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gk^{A456V} - Mouse Strain RES191

Mouse Information

Common Name:	gk^{A456V}
MGI Official Name:	Gck^{tm3Mgn}
Description:	This line of mice provides a model for Persistant Hyperinsulinemic Hypoglycemia of Infancy, or PHHI-GK, a rare genetic disease of humans. A gk^{A456V} mutation, originally identified in a human pedigree with PHHI-GK, was introduced into these mice by gene knock-in. These mice may be useful for studies of sustained hypoglycemia. The mutation has been bred into a C57BL/6J strain thereby facilitating direct comparisons to both wild type C57BL/6J animals and to animals with a gk^{K414E} mutation.
Categories:	None specified.

Genetic Alterations

1) Targeted Mutagenesis	
Type of Allele	Global Mutation
Targeted Gene	Glucokinase (Gck - NCBI GeneID:103988)
Targeted Allele	targeted mutation 3 (Gck^{tm3Mgn} - MGI:3701764)
Description of Targeting Vector	A single base mutation was introduced into exon 10 via site specific mutagenesis to change amino acid 456 from alanine to valine. Genotype by DNA PCR using primers 5'-TGT CTC AAT TTG CTG TGT CCT CCA-3' and 5'-ATG TGT GAG TGT GCC AAT ATG AGT-3'. These primers will amplify a 636 bp fragment from the wild type allele and a 741 bp fragment from the mutant allele. Homozygous mutant mice, which have a phenotype of moderate hypoglycemia, are viable and breed well. Heterozygous animals are mildly hypoglycemic.
Targeting Vector Genbank File	pBOB2.A456V.gb

Citations

PubMedID	Citation
17353190	Glucokinase thermolability and hepatic regulatory protein binding are essential factors for predicting the blood glucose phenotype of missense mutations. (2007) *J Biol Chem* **282**: 13906-16 (Added 2008-03-29 16:59:08)

Strain Information

Strain Type:	Congenic Strain
Chimera/Founder Genetic Background:	129S6/SvEvTac
Current Genetic Background:	C57BL/6J (date recorded: Not provided)
Strain Description:	After achieving germline transmission mice carrying the mutant gk^{A456V} allele were bred to EIIa-Cre transgenic mice in order to delete a neomycin resistance (neoR) cassette. Mice lacking neoR were then backcrossed for a total of twelve generations with C57BL/6J mice to obtain a congenic line.

Access Status

This resource is publicly viewable.

Request this Resource

Request from a **repository**

Primary contributor: **Magnuson Lab**

Resource Tags

Gck, gk^{A456V}, mouse, mouse strain

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Resource History & Actions

Approved on Feb 02, 2007
Last modified on Nov 02, 2011

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Related resources

BCBC

No matching resources

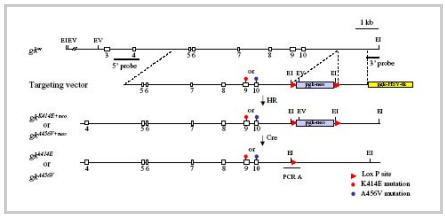
Other Consortia

No matching resources

Data courtesy of dkCOIN. Only public resources are displayed.

Associated Images

Image 1



Description:
Gene targeting was used to introduce a point mutation in the gk gene. Uppermost map is a diagram of the mouse gk gene showing locations of exons 3 to 10 (indicated by open boxes). The locations of the DNA fragments used as 5' and 3' hybridization probes are shown. EI, EcoRI; EV, EcoRV. Second map from top is of the gene targeting vector carrying either the K414E mutation in exon 9 depicted by the red circle or the A456V mutation in exon 10 as indicated by the blue circle. A neomycin resistance cassette (pgk-neoR), which is flanked with two loxP sites depicted by red triangles, and a HSV-thymidine kinase cassette (pgk-HSV-TK), were used for positive and negative selection, respectively. Third map from top is of the recombinant gk allele after homologous recombination (HR) carrying a floxed pgk-neoR cassette and the respective point mutation in exon 9 or 10. Fourth map from top is of the mutant gk allele after Cre recombination.

Reference:
17353190

Repositories

MMRRC

Request via www.mmrrc.org website

Stock #: 015249-UCD
Availability Notes: *Not provided*

BCBC members may Login to request this resource.

Contact Information

Preferred Contact

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Associated Publications

No publications associated

Comments

There are no comments for this entry.

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