Mouse Models SPG7 Publication

Spg7?/?Afg3l2Emv66/+ mice

IDENTIFICATION

Causal gene(s)	SPG7
Repeat size or mutation	Spg7 homozygous knockout and heterozygous Afg3l2 mice
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57BL6-129Sv x FVB
Type of model	Knockout

PHENOTYPE

Progression	Progressive
Hallmark features	Loss of balance, tremor, ataxia, axonopathy, cerebellar degeneration, loss of Purkinje cells and parallel fibers, reactive astrogliosis, mitochondrial abnormalities

SOURCE & PUBLICATIONS

Originating lab / institution	Istituto Neurologico C. Besta
Links to publications or public resources	Genetic interaction between the m-AAA protease isoenzymes reveals novel roles in cerebellar degeneration - PubMed https://pubmed.ncbi.nlm.nih.gov/19289403/