Cell Lines SACS/ARSACS/SPAX6 Publication

Patient line 1

IDENTIFICATION

Causal gene(s)	SACS
Repeat size or mutation	c.8339T>C (p.Phe2780Cys) and c.12416T>C (p.Leu4139Ser)/c.11675C>G (p.Ser3892X) (sibling from patient line 2)
Cell type	Fibroblast

SOURCE & PUBLICATIONS

Originating lab / institution	UCL Queen Square Institute of Neurology
Links to publications or public resources	Hsp90 Inhibition: A Promising Therapeutic Approach for ARSACS - PubMed https://pubmed.ncbi.nlm.nih.gov/34769152/