BALB/cNctr-Npc1<sup>m1N</sup>/J

Stock No: 003092 | npc<sup>nih</sup>

Spontaneous Mutation

Overview

Also Known As: npc<sup>nih</sup>

Mice homozygous for the recessive NIH allele of the Niemann Pick type C1 gene (Npc1<sup>m1N</sup>) show a dual deficiency of sphingomyelinase and glucocerebrosidase activity. The overall phenotype resembles the sphingomyelinosis condition seen in mice homozygous for the sphingomyelinosis allele (Npc1<sup>spm</sup>, Stock No. 002760). Sphingomyelinosis closely resembles that of human Niemann-Pick Type C disease patients.

Genetic overview

<table>
<thead>
<tr>
<th>Genetic Background</th>
<th>Generation</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>?+F44</td>
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<tr>
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<td>(19-JUL-17&lt;)</td>
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</tbody>
</table>

Npc1<sup>m1N</sup>

<table>
<thead>
<tr>
<th>Allele Type</th>
<th>Gene Symbol</th>
<th>Gene Name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spontaneous</td>
<td>Npc1</td>
<td>NPC intracellular cholesterol:transporter 1</td>
</tr>
</tbody>
</table>

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