Overview

Also Known As: early-onset glaucoma 1
This R115L point substitution provides a fully penetrant model for early-onset glaucoma, with elevated intraocular pressure by five weeks of age.

GENETIC OVERVIEW

<table>
<thead>
<tr>
<th>Genetic Background</th>
<th>Generation</th>
</tr>
</thead>
<tbody>
<tr>
<td>C57BL6J x gl2 Boc</td>
<td>N1F32</td>
</tr>
<tr>
<td></td>
<td>(2019-01-29 00:00:00)</td>
</tr>
</tbody>
</table>

\( Pitx2^{egl1} \)

<table>
<thead>
<tr>
<th>Allele Type</th>
<th>Gene Symbol</th>
<th>Gene Name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chemically induced (ENU) (Not Specified)</td>
<td>Pitx2</td>
<td>paired-like homeodomain transcription factor 2</td>
</tr>
</tbody>
</table>
Mice homozygous for the \( \text{Pitx2}^{\text{R115L}} \) point substitution on this C57BL/6J background provide a fully penetrant model for early-onset glaucoma, developing elevated intraocular pressure by five weeks of age, which subsequently becomes more severe. The size of the eye is similar to that of controls until 13 weeks of age when the eyes are larger. Image guided optical coherence tomography at three months of age shows enlargement of the anterior chamber and optic cupping, which presents as a ring-like feature around the optic nerve head by indirect ophthalmoscopy. Histology at 3 months of age shows optic head cupping, anterior synechia and a complete loss of the inner retinal layers. Functionally, at three months of age the rod a-wave is normal but there is a loss of rod b-waves and lower cone electroretinogram response. This ENU-induced mutation was mapped between D3Mit106 and D3Mit291 and found to be a single G to T transversion in \( \text{Pitx2} \) exon 2 that changes amino acid 115 from arginine to leucine.
Genotyping Protocols
Genotyping resources and troubleshooting

Mating System
Homozygote x Homozygote

Citation
When using the early-onset glaucoma 1 mouse strain in a publication, please cite the originating article(s) and include JAX stock #004240 in your Materials and Methods section.

Pricing & Availability

Domestic
International
Pricing effective for USA, Canada and Mexico shipping destinations

<table>
<thead>
<tr>
<th>AGE</th>
<th>SEX</th>
<th>GENOTYPE</th>
<th>PRICE</th>
</tr>
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<tbody>
<tr>
<td>Approx 4-8 weeks</td>
<td>Female</td>
<td>homozygous for Pitx2&lt;sup&gt;egl1&lt;/sup&gt;</td>
<td>$255.00</td>
</tr>
<tr>
<td></td>
<td>Male</td>
<td>homozygous for Pitx2&lt;sup&gt;egl1&lt;/sup&gt;</td>
<td>$255.00</td>
</tr>
</tbody>
</table>

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All

By Allele

By Gene

By Collection

All Related Strains