Mouse Models FXTAS Publication

Fmr1 (CGG)98 knock-in mice

IDENTIFICATION

Causal gene(s)	FMR1
Repeat size or mutation	CGG knock-in mouse with 98 CGG
Animal model	Mouse

MODEL DETAILS

Type of model	Knock-in

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	The mouse (CGG)9 repeat was exchanged for a human (CGG)98 repeat
Transgenic construct: details	Construct with mouse Fmr1 promoter where the mouse (CGG)8 was exchanged for a human (CGG)98 repeat
Promoter: gene	Fmr1
Promoter: species	Mouse

PHENOTYPE

Progression	Progressive
Hallmark features	Cognitive decline and neutomotor disturbances

SOURCE & PUBLICATIONS

Originating lab / institution	Erasmus University Rotterdam
Links to publications or public resources	Instability of a (CGG)98 repeat in the Fmr1 promoter - PubMed https://pubmed.ncbi.nlm.nih.gov/11487573/ Cognitive decline, neuromotor and behavioural disturbances in a mouse model for fragile-X-associated tremor/ataxia syndrome (FXTAS) - PubMed https://pubmed.ncbi.nlm.nih.gov/15876460/