Mouse Models SCA38 Publication

Elovl5-/- mice

IDENTIFICATION

Causal gene(s)	ELOVL5
Repeat size or mutation	Homozygous Elovl5 knock-out mice
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57BL/6J
Type of model	Knockout

TRANSGENIC CONSTRUCT

Transgenic construct: details	ES cell clone (DTM098) harbors an insertion of gene trap vector 7.9 kb downstream of exon 1 of Elovl5 resulting in knock-out. ES cells were injected in mice blastocyst

PHENOTYPE

Hallmark features	Motor coordination impairment, disruption of cerebellar architecture, alterations in cerebellar excitatory presynaptic mechanisms

SOURCE & PUBLICATIONS

Originating lab / institution	University of Texas Southwestern Medical Center at Dallas
Links to publications or public resources	Deletion of ELOVL5 leads to fatty liver through activation of SREBP-1c in mice - PubMed https://pubmed.ncbi.nlm.nih.gov/18838740/ Effects of the administration of Elovl5-dependent fatty acids on a spino-cerebellar ataxia 38 mouse model - PubMed https://pubmed.ncbi.nlm.nih.gov/35933444/ Impact of Elovl5 Deficiency on Cerebellar Excitatory Synaptic Transmission in Mice - PubMed https://pubmed.ncbi.nlm.nih.gov/40789653/