Cell Lines SACS/ARSACS/SPAX6 Publication

PN1

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	p.[(Arg3636Gln;Pro3652Thr)];[Leu3745fsTer1]
Cell type	Fibroblast

DONOR INFORMATION

Donor gender	Male
Age at disease onset (years)	15

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 https://pubmed.ncbi.nlm.nih.gov/34649874/