Mouse Models FXTAS Publication

FMRP-P608L mutation mice

IDENTIFICATION

Causal gene(s)	FMR1
Repeat size or mutation	Fmr1 missense mutation c.1823C>T (Pro608Leu)
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57BL/6J
Type of model	CRISPR/Cas9-induced mutation

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	Fmr1 missense mutation c.1823C>T (Pro608Leu) in exon 17 was generated with CRISPR/Cas9-mediated genome engineering

PHENOTYPE

Progression	Progressive
Hallmark features	Impaired motor behavior, decreased striatal dopamine levels

SOURCE & PUBLICATIONS

Originating lab / institution	The Second Affiliated Hospital of Guangzhou Medical University
Links to publications or public resources	Age-dependent impairment of dopamine D1 receptor signalling in mouse striatum by FMR1 variant P626L - PubMed https://pubmed.ncbi.nlm.nih.gov/40980401/