Mouse Models DRPLA Publication

Transgenic DRPLA 129Q Mice

IDENTIFICATION

Causal gene(s)	ATN1
Repeat size or mutation	129Q
Animal model	Mouse

MODEL DETAILS

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

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Entire mutant DRPLA gene
Transgenic construct: details	Accidental expansion of 76Q repeat at the early embryonic stage resulted in a Q129 mouse
Promoter: gene	SV40

PHENOTYPE

Progression	Progressive
Hallmark features	myoclonic movements, ataxia, epilepsy, dysfunction of pallidal and cerebellar neurons, dysfunction of synaptic transmission in hippocampal CA1 pyramidal cells, intranuclear inclusions

SOURCE & PUBLICATIONS

Originating lab / institution	Niigata University
Links to publications or public resources	Severe neurological phenotypes of Q129 DRPLA transgenic mice serendipitously created by en masse expansion of CAG repeats in Q76 DRPLA mice - PubMed https://pubmed.ncbi.nlm.nih.gov/19039037/