

[Cell Lines](#) [SCA28](#) [Publication](#)

## F664S (p1)

### IDENTIFICATION

Causal gene(s)	AFG3L2
Repeat size or mutation	p.F664S (p1)
Cell type	Fibroblast

### DONOR INFORMATION

Donor gender	Male
Ethnicity	Italian
Age at disease onset (years)	34
Age at collection (years)	39

### 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>