Cell Lines SCA28 Publication

L-10182

IDENTIFICATION

Causal gene(s)	AFG3L2
Repeat size or mutation	c.2167G>A p.V723M c.1847A>G p.Y616C (comp. heterozygous); affected son of L-11388
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Ethnicity	German/Romanian
Age at disease onset (years)	20
Age at collection (years)	39

SOURCE & PUBLICATIONS

Originating lab / institution	University of Lübeck
Links to publications or public resources	Spinocerebellar Ataxia Type 28-Phenotypic and Molecular Characterization of a Family with Heterozygous and Compound-Heterozygous Mutations in AFG3L2 - PubMed https://pubmed.ncbi.nlm.nih.gov/31111429/