Mouse Models AOA2/SCAN2/SCAR1 Publication

Setx-/-

IDENTIFICATION

Causal gene(s)	SETX
Repeat size or mutation	Homozygous Setx knock-out mice
Animal model	Mouse

MODEL DETAILS

Mouse strain / background	C57BL6 and 129Sv
Type of model	Knockout

TRANSGENIC CONSTRUCT

Transgenic construct: sequence type	A Cre-LoxP system was used to delete exon 4 of the Setx gene.
Transgenic construct: details	A Neo cassette with a LoxP site was inserted into a BAC clone corresponding to mouse chromosome 2 and the Setx genomic sequence. These were transfected in ES cells and Neo+ cells were selected. Neo cassette was excised by crossing with a Cre deleter strain to generate the Setx-/- mice

PHENOTYPE

Hallmark features	No phenotype related to AOA2, more DNA double-strand breaks, defect in disassembly of Rad51 filaments, accumulation of DNA/RNA hybrids (R loops), failure of crossing-over

SOURCE & PUBLICATIONS

Originating lab / institution	University of Queensland
Links to publications or public resources	R-loops in proliferating cells but not in the brain: implications for AOA2 and other autosomal recessive ataxias - PubMed https://pubmed.ncbi.nlm.nih.gov/24637776/