Cell Lines SCA28 Publication

M666T

IDENTIFICATION

Causal gene(s)	AFG3L2
Repeat size or mutation	p.M666T
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Female
Age at disease onset (years)	23
Age at collection (years)	50

SOURCE & PUBLICATIONS

Originating lab / institution	IRCCS Ospedale San Raffaele
Links to publications or public resources	Pathogenic variants in the AFG3L2 proteolytic domain cause SCA28 through haploinsufficiency and proteostatic stress-driven OMA1 activation - PubMed https://pubmed.ncbi.nlm.nih.gov/30910913/