Mouse Models FXTAS Publication

L7CGG90Fmr1

IDENTIFICATION

Causal gene(s)	FMR1
Repeat size or mutation	Specific expression of CGG repeats in Fmr1 in Purkinje neurons
Animal model	Mouse

MODEL DETAILS

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

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	human (CGG)98 repeat
Transgenic construct: details	FLAG-tagged Fmr1 with (CGG)90 repeats cDNA constructs under the L7/pcp2 promoter
Promoter: gene	L7/pcp2
Promoter: species	Mouse

PHENOTYPE

Progression	Progressive
Hallmark features	Intranuclear inclusions in Purkinje neurons, purkinje cell death, behavioral deficits

SOURCE & PUBLICATIONS

Originating lab / institution	Baylor College of Medicine
Links to publications or public resources	Ectopic expression of CGG containing mRNA is neurotoxic in mammals - PubMed https://pubmed.ncbi.nlm.nih.gov/19377084/