Cell Lines SACS/ARSACS/SPAX6 Publication

ARSACS 3

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	p.F2780C & p.S3892* (missense/nonsense mutation)
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	Queen Mary University of London
Links to publications or public resources	Altered organization of the intermediate filament cytoskeleton and relocalization of proteostasis modulators in cells lacking the ataxia protein sacsin - PubMed https://pubmed.ncbi.nlm.nih.gov/28535259/