Mouse Models SCAR8/ARCA1 Publication

Nes1g STOP/STOP

IDENTIFICATION

Causal gene(s)	SYNE1
Repeat size or mutation	Depletion of giant isoforms of Nesprin1
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57/BL6
Type of model	CRISPR/Cas9-induced stop codon

TRANSGENIC CONSTRUCT

Transgenic construct: details	To specifically inhibit the expression of giant isoforms of Nesprin1, 16 bases including STOP codons in all reading frames and an EcoRI restriction site were inserted within exon ?64 of Syne1 used a CRISPR/Cas9 approach

PHENOTYPE

Progression	3 months
Hallmark features	Enlargement of verntricular volume

SOURCE & PUBLICATIONS

Originating lab / institution	Washington University School of Medicine
Links to publications or public resources	The KASH-containing isoform of Nesprin1 giant associates with ciliary rootlets of ependymal cells - PubMed https://pubmed.ncbi.nlm.nih.gov/29630990/