

C.129-*Mthfr*^{tm1Rzn}/J

Stock No: **024503**

 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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Donating Investigator

Rima Rozen, McGill University Health Centre

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GENETIC OVERVIEW

Genetic Background

Generation

Mthfr^{tm1Rzn}

Alele Type

Gene Symbol

Gene Name

Targeted (Null/Knockout)

Mthfr

methylenetetrahydrofolate reductase

VIEW GENETICS

RESEARCH APPLICATIONS

Cell Biology Research

Metabolism Research

Cardiovascular Research

Developmental Biology 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

Mthfr KO mice have a neo cassette disrupting exon 3 of the 5,10-methylenetetrahydrofolate reductase (*Mthfr*) gene, abolishing gene expression. MTHFR is the rate-limiting enzyme in the methyl cycle. It is involved in the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, which is a cosubstrate for homocysteine remethylation to methionine. Homozygous KO of MTHFR causes an abnormally high level of homocysteine in blood and urine, called homocysteinemia. Patients with this disorder exhibit developmental delay, thrombosis, motor and gait dysfunction, seizures, and other neurological symptoms. Mild MTHFR deficiency due to polymorphic variation causes a more modest elevation of plasma homocysteine (mild hyperhomocysteinemia) which is associated with increased risk for some common multifactorial disorders. Homozygous *Mthfr* KO mice low viability, with 25% surviving at 5 weeks of age. They have a 10 fold increase in plasma homocysteine levels. They are smaller and show developmental retardation with cerebellar pathology, abnormal lipid deposition in the proximal portion of the aorta, kinked tail, kyphosis, facial anomalies, and liver steatosis. Heterozygotes are normal in size and survival, and have a 1.6 fold increase in plasma homocysteine levels over wildtype. Heterozygotes are often studied as a model for mild MTHFR deficiency. These mice on a BALB/cJ background have a more deleterious phenotype than on a C57BL/6NJ background (Stock No. [024316](#)).

Development

Control Suggestions

Selected References

Genetics

Mthfr^{tm1Rzn}

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

Standard PCR:[Mthfr](#)

[Genotyping resources and troubleshooting](#)

Breeding Considerations

When maintaining a live colony, heterozygous mice may be bred together. The donating investigator reports that only 25% of homozygotes survive to 5 weeks of age.

[Additional Breeding and Husbandry Support](#)

Citation

When using the C.129-*Mthfr*^{tm1Rzn}/J mouse strain in a publication, please [cite the originating article\(s\)](#) and include JAX stock #024503 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 **International**

Pricing effective for USA, Canada and Mexico shipping destinations

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	Heterozygous or wildtype for Mthfr<tm1Rzn>	\$2,854.50

RELATED PRODUCTS AND SERVICES		
Frozen Mouse Embryo	C.129-Mthfr<tm1Rzn>/J Frozen Embryo	\$2595.00

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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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Q U E S T I O N S A B O U T T E R M S O F U S E

LICENSING INFORMATION

Phone: 207-288-6470
 Email: TechTran@jax.org

Related Strains

All

By Allele

By Gene

By Collection



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