Mouse Models SCA36 Publication

NOP56-(TG3C2)62

IDENTIFICATION

Causal gene(s)	NOP56
Repeat size or mutation	AAV9 with human NOP56 (TG3C2)62
Animal model	Mouse

MODEL DETAILS

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

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	human NOP56 gDNA with 62 (TG3C2) repeats
Transgenic construct: details	Expression vector containing NOP56 (TG3C2)62 human gDNA was subcloned in AAV expression vector (pAM/CBA-pl-WPRE-BGH) containing inverted repeats of serotype 2 and packaged into AAV9 capsids following intracerebroventricular AAV injections

PHENOTYPE

Progression	Progressive
Hallmark features	Clasping phenotype, locomotor defects, loss of cerebellar Purkinje cells, narrowing of molecular cell layer, presence of RNA foci in brain, RAN translation

SOURCE & PUBLICATIONS

Originating lab / institution	Mayo Clinic
Links to publications or public resources	Hexanucleotide Repeat Expansions in c9FTD/ALS and SCA36 Confer Selective Patterns of Neurodegeneration In Vivo - PubMed https://pubmed.ncbi.nlm.nih.gov/32375043/