Mouse Models SCA31 Publication

Tk2-/- KO mice (no ataxia phenotype)

IDENTIFICATION

Causal gene(s)	BEAN1/TK2
Repeat size or mutation	Homozygous Tk2 knock-out mice
Animal model	Mouse

MODEL DETAILS

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

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Replacement of exon 4 and part of exon 5 of the Tk2 gene by a neomycin resistance cassette
Transgenic construct: details	A targeting vector containing exon 3 till 6 of the Tk2 gene, the region containing exon 4 and 5 (bp 7066-9753) was replaced by a neomycin resistance cassette

PHENOTYPE

Progression	Progressive
Hallmark features	Growth retardation, high rate of early mortality, hypothermia, progressive depletion of mtDNA levels, cardiomyopathy, lipodystrophy

SOURCE & PUBLICATIONS

Originating lab / institution	Karolinska Institute
Links to publications or public resources	Progressive loss of mitochondrial DNA in thymidine kinase 2-deficient mice - PubMed https://pubmed.ncbi.nlm.nih.gov/18434326/