Overview

In mice homozygous for Nr2e3<sup>mut</sup>, evenly distributed white spots cover the retina and have been detected by Fundus examination as early as 16.5 days of age. Nr2e3 is a retinal transcription factor important in the developmental pathways of photoreceptor cells. Enhanced S-cone syndrome has been associated with mutations in human NR2E3 and mice homozygous for the Nr2e3<sup>mut</sup> mutation offer a model for this disease.

Genetic Overview

<table>
<thead>
<tr>
<th>Genetic Background</th>
<th>Generation</th>
</tr>
</thead>
<tbody>
<tr>
<td>000664 C57BL/6J</td>
<td>N8F16</td>
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<td>(2017-01-04 00:00:00)</td>
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</tbody>
</table>
**Details**

**Detailed Description**

Nr2e3 is a retinal transcription factor important in the developmental pathways of photoreceptor cells. In mice homozygous for Nr2e3<sup>3rd7</sup>, evenly distributed white spots cover the retina and have been detected by Fundus examination as early as 16.5 days of age. Whorls and rosettes in the outer nuclear layer can first be detected at 12.5 days of age, before the eyes open. These whorls likely underlie the appearance of the white spots on the retina and the white spots and whorls are both present at one month of age then are reduced in number by 5 months, and disappear by 16 months. Electroretinographs give normal signals until 5 months of age when both rod and cone signals begin to show a progressive reduction. Attenuated retinal vessels and mottled pigment are found by 16 months of age, and the outer nuclear layer is only half normal thickness subsequent to progressive loss of cones and rods. Immunohistochemical assessment revealed that the whorls are filled with and surrounded by cone cells and there is an increase in the percentage of blue opsin expressing cone cells. Thus, NR2E3 regulates photoreceptor cell differentiation. Enhanced S-cone syndrome has been associated with mutations in human NR2E3 and mice homozygous for the Nr2e3<sup>3rd7</sup> mutation offer a model for this disease. (Chang et al., 1998; Akhmedov et al., 2000; Haider et al., 2000 and 2001.)

**Development**

**Control Suggestions**

**Genetics**

**Nr2e3<sup>3rd7</sup>**
Genotyping Protocols
Genotyping resources and troubleshooting

Mating System
Homozygote x Homozygote

Appearance
black, retinal degeneration
Related Genotype: a/a Nr2e3<sup>+/</sup>/Nr2e3<sup>+/</sup>

Citation
When using the B6.Cg-Nr2e3<sup>+/</sup> J mouse strain in a publication, please cite the originating articles and include JAX stock #004643 in your Materials and Methods section.
Facility Barrier Level Descriptions
M6L.277 (Low)

Pricing & Availability

<table>
<thead>
<tr>
<th>Live Mouse</th>
<th>Pricing effective for USA, Canada and Mexico shipping destinations</th>
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</thead>
<tbody>
<tr>
<td>AGE</td>
<td>SEX</td>
</tr>
<tr>
<td>Approx 4-8 weeks</td>
<td>Female</td>
</tr>
<tr>
<td></td>
<td>Male</td>
</tr>
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</table>

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- By Allele
- By Gene
- By Collection

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