Cell Lines SACS/ARSACS/SPAX6 Publication

SK10A

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.9248dupC/p.Ala3096Cysfs*2
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	Obtained from: Queen Mary University of London
Links to publications or public resources	The ARSACS disease protein sacsin controls lysosomal positioning and reformation by regulating microtubule dynamics - PubMed https://pubmed.ncbi.nlm.nih.gov/35933016/