Cell Lines SCA28 Publication

Y689H

IDENTIFICATION

Causal gene(s)	AFG3L2
Repeat size or mutation	p.Y689H
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	43
Age at collection (years)	60

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/