Mouse Models AOA1 Publication

AtmR35X/R35X; Aptx-/- mice

IDENTIFICATION

Causal gene(s)	APTX
Repeat size or mutation	Aptx homozygous knockout with homozygous R35X mutation in Atm gene (causing ataxia telangiectasia) mice
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57Bl/6 and 129 x C57Bl/6J
Type of model	Knockout

PHENOTYPE

Progression	Progressive
Hallmark features	Ataxic phenotype, atrophy of cerebellar molecular layer, Purkinje cell signaling dysfunction, predisposition to develop cancer, immune abnormalities

SOURCE & PUBLICATIONS

Originating lab / institution	Harbor UCLA medical Center
Links to publications or public resources	A novel, ataxic mouse model of ataxia telangiectasia caused by a clinically relevant nonsense mutation - PubMed https://pubmed.ncbi.nlm.nih.gov/34723800/