

These mice, which contain an activating, A456V mutation within the Gck gene, provide a model for Persistent Hyperinsulinemic Hypoglycemia of Infancy, or PHHI-GK, a rare genetic disease of humans.

During the generation of this line, male chimeric mice derived by injecting correctly targeted TL1 mESCs into blastocysts from C57Bl/6 animals were mated directly with 129S6 female mice, thereby maintaining the mutation in an inbred state.

Cryostocks of both 8-cell embryos and sperm from this line exist only at the VCMR.

Keywords: [glucokinase](#) [Gck](#) [PHHI](#) [gck^{A456V}](#) [Mgn](#)

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Mouse Information

Common Name	gk ^{A456V}
Research Applications	<i>Not provided</i>
MMRRC ID	<i>Not provided</i>
Jackson Laboratories Stock No	<i>Not provided</i>
VCMR ID	DE, DS
Additional Strain Information	<i>Not provided</i>

Genetic Alteration

Mutation #1: Targeted Mutagenesis	
Type of Allele	Global Mutation
Targeted Gene	Name: Glucokinase Symbol: Gck NCBI: 103988
Allele	Name: glucokinase; targeted mutation 3, Mark A Magnuson Symbol: Gck ^{tm3Mgn} MGI: 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.
Vector Genbank File	pBOB2.A456V.gb

Allele Map	<i>Not Provided</i>
PCR Genotyping Protocol	Gck.A456V_PCR_genotyping_protocol.docx
Citations	<p>Publication</p> <p><u>Glucokinase thermolability and hepatic regulatory protein binding are essential factors for predicting the blood glucose phenotype of missense mutations.</u> (2007) <i>J Biol Chem</i> 282: 13906-16 (Added 12/10/2013) PMID: 17353190</p>

Background Strain Information

Strain Type	Congenic Strain
Chimera/Founder Genetic Background	129S6/SvEvTac
Current Genetic Background	<i>Not provided</i>
Number of Generations Backcrossed	>10
Strain Description	<p>Need to check how neo cassette was removed.</p> <p>129SvEv at cryopreservation</p> <p>Cryopreserved in 2005.</p> <p>Vial DE - cryopreserved embryos at the 8 cell stage</p> <p>Vial DS - cryopreserved sperm in vials</p>

Attachments

 [gka456v.neo_wt_protocol.doc](#) - Added on July 27, 2010 at 10:38 AM by Jill Lindner

PCR protocol for genotyping mice

 [K414EA456V_targeting_vector.jpg](#) - Added on July 19, 2010 at 10:16 AM by Mark Magnuson

K414E and A456V targeting vectors



