Mouse Models SCA21 Publication

R116C mutant TMEM-FL

IDENTIFICATION

Causal gene(s)	TMEM240
Repeat size or mutation	AAV9 with R116C mutant TMEM-FL in cerebellar neurons
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57BL/6J
Type of model	Induced (AAV9 infection)

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	TMEM240 human CDNA
Transgenic construct: details	TMEM240 human CDNA were cloned in AAV9 vectors under the regulation of a neuron-specific synapsin 1 promoter. The R116C mutation was introduced using PrimeStar mutagenesis basal kit. AAV9 vectors were injected in cerebella of 4 week-old-mice
Promoter: gene	synapsin 1

PHENOTYPE

Progression	Progressive
Hallmark features	Impaired motor coodrination, activation of microglia and astrocytes, autophagic lysosomal protein degradation, neural cell death

SOURCE & PUBLICATIONS

Originating lab / institution	Kumamoto University
Links to publications or public resources	Lysosomal dysfunction and early glial activation are involved in the pathogenesis of spinocerebellar ataxia type 21 caused by mutant transmembrane protein 240 - PubMed https://pubmed.ncbi.nlm.nih.gov/30184469/