

[Mouse Models](#)[AOA1](#)[Publication](#)

AtmR35X/+; Aptx-/- mice

IDENTIFICATION

Causal gene(s)	APT _X
Repeat size or mutation	Aptx homozygous knockout with heterozygous 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

Hallmark features	Ataxic phenotype, atrophy of cerebellar molecular layer, Purkinje cell signaling dysfunction,
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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/