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**gk<sup>A456V</sup> - Mouse Strain RES191****Mouse Information**

<b>Common Name:</b>	gk <sup>A456V</sup>
<b>MGI Official Name:</b>	Gck <sup>tm3Mgn</sup>
<b>Description:</b>	This line of mice provides a model for Persistent Hyperinsulinemic Hypoglycemia of Infancy, or PHHI-GK, a rare genetic disease of humans. A gk <sup>A456V</sup> 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 <sup>K414E</sup> mutation.
<b>Categories:</b>	None specified.

**Genetic Alterations**


<b>1) Targeted Mutagenesis</b>	
<b>Type of Allele</b>	Global Mutation
<b>Targeted Gene</b>	Glucokinase (Gck - <a href="#">NCBI GeneID:103988</a> )
<b>Targeted Allele</b>	targeted mutation 3 (Gck <sup>tm3Mgn</sup> - <a href="#">MGI:3701764</a> )
<b>Description of Targeting Vector</b>	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.
<b>Targeting Vector Genbank File</b>	<a href="#">pBOB2.A456V.gb</a>

<b>Citations</b>	<table border="1"> <thead> <tr> <th>PubMedID</th> <th>Citation</th> </tr> </thead> <tbody> <tr> <td><a href="#">17353190</a></td> <td><a href="#">Glucokinase thermolability and hepatic regulatory protein binding are essential factors for predicting the blood glucose phenotype of missense mutations.</a> (2007) <i>J Biol Chem</i> <b>282</b>: 13906-16 (Added 2008-03-29 16:59:08)</td> </tr> </tbody> </table>	PubMedID	Citation	<a href="#">17353190</a>	<a href="#">Glucokinase thermolability and hepatic regulatory protein binding are essential factors for predicting the blood glucose phenotype of missense mutations.</a> (2007) <i>J Biol Chem</i> <b>282</b> : 13906-16 (Added 2008-03-29 16:59:08)
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**Strain Information**

<b>Strain Type:</b>	Congenic Strain
<b>Chimera/Founder Genetic Background:</b>	129S6/SvEvTac
<b>Current Genetic Background:</b>	C57BL/6J (date recorded: Not provided)
<b>Strain Description:</b>	After achieving germline transmission mice carrying the mutant gk <sup>A456V</sup> allele were bred to Ella-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**


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Primary contributor: [Magnuson Lab](#)

**Resource Tags**

Gck, gk<sup>A456V</sup>, 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

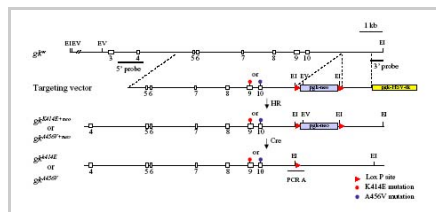
**Other Consortia**

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.mmrc.org website](http://www.mmrc.org)

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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