Mouse Models SCA11 Publication

TTBK2 knockin mice

IDENTIFICATION

Causal gene(s)	TTBK2
Repeat size or mutation	Heterozygous TTBK2 knock-in of one-base insertion in exon 13 at nucleotide 1329
Animal model	Mouse

MODEL DETAILS

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

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Human TTBK2 exon 13 with an one-base insertion at nucleotide 1329
Transgenic construct: details	Knock-in construct containing exon 13 with the one-base insertion of adenosine at nucleotide 1329 within LoxP sites and a puromycin resistance gene. After homologous recombination, Flp recombination removes the puromycin resistance gene.

SOURCE & PUBLICATIONS

Originating lab / institution	University of Dundee
Links to publications or public resources	TTBK2 kinase substrate specificity and the impact of spinocerebellar-ataxia-causing mutations on expression, activity, localization and development - PubMed https://pubmed.ncbi.nlm.nih.gov/21548880/