C57BLKS/J-Npc1<sup>spm/J</sup>

Stock No: 002760 | sphingomyelinosis

- Spontaneous Mutation

PLACE ORDER

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

Overview

Also Known As: sphingomyelinosis

This spontaneous mutation provides a model for Niemann-Pick disease types C1 and D, with severe Purkinje cell loss. Homozygotes on this C57BLKS/J background live longer than mice homozygous for this mutation on a DBA/2J or C57BL/6J background but develop a massive hepatomegaly not seen on these other backgrounds.

READ MORE +
**Npc1<sup>spm</sup>**

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<tr>
<th>Allele Type</th>
<th>Gene Symbol</th>
<th>Gene Name</th>
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<tr>
<td>Spontaneous</td>
<td>Npc1</td>
<td>NPC intracellular cholesterol transporter 1</td>
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**RESEARCH APPLICATIONS**

- Metabolism Research
- Internal/Organ Research
- Neurobiology Research
- Mouse/Human Gene Homologs

**BASE PRICE**

Starting at:

$2,854.50 Domestic price Cryo Recovery

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**Details**

**Detailed Description**

NPC1 is a transmembrane protein involved in the transport and export of LDL-derived cholesterol and other lipids from late endosomes and lysosomes. NPC1 Defects cause Niemann-Pick disease types C1 and D, lysosomal storage diseases in which cholesterol and other lipids accumulate in lysosomes of many organs. On this C57BLKS/J background sphingomyelinosis (spm) homozygotes begin to display weight loss and progressive neurological symptoms, such as tremor and ataxia, at approximately 7 weeks of age and die between 11 and 15 weeks of age with mean longevity of approximately 85 days for females and 90 days for males. Hepatosplenomegaly is found as early as 4 weeks of age, and peaks at 6 weeks of age just before noticeable weight loss. Massive foam cell clusters are found in the liver and spleen, and the liver has irregularly shaped white areas while the spleen has a whitish color. The lymph nodes are enlarged, including the mesenteric, inguinal and submandibular lymph nodes. (Miyawaki et al., 1982, 1986)

Beginning at 4 weeks of age, neuronal swelling is found in the neocortex, piriform cortex, hippocampus and basal ganglia and this spreads with age to include the thalamus, midbrain, cerebellum and brainstem by 11 to 12 weeks of age. Neuronal cell loss is restricted to the Purkinje cell layer and the ventral posterior medial and lateral nuclei of the thalamus, with the thalamic neurons degenerating between 4 and 13 weeks of age and the Purkinje cell loss delayed to 5 weeks of age and nearly complete loss by 9 weeks of age (Yamada et al., 2001). Purkinje cell loss begins with zebrin II negative Purkinje cells, which results in a stripping pattern of cell loss. At 45 days of age there is significant loss in both the posterior lobe, and anterior lobe, by 60 days of age there is further loss, especially in the posterior lobe hemispheres, and by 90 days of age most Purkinje cells are gone, but some remain in the
flocculus/paraflocculus, lobules IX and X of the nodular zone, and 3 stripes remain in the anterior zone. These sphingomyelinosis homozygotes have a faster, more severe Purkinje cell loss than do Npc1<sup>1<sup>M1</sup>N</sup> homozygotes on a BALB/c background, but survive approximately 15 days longer (Sarna et al., 2003).

Genetic background impacts the phenotype for this mutation. On this C57BLKS/J background the foam cells stain slightly reddish with H&E stain, but on a DBA/2J congenic background the foam cells appear vacuolated, the massive hepatomegaly is not seen, weight loss begins just after 6 weeks of age and is faster with most dying between 9 and 10 weeks of age and the mean longevity for females and males is approximately 66 days. On a C57BL/6J congenic background homozygotes have a phenotype closer to that found in DBA/2J than in C57BLKS/J. (Miyawaki et al., 1986)

Sphingomyelinosis homozygotes fail to breed, but homozygous ovarian transplants and in vitro fertilization using sperm from homozygotes have each been used successfully for this strain.

Genotyping Protocols
Pyrosequencing: Npc1<sup>1<sup>spm</sup></sup>
Standard PCR: Npc1<sup>1<sup>spm</sup></sup>
Genotyping resources and troubleshooting
Breeding Considerations
Sphingomyelinosis homozygotes fail to breed, but homozygous ovarian transplants and in vitro fertilization using sperm from homozygotes have each been used successfully for this strain.

Additional Breeding and Husbandry Support

Appearance
black, ataxic, tremors
Related Genotype: a/a Npc<sup>spm</sup>/Npc<sup>spm</sup>

black, unaffected
Related Genotype: a/a Npc<sup>spm</sup>/+ or a/a +/-

Citation
We recommend the sphingomyelinosis mouse strain in a publication, please cite the originating article(s) and include JAX stock #002760 in your Materials and Methods section.

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

Pricing & Availability

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

<table>
<thead>
<tr>
<th>Domestic</th>
<th>International</th>
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<td>Pricing effective for USA, Canada and Mexico shipping destinations</td>
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<tr>
<th>SERVICE</th>
<th>GENOTYPE</th>
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<tr>
<td>Cryo Recovery</td>
<td>Heterozygous for Npc&lt;sup&gt;spm&lt;/sup&gt;</td>
<td>$2,854.50</td>
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We will fulfill your order by providing at least two carriers for each strain ordered. The total number, sex, and genotypes provided will vary, although typically 8 or more animals are provided. Please check genotypes which will be recovered. While the genotypes of all animals produced will be communicated to you prior to scheduling shipment, the genotypes of animals provided may not reflect the mating scheme and genotypes described in the strain description. Animals are typically ready to ship in 11-14 weeks. If a second recovery is required to produce the minimum number of animals, then delivery time would increase to approximately 25 weeks. If we fail to produce animals of the correct genotype, you will not be charged. We cannot guarantee the reproductive success of mice shipped to your facility. If the mice are lost after the first three days (post-arrival) or do not produce progeny at your facility, a new order and fee will be necessary.

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