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IDENTIFICATION

Causal gene(s)	ATXN3
Repeat size or mutation	72 CAG
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	30
Age at collection (years)	56

SOURCE & PUBLICATIONS

Originating lab / institution	University Medical Center Groningen (UMCG)
Links to publications or public resources	Levels of DNAJB family members (HSP40) correlate with disease onset in patients with spinocerebellar ataxia type 3 - PubMed https://pubmed.ncbi.nlm.nih.gov/20726892/ The HSPB8-BAG3 chaperone complex is upregulated in astrocytes in the human brain affected by protein aggregation diseases - PubMed https://pubmed.ncbi.nlm.nih.gov/21696420/