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# 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 <a href="https://pubmed.ncbi.nlm.nih.gov/30910913/">https://pubmed.ncbi.nlm.nih.gov/30910913/</a>