Mouse Models DRPLA Publication

At65QmNES mice

IDENTIFICATION

Causal gene(s)	ATN1
Repeat size or mutation	65Q with alteration of nuclear export signal
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C3H/B6
Type of model	Transgenic

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Full-length cDNA
Transgenic construct: details	Full-length cDNA of atrophin-1 encoding 65Q and alterations in the nuclear export signal (amino acids 1033-1041) were cloned into an expression plasmid leading to expression of transgenes in CNS neurons
Promoter: gene	PrP
Promoter: species	Mouse

PHENOTYPE

Progression	Progressive
Hallmark features	Increased nuclear accumulation and aggregation of mutant atropin-1 in neurons, shorter lifespan, reduced survival, locomotor abnormalities, astrogliosis

SOURCE & PUBLICATIONS

Originating lab / institution	Johns Hopkins University School of Medicine
Links to publications or public resources	The nuclear export signal mediates mutant atrophin-1-induced neuropathology in a mouse model of DRPLA - PubMed https://pubmed.ncbi.nlm.nih.gov/40263757/