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# PN2

## IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	p.[Cys72fsTer4];[(Arg3636Gln;Pro3652Thr)]
Cell type	Fibroblast

## DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	>25

## SOURCE & PUBLICATIONS

Originating lab / institution	University of Milan
Links to publications or public resources	Assessment of Sacsin Turnover in Patients With ARSACS: Implications for Molecular Diagnosis and Pathogenesis - PubMed <a href="https://pubmed.ncbi.nlm.nih.gov/34649874/">https://pubmed.ncbi.nlm.nih.gov/34649874/</a>