Mouse Models SCA19/22 Publication

Kcnd3 F227del mutant mice

IDENTIFICATION

Causal gene(s)	KCND3
Repeat size or mutation	Knock-in of Kcnd3 F227del, both heterozygous and homozygous
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	B6N x C57BL/6J
Type of model	Knock-in

PHENOTYPE

Hallmark features	Motor coordination impairments, Purkinje cell degeneration, neuroinflammation, degeneration of Golgi apparatus, degeneration of mitochondria, degeneration of ER

SOURCE & PUBLICATIONS

Originating lab / institution	National Yang Ming Chiao Tung University
Links to publications or public resources	A dominant negative Kcnd3 F227del mutation in mice causes spinocerebellar ataxia type 22 (SCA22) by impairing ER and Golgi functioning - PubMed https://pubmed.ncbi.nlm.nih.gov/39562497/