

[Cell Lines](#) [SACS/ARSACS/SPAX6](#) [Publication](#)

# PN9

## IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	p.[Pro536Leu];[Arg632Trp]
Cell type	Fibroblast

## DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	16

## 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>