Mouse Models DRPLA Publication

Transgenic DRPLA 76Q Mice

IDENTIFICATION

Causal gene(s)	ATN1
Repeat size or mutation	Q76
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	A clone containing the entire mutant DRPLA gene with 78 repeats, a SV40 promoter and neomycin gene
Promoter: gene	SV40

PHENOTYPE

Hallmark features	Intergenerational and somatic instabilites of expanded CAG repeats

SOURCE & PUBLICATIONS

Originating lab / institution	Niigata University
Links to publications or public resources	Transgenic mice harboring a full-length human mutant DRPLA gene exhibit age-dependent intergenerational and somatic instabilities of CAG repeats comparable with those in DRPLA patients - PubMed https://pubmed.ncbi.nlm.nih.gov/9887337/ 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/