Mouse Models FXTAS Publication

Fragile X premutation knock-in mice

IDENTIFICATION

Causal gene(s)	FMR1
Repeat size or mutation	Knock-in of mouse Fmr1 gene with expanded CGG repeat
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57/BL6
Type of model	Knock-in

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Mouse Fmr1 gene fragment (promoter, exon1, intron1) with an expanded CGG repeat (118 CGG)
Transgenic construct: details	Targeting construct containing the promoter, exon1 and intron 1 of the mouse Fmr1 gene. A repeat tract expansion (118 CGG) in exon 1 was generated using a serial ligation strategy
Promoter: gene	Fmr1
Promoter: species	Mouse

PHENOTYPE

Hallmark features	Neuronal intranuclear inclusions, PC pathology, regional Fmrp deficits (more in thalamus and inferior olivary complex), repeat instability

SOURCE & PUBLICATIONS

Originating lab / institution	National Institutes of Health
Links to publications or public resources	Regional FMRP deficits and large repeat expansions into the full mutation range in a new Fragile X premutation mouse model - PubMed https://pubmed.ncbi.nlm.nih.gov/17442505/