The Jackson Laboratory

B6.129-*Npc1* ^{tm1Dso}/J

Stock No: **027704** | Npc1 $^{tm(I1061T)Dso}$

Congenic, 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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as the I1061T missense mutation commonly found in humans with the cholesterol–sphingolipid lysosomal storage disorder, Niemann-Pick type C1 (NPC1) disease.

Donating Investigator

Daniel S. Ory, Washington University School of Medicine

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GENETIC OVERVIEW

Genetic Background **Generation**

VIEW GENETICS

RESEARCH APPLICATIONS

Neurobiology Research
Developmental Biology Research
Metabolism Research
Research Tools

VIEW ALL RESEARCH APPLICATION

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

VIEW PRICE LIST

Details

Detailed Description

Npc1$^{tm(I1061T)Dso}$ mutant mice possess *loxP* sites flanking exons 14-20 of the Niemann-Pick type C1 (*Npc1*) gene. They also contain the I1061T missense mutation in exon 21, which is commonly found in humans with Niemann-Pick type C1 (NPC1) disease. NPC1 is a lysosomal membrane protein that mediates intracellular cholesterol trafficking. Mutations in *Npc1* are responsible for Niemann-Pick type C (NPC) disease, a fatal neurodegenerative disease caused by a defect in cholesterol–sphingolipid lysosomal storage. Niemann-Pick Type C1 (NPC1) disease is a rare form of the disease and is characterized by ataxia, motor impairment, progressive intellectual decline, and dementia. Mice that are homozygous for this allele are viable with an average lifespan of 125 days. At 10 weeks of age mice begin to lose weight compared to controls and heterozygous littermates. Weight loss is progressive during later stages of disease. At 8 weeks of age mice exhibit visible resting tremors, and by 12 weeks the mice are no longer able to maintain balance on a rotating rod. NPC1^{I1061T} protein has a reduced half-life in vivo, consistent with protein misfolding and rapid endoplasmic reticulum-associated degradation. Mice also showed progressive Purkinje cell degeneration in the cerebellum

When bred to mice that express tissue-specific Cre recombinase, resulting offspring will have exons 14-20 deleted in the *cre*-expressing tissues, creating a *Npc1* null mouse model. Homozygotes KO mice typically exhibit lysosomal storage of non-esterified cholesterol, neurodegeneration, ataxia, presence of foam cells, sterility, and a shortened lifespan.

Development

Control Suggestions

Selected References

Genetics

Currently there are no related genes or alleles for this strain.

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

CONTACT TECHNICAL SUPPORT

Genotyping Protocols
Sanger sequencing:Npc1
Standard PCR:Npc1
Genotyping resources and troubleshooting

Breeding Considerations

When maintaining a live colony, heterozygous mice may be bred together. Homozygous mice have reduced lifespan, with an average survival of 125 days. The donating investigator has not attempted to breed homozygous mice.

Additional Breeding and Husbandry Support

Citation
When using the Npc1$^{tm(I1061T)Dso}$ mouse strain in a publication, please cite the originating article(s) and include JAX stock #027704 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 or wildtype for Npc1<tm1Dso>	$2,854.50

RELATED PRODUCTS AND SERVICES

Frozen Mouse Embryo	B6.129-Npc1<tm1Dso>/J Frozen Embryo	$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

ADDITIONAL USE RESTRICTIONS APPLY

Use of MICE by companies or for-profit entities requires a license prior to shipping.

LICENSING INFORMATION

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

Related Strains

All

By Allele

By Gene

By Collection

Choose other country or region

Did you find what you were looking for?

Yes No