The Jackson Laboratory

STOCK *Mecp2* $^{tm1.1Jtc}$ /SchvJ

Stock No: **024990** | B6J;129S6.MeCP2 ^{R168X}

Targeted Mutation

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

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express a truncated R168X protein with a partial NLS and lacking the transcriptional repression domain (TRD) for interaction with corepressors. This RTT model is useful for testing highly robust behavioral paradigms in preclinical drug trials.

Donating Investigator

Laura R Schaevitz, Tufts University

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GENETIC OVERVIEW

Genetic Background	Generation

Mecp2$^{tm1.1Jtc}$

Allele Type	Gene Symbol	Gene Name
Targeted (Humanized sequence)	*Mecp2*	methyl CpG binding protein 2

VIEW GENETICS

RESEARCH APPLICATIONS

Neurobiology Research
Developmental Biology Research

VIEW ALL RESEARCH APPLICATIO

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

VIEW PRICE LIST

Details

Detailed Description

The early truncating MeCP2^{R168X} nonsense point mutation in the methyl-CpG binding domain of MeCP2 is one of the most common MeCP2 mutations associated with Rett syndrome (RTT). The largely truncated R168X protein retains the capacity to bind methylated DNA and carries a partial nuclear localization signal, but lacks the transcriptional repression domain for interaction with co-repressors. A second termination (*BspH*1 restriction site) downstream of the R168X mutation is reported not to affect the mutant product. MeCP2^{R168X} mice are a model of RTT; exhibiting respiratory, neuromuscular and behavioral abnormalities similar, but not identical, to that of Mecp2 null mice. The phenotype is described in detail below. These mice also underscore the importance of including Mecp2 mutant females in preclinical studies.

MeCP2^{R168X} mice on a mixed genetic background of ~87-93% C57BL/6J and ~13-6% 129S6/SvEvTac (B6J;129S6.MeCP2^{R168X}) are Stock No. 024990. In an attempt to offer alleles on well-characterized or multiple genetic backgrounds, alleles are frequently moved to a genetic background different from that on which an allele was first characterized. It should be noted that the phenotype of these B6J;129S6.MeCP2^{R168X} mice could vary from that originally described on other genetic backgrounds. We will modify the strain description if necessary as published results become available.

Lawson-Yeun *et al.* 2007 Brain Res 1180:1 reports the phenotype of Mecp2^{R168X} animals backcrossed at least 10 generation onto the 129S6/SvEvTac genetic background (129S6.Mecp2^{R168X}) as: hemizygous males (MeCP2$^{R168X/Y}$) have a shortened lifespan (average 86 days) with forelimb stereotypies, hindlimb atrophy, hypoactivity and breathing irregularities. By 7 weeks of age, significant hindlimb clasping is evident. Female heterozygotes (MeCP2$^{R168X/+}$) manifest significant defects by approximately 6 months (hindlimb clasping, breathing irregularities) and can survive past 1 year of age.

Schaevitz *et al.* 2013 Genes Brain Behav 12:732 reports the phenotype of 129S6.Mecp2^{R168X} animals backcrossed 1-2 generations onto C57BL/6J as: MeCP2^{R168X} mutants mirror many clinical features of human RTT. MeCP2$^{R168X/Y}$ males exhibit growth, motor, respiratory and cognitive abnormalities, and reduced anxiety. The phenotype is less severe and with later onset in MeCP2$^{R168X/+}$ females with the exception of seizures; ~4% of MeCP2$^{R168X/+}$ females exhibit tonic-clonic seizures that typically result in death. The phenotype in MeCP2$^{R168X/Y}$ males is similar to that reported for Mecp2 null males (such as *Mecp2$^{tm1.1Bird}$*; Stock No. 003890). Compared to females heterozygous for the Mecp2 null mutation, MeCP2$^{R168X/+}$ females exhibit delayed motor defect onset, normal anxiety-like behavior and increased seizure susceptibility.

Bissonnette *et al.* 2014 Neuroscience 267:166 reports the phenotype of Mecp2^{R168X} animals on a mixed C57BL/6J;129S6/SvEvTac genetic background as: MeCP2$^{R168X/+}$ females display augmented hypoxic ventilatory responses and depressed hypercapnic responses: the incidence of apnea is much greater in MeCP2$^{R168X/+}$ females (189 per hour) than MeCP2$^{T158A/+}$ females (41 per hour).

For B6J;129S6.MeCP2^{R168X} mice, the donating investigator reports that mating hemizygous males with wildtype females does not produce offspring; it is not known if the hemizygous males are sterile or if they simply do not breed as a result of their Rett syndrome-like phenotype. The donating investigator also reports that backcrossing MeCP2^{R168X} mice onto the C57BL/6 genetic background leads to increased phenotype severity.

The phenotype differences observed between male and female mice is because *Mecp2* is located on the X chromosome. Due to X-chromosome inactivation, heterozygous females have mosaic expression of wildtype MeCP2.

Development

Control Suggestions

Selected References

Genetics

Mecp2$^{tm1.1Jtc}$

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

CONTACT TECHNICAL SUPPORT

Genotyping Protocols
Sanger sequencing:Mecp2
Genotyping resources and troubleshooting

Breeding Considerations

The MeCP2^{R168X} mutant allele is located on the X chromosome. Hemizygous males exhibit features of Rett syndrome and have a shortened lifespan. Heterozygous females may live more than one year and have a less severe phenotype with later onset. When maintaining a live colony, heterozygous females may be bred with wildtype males from the colony.
The donating investigator reports that mating hemizygous males with wildtype females does not produce offspring; it is not known if the hemizygous males are sterile or if they simply do not breed as a result of their Rett syndrome-like phenotype.
The donating investigator also reports that backcrossing MeCP2^{R168X} mice onto the C57BL/6 genetic background leads to increased phenotype severity. The expected coat colors are agouti and black.

Additional Breeding and Husbandry Support

Citation

When using the B6J;129S6.MeCP2^{R168X} mouse strain in a publication, please cite the originating article(s) and include JAX stock #024990 in your Materials and Methods section.

Animal Health Reports

Facility Barrier Level Descriptions

Production of mice from cryopreserved embryos or sperm occurs in a maximum barrier room, G200

Pricing & Availability

Cryo Recovery

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

DomestiqInternatio

Pricing effective for USA, Canada and Mexico shipping destinations

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	X linked = Females are heterozygous and males are wildtype for Mecp2<tm1.1Jtc> X linked	$2,854.50

RELATED PRODUCTS AND SERVICES

Frozen Mouse Embryo	STOCK Mecp2<tm1.1Jtc>/SchvJ	$2595.00

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limited to outside agencies and court filings.

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The Jackson Laboratory has rigorous genetic quality control and mutant gene genotyping programs to ensure the genetic background of JAX® Mice strains as well as the genotypes of strains with identified molecular mutations. JAX® Mice strains are only made available to researchers after meeting our standards. However, the phenotype of each strain may not be fully characterized and/or captured in the strain data sheets. **Therefore, we cannot guarantee a strain's phenotype will meet all expectations.** To ensure that JAX® Mice will meet the needs of individual research projects or when requesting a strain that is new to your research, we suggest ordering and performing tests on a small number of mice to determine suitability for your particular project. We do not guarantee breeding performance and therefore suggest that investigators order more than one breeding pair to avoid delays in their research.

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Related Strains

All

By Allele

By Gene

By Collection

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