene(s)	ATXN3
Repeat size or mutation	76 CAG
Cell type	hiPSC

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	21
Age at collection (years)	29

SOURCE & PUBLICATIONS

Originating lab / institution	University of Tübingen
Links to publications or public resources	A combinatorial approach to identify calpain cleavage sites in the Machado-Joseph disease protein ataxin-3 - PubMed https://pubmed.ncbi.nlm.nih.gov/28334907/