The Jackson Laboratory

B6.Cg-*Spef2* ^{bgh} /RcMdfLleeJ

Stock No: **027799** | big giant head (bgh)

Chemically Induced Mutation, Congenic

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

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primary ciliary dyskinesia (PCD; including hydrocephalus, sinusitis and male infertility), as well as reduced respiratory ciliary motility. These C57BL/6J-congenic bgh mice (B6J.bgh) may be useful in studying PCD, as well as the role of mucociliary clearance in host defense.

Donating Investigator

Lance Lee, Sanford Children's Health Research Center (Sanford Research Center)

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GENETIC OVERVIEW

Genetic Background	Generation

Spef2^{bgh}

Allele Type	Gene Symbol	Gene Name
Chemically induced (ENU) (Null/Knockout)	*Spef2*	sperm flagellar 2

VIEW GENETICS

RESEARCH APPLICATIONS

Developmental Biology Research
Cell Biology Research
Research Tools
Immunology, Inflammation and Autoimmunity Research
Reproductive Biology Research

VIEW ALL RESEARCH APPLICATIO

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

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Details

Detailed Description

Mice homozygous for the "big giant head" mutation (*bgh/bgh*; *Spef2*$^{bgh/bgh}$) have several abnormalities commonly associated with primary ciliary dyskinesia (PCD); including hydrocephalus, sinusitis and male infertility. Homozygous mice do not exhibit *situs inversus*. Heterozygous mice are viable and fertile with no abnormal phenotype. The absence of full-length protein was confirmed by western blot analysis on testis extracts from homozygotes.

The donating investigator has assessed homozygous phenotype on three different backgrounds: C57BL/6J-congenic (B6J.bgh/bgh), 129S6/SvEvTac-congenic (129S6.bgh/bgh) and an F1 cross of these two backgrounds ((B6x129)F1.bgh/bgh). Overall, the severity of morphological brain damage is background strain-dependent. Alterations in astrocytosis, microglial activation, myelination, and the neuronal population were identified and are generally more severe on the C57BL/6J background. Specific details follow.

B6J.bgh/bgh mice exhibit lethal hydrocephalus (average death ~ 1 month of age) with damage to multiple cell types, as well as cilia-driven cerebrospinal fluid flow deficit, sinusitis and male infertility. B6J.bgh/bgh also exhibit upper airway abnormalities (reduced respiratory ciliary motility, but no ultrastructural defects); although no major cellular, developmental or inflammatory abnormalities in the lower airway are reported. The donating investigator reports that B6J.bgh/bgh mice do not exhibit *situs inversus*. Heterozygous mice (B6J.bgh/+) are viable and fertile with no abnormal phenotype.

129S6.bgh/bgh mice do not develop gross hydrocephalus or exhibit early mortality, although they have cilia-driven cerebrospinal fluid flow deficits and variable levels of ventricular enlargement and brain damage. The sinusitis phenotype and male fertility phenotype for 129S6.bgh/bgh mice has not been characterized to date (although the donating investigator expects it to be similar to B6J.bgh/bgh animals). The specific upper airway vs. lower airway phenotype described for B6J.bgh/bgh has not been tested in 129S6.bgh/bgh to date (July 2015). The donating investigator reports that 129S6.bgh/bgh mice do not exhibit *situs inversus*. Heterozygous mice (129S6.bgh/+) are viable and fertile with no abnormal phenotype.

(B6x129)F1.bgh/bgh mice exhibit sinusitis and male infertility, but no evidence of gross hydrocephalus or early mortality. (B6x129)F1.bgh/bgh also exhibit upper airway abnormalities (reduced respiratory ciliary motility, but no ultrastructural defects); although no major cellular, developmental or inflammatory abnormalities in the lower airway are reported. The infertility in (B6x129)F1.bgh/bgh males results from reduction in the number of elongating spermatids during spermiogenesis and structural defects in sperm flagella (this could not be tested directly in B6J.bgh/bgh males because they die before sexual maturity). The donating investigator reports that (B6x129)F1.bgh/bgh mice do not exhibit *situs inversus*. Heterozygous mice ((B6x129)F1.bgh/+) are viable and fertile with no abnormal phenotype. Compared to similarly treated (B6x129)F1 wildtype mice, the (B6x129)F1.bgh/bgh mice show increased lymphocytic response when challenged with *Streptococcus pneumonia* infection.

Development

Control Suggestions

Selected References

Genetics

Spef2^{bgh}

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

CONTACT TECHNICAL SUPPORT

Genotyping Protocols

Sanger sequencing:Spef2
Genotyping resources and troubleshooting

Breeding Considerations

Homozygous mice on the C57BL/6J genetic background (B6J.bgh/bgh) exhibit lethal hydrocephalus (average death ~ 1 month of age) and male infertility. Therefore, when maintaining a live colony, heterozygous mice may be bred together, to wildtype mice from the colony or to C57BL/6J inbred mice (Stock No. 000664).

Additional Breeding and Husbandry Support

Mating System

Wild-type x Heterozygote
Heterozygote x Wild-type

Citation

When using the big giant head (bgh) mouse strain in a publication, please cite the originating article(s) and include JAX stock #027799 in your Materials and Methods section.

Animal Health Reports

Facility Barrier Level Descriptions

Production of mice from cryopreserved embryos or sperm occurs in a maximum barrier room, G200

Pricing & Availability

Cryo Recovery

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

Domestic **Internatic**

Pricing effective for USA, Canada and Mexico shipping destinations

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	Heterozygous or wildtype for Spef2<bgh>	$2,854.50

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The Jackson Laboratory has rigorous genetic quality control and mutant gene genotyping programs to ensure the genetic background of JAX® Mice strains as well as the genotypes of strains with identified molecular mutations. JAX® Mice strains are only made available to researchers after meeting our standards. However, the phenotype of each strain may not be fully characterized and/or captured in the strain data sheets. **Therefore, we cannot guarantee a strain's phenotype will meet all expectations.** To ensure that JAX® Mice will meet the needs of individual research projects or when requesting a strain that is new to your research, we suggest ordering and performing tests on a small number of mice to determine suitability for your particular project. We do not guarantee breeding performance and therefore suggest that investigators order more than one breeding pair to avoid delays in their research.

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