Mouse Models DRPLA Publication

Atn1Q112/+ mouse

IDENTIFICATION

Causal gene(s)	ATN1
Repeat size or mutation	112Q
Animal model	Mouse

MODEL DETAILS

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

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	human ATN1 DNA
Transgenic construct: details	endogenous mouse Atn1 allele was replaced with the human ATN1 sequence, from 5'UTR to 3'UTR with expanded pure 112 CAG repeats in exon 5 using CRISPR/Cas9

PHENOTYPE

Progression	Progressive
Hallmark features	Incoordination, gait disturbances, tremors, reduced brain size, aggregate accumulation

SOURCE & PUBLICATIONS

Originating lab / institution	University of Washington
Links to publications or public resources	Atrophin-1 antisense oligonucleotide provides robust protection from pathology in a fully humanized DRPLA model - PubMed https://pubmed.ncbi.nlm.nih.gov/41624332/