Mouse Models SCA11 Publication

conditional Ttbk2 KO (Ttbk2pcp2)

IDENTIFICATION

Causal gene(s)	TTBK2
Repeat size or mutation	Conditional Ttbk2 knockout specifically in the Purkinje cells
Animal model	Mouse

MODEL DETAILS

Type of model	Knockout

TRANSGENIC CONSTRUCT

Transgenic construct: details	a conditional allele of Ttbk2 from the European Mutant Mouse Cell Repository (Ttbk2tm1c(EUCOMM)Hmgu) was crossed with a mouse line expressing tamoxifen-inducible Cre recombinase driven by a Pcp2 promoter
Promoter: gene	Pcp2
Promoter: species	Mouse

PHENOTYPE

Hallmark features	lower number of primary cilia, loss of VGLUT2 synapses in cerebellum, locomotor deficiencies

SOURCE & PUBLICATIONS

Originating lab / institution	Duke University
Links to publications or public resources	TTBK2 and primary cilia are essential for the connectivity and survival of cerebellar Purkinje neurons - PubMed https://pubmed.ncbi.nlm.nih.gov/31934864/