

C57BL/6J-*Pitx2*^{egl1}/Boc

Stock No: 004240 | early-onset glaucoma 1

 Chemically Induced Mutation, Coisogenic

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

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GENETIC OVERVIEW

Genetic Background

Generation

N1F36
(2020-05-07 00:00:00)

Pitx2^{egl1}

Alele Type

Chemically induced (ENU)
(Not Specified)

Gene Symbol

Pitx2

Gene Name

paired-like homeodomain transcription factor 2

VIEW GENETICS

RESEARCH APPLICATIONS

Sensorineural Research

VIEW ALL RESEARCH APPLICATIONS

BASE PRICE

Starting at:

\$305.00 Domestic price for female

V I E W P R I C E L I S T

Details

Detailed Description

Mice homozygous for the *Pitx2*^{egl1} 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 *Pitx2* exon 2 that changes amino acid 115 from arginine to leucine.

Development

Control Suggestions

Selected References

Genetics

Pitx2^{egl1}

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

- Technical Support

C O N T A C T T E C H N I C A L S U P P O R T

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.

Animal Health Reports
[Facility Barrier Level Descriptions](#)

 [MGL277 \(Low\)](#)

- Pricing & Availability



Availability
Varies

Domestic **International**

Pricing effective for USA, Canada and Mexico shipping destinations

Domestic Pricing for

- Commercial & For-Profit
- Not-For-Profit & Academic

LIVE MOUSE

AGE	SEX	GENOTYPE	PRICE
Approx 4-8 weeks	Female	Homozygous for Pitx2 ^{egl1}	\$305.00
	Male	Homozygous for Pitx2 ^{egl1}	\$305.00

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LICENSING INFORMATION

Phone: 207-288-6470

Email: TechTran@jax.org

Related Strains

All

By Allele

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