Mouse Models FXTAS Publication

?5'UTR FMR1 (CGG)99

IDENTIFICATION

Causal gene(s)	FMR1
Repeat size or mutation	Full human FMR1 5'UTR with 99 expanded CGG expressing only CGG RNA
Animal model	Mouse

MODEL DETAILS

Type of model	Transgenic

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Full human FMR1 5'UTR with 99 expanded CGG expressing only CGG RNA and not the FMRpolyG protein
Transgenic construct: details	Full human FMR1 5'UTR with 99 expanded CGG expressing only CGG RNA and not the FMRpolyG protein with CAG promoter inserted in Rosa26 locus
Promoter: gene	CAG

SOURCE & PUBLICATIONS

Originating lab / institution	Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC)
Links to publications or public resources	Translation of Expanded CGG Repeats into FMRpolyG Is Pathogenic and May Contribute to Fragile X Tremor Ataxia Syndrome - PubMed https://pubmed.ncbi.nlm.nih.gov/28065649/