Mouse Models DRPLA Publication

AT-FL-65Q;GFP-LC3

IDENTIFICATION

Causal gene(s)	ATN1
Repeat size or mutation	65Q with GFP-LC3
Animal model	Mouse

MODEL DETAILS

Type of model	Transgenic

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Full-length cDNA for Atn1 and DNA fragment for GFP-LC3
Transgenic construct: details	Full-length cDNA of atrophin-1 encoding 65Q constructs were ligated into the mouse prion protein promoter vector ensuring expression in neurons throughout the brain; A 1.8-kbp DNA fragment containing the rat LC3 cDNA fused to EGFP at the N-terminus (GFP-LC3)

PHENOTYPE

Hallmark features	Stalled canonical autophagy shown by accumulation of p62, downregulation of LC3-I/II conversion and reduced Tfeb expression

SOURCE & PUBLICATIONS

Originating lab / institution	King's College London
Links to publications or public resources	Stall in Canonical Autophagy-Lysosome Pathways Prompts Nucleophagy-Based Nuclear Breakdown in Neurodegeneration - PubMed https://pubmed.ncbi.nlm.nih.gov/29174892/