

B6.129S2-Ampd2^{tm1Tm}/J

Stock No: **024913**

 Congenic, Targeted Mutation

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

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seen.

Donating Investigator

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GENETIC OVERVIEW

Genetic Background

Generation

Ampd2^{tm1Tm}

Alele Type

Targeted (Null/Knockout)

Gene Symbol

Ampd2

Gene Name

adenosine monophosphate deaminase 2

VIEW GENETICS

RESEARCH APPLICATIONS

Neurobiology Research

Cell Biology Research

Internal/Organ Research

VIEW ALL RESEARCH APPLICATIONS

BASE PRICE

Starting at:

\$2,854.50 Domestic price Cryo Recovery

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

Details

Detailed Description

Ampd2 (adenosine monophosphate deaminase 2) plays a role in the maintenance of cellular guanine nucleotide pools by regulating the feedback inhibition of adenosine derivatives on de novo purine synthesis. Mutations in the gene have been associated with pontocerebellar hypoplasia (PCH), a rare, inherited, progressive neurodegenerative disorder with prenatal onset that is associated with a loss of brainstem and cerebellar parenchyma.

Mice homozygous for this *Ampd2* knock-out allele show no histological neuronal phenotype, and are viable and fertile. A significant increase in AMP and decrease in ATP and GTP are observed in the kidneys of knockout mice at 12-24 weeks of age. In addition to changes in nucleotide metabolism in the kidneys, proteinuria is discernable at 3 weeks of age, increasing to transient peak levels at 6 weeks of age. Ultrastructural studies of glomerular specimens show effacement of the podocyte processes, resembling minimal-change nephropathy in humans.

Pups homozygous for both this *Ampd2* mutation and a knock-out of *Ampd3* (*Ampd3*^{tm2a(KOMP)Wtsi}, available from the [KOMP Repository](#)) are born in the expected Mendelian ratio, but show slightly reduced body weight and have a severely shortened lifespan. Most die by postnatal day 21. Although the brain of newborns is slightly reduced in size, double knock-out mice show little evidence of the characteristic neuronal loss observed in AMPD2 mutant patients. A neurodegenerative phenotype is observed, mostly affecting the CA3 pyramidal neurons in the hippocampus and some sparse pyknotic cells all over the cortex and cerebellum. This is associated with a behavioral gait disturbance after postnatal day 10. Brains of 2 week old double knock-out animals show a 25% increase in ATP nucleotide levels and a 33% decrease in GTP levels, as compared to wildtype.

Development

Control Suggestions

Selected References

Genetics

Ampd2^{tm1Tm}

⊖ Disease/Phenotype

[+ Disease Terms](#)

[+ Research Areas By Phenotype](#)

[+ Mammalian Phenotype Terms by Genotype](#)

[+ References](#)

⊖ 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

Separated PCR:[Ampd2](#)

[Genotyping resources and troubleshooting](#)

Breeding Considerations

Heterozygotes and homozygotes are viable and fertile.

[Additional Breeding and Husbandry Support](#)

Citation

When using the B6.129S2-*Ampd2*^{tm1Tm}/J mouse strain in a publication, please [cite the originating article\(s\)](#) and include JAX stock #024913 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



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Domestic International

Pricing effective for USA, Canada and Mexico shipping destinations

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	Heterozygous or wildtype for -Ampd2<tm1Tm>	\$2,854.50

RELATED PRODUCTS AND SERVICES

Frozen Mouse Embryo	B6.129S2-Ampd2<tm1Tm>/J Frozen Embryo	\$2595.00
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THE JACKSON LABORATORY'S GENOTYPE PROMISE

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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