The Jackson Laboratory

MRL/MpJ-*Atp6v1b1* ^{vtx}/Kjn

Stock No: **021771** | vortex

Coisogenic, Spontaneous Mutation

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

PLACE ORDER

Email Download PDF Help

human genetic disease distal renal tubular acidosis with progressive nerve deafness. On the MRL/MpJ inbred background vortex homozygotes have inner ear defects that are not found in C57BL/6J congenic vortex homozygotes. On both backgrounds this mutation causes hypocalciuria, with greater severity on the MRL/MpJ background, but increased urine pH is only found on the C57BL/6J background, possibly due to the inherently high urine pH of the MRL/MpJ inbred strain.

READ MORE +

GENETIC OVERVIEW

Genetic Background

000486 MRL/MpJ

Generation

Atp6v1b1^{vtx}

Allele Type	Gene Symbol	Gene Name
Spontaneous (Not Specified)	*Atp6v1b1*	ATPase, H+ transporting, lysosomal V1 subunit B1

VIEW GENETICS

RESEARCH APPLICATIONS

Internal/Organ Research
Metabolism Research
Sensorineural Research
Cell Biology Research

VIEW ALL RESEARCH APPLICATIO

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

VIEW PRICE LIST

Details

Detailed Description

The vortex mutation is a spontaneous point mutation causing a G78D amino acid substitution in *Atp6v1b1*, a component of the vacuolar H+ ATPase pump that is active in the kidneys and inner ear. Mutations in the human ortholog, including two separate instances of G78D substitutions, have been shown to cause distal renal tubular acidosis with progressive nerve deafness (OMIM #267300). On the MRL/MpJ background the vortex mutation causes developmental defects of the inner ear with profound hearing and vestibular impairment in homozygotes, likely the result of failed fluid homeostasis. Although they are generally healthy and have normal fertility, vortex homozygotes on the MRL/MpJ background can be identified before wean age by their circling and head tilting. They are deaf, with ABR thresholds essentially absent across a broad range of frequencies and measurements of the endocochlear potential are nearly absent. At E15 the membranous labyrinth is dilated relative to controls, with the endolymphatic sac, duct, utricle, saccule, and cochlear duct showing swelling; at P1 the cochlear duct is enlarged and what little there is of utriclular or saccular otoconia is dispersed; and in adults histology finds lateral expansion of cochlear fluid spaces, basal displacement of the organ of Corti, small scala tympani, expanded scala media, displacement of the Reissner's membrane, elongated yet stubby spiral limbus, very thin spiral ligament, very few type I or III fibrocytes, and the boney interscalar septum between the lower cochlear base and lower apical turn is often absent. The inner hair cells are reasonably normal, but the outer hair cells are lacking in the cochlear base and variably present in the apex. While the vestibular organs show normal cristae and no distinct hair cell or neural loss, there are very few otoconia but there are some very large otoconia, mostly in the saccule, that have triangular shapes and in 5 of 9 homozygotes assessed these large, misshapen otoconia were also found in the cochlear scala media.

While vortex homozygotes on either the MRL/MpJ or C57BL/6J background have normal potassium, sodium, and chloride concentrations in blood and urine, both show decreased calcium in the urine, with only 45.9 +/- 6.01 mg/g in MRL/MpJ homozygotes relative to 163.3 +/- 17.4 mg/g in MRL/MpJ controls, a reduction of approximately 70%, and 99 +/- 9.6 mg/g in C57BL/6J congenic homozygotes versus 139.2 +/- 9.1 in C57BL/6J controls, a reduction of approximately 30%. No elevation in urine pH was found on the MRL background, but the MRL/MpJ inbred strain has a naturally high urine pH relative to other inbred strains. On the C57BL/6J congenic background (see Stock No. 021772) an elevation in urine pH is found, but the inner ear develops normally and ABR thresholds and endocochlear potential measurements are normal. Vortex homozygotes on an (MRL x B6)F1 hybrid background have ABR thresholds much closer to normal than do homozygotes on the MRL/MpJ background. An MRL/MpJ modifier that contributes to the vortex hearing loss phenotype was mapped to Chromosome 13 between rs3719347 and rs3023390. No abnormal phenotypes have been identified in heterozygotes on either inbred genetic background. (Tian et al., 2017.)

Development

Control Suggestions

Selected References

Genetics

Atp6v1b1^{vtx}

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

CONTACT TECHNICAL SUPPORT

Genotyping Protocols

Separated PCR:GAL Panel
Genotyping resources and troubleshooting

Mating System

Homozygote x Heterozygote
Heterozygote x Homozygote

Citation

When using the vortex mouse strain in a publication, please cite the originating article(s) and include JAX stock #021771 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.

Domestic **Internatic**

Pricing effective for USA, Canada and Mexico shipping destinations

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	Heterozygous for Atp6v1b1<vtx>	$2,854.50

RELATED PRODUCTS AND SERVICES

Frozen Mouse Embryo	MRL/MpJ-Atp6v1b1<vtx>/Kjn	$2595.00

PAYMENT TERMS AND CONDITIONS

Terms are granted by individual review and stated on the customer invoice(s) and account statement. These transactions are payable in U.S. currency within the granted terms. Payment for services, products, shipping containers, and shipping costs that are rendered are expected within the payment terms indicated on the invoice or stated by contract. Invoices and account balances in arrears of stated terms may result in The Jackson Laboratory pursuing collection activities including but not limited to outside agencies and court filings.

THE JACKSON LABORATORY'S GENOTYPE PROMISE

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.

Terms Of Use

TERMS OF USE

General Terms and Conditions

QUESTIONS ABOUT TERMS OF USE

LICENSING INFORMATION

Phone: 207-288-6470
Email: TechTran@jax.org

Related Strains

All

By Allele

By Gene

By Collection

Leading the search for

TOMORROW'S CURES