Overview

*Mthfr* KO mice may be useful for studying DNA methylation control.

Donating Investigator

Rima Rozen, McGill University Health Centre
### GENETIC OVERVIEW

**Genetic Background**

<table>
<thead>
<tr>
<th>Allele Type</th>
<th>Gene Symbol</th>
<th>Gene Name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Targeted (Null/Knockout)</td>
<td>Mthfr</td>
<td>methylenetetrahydrofolate reductase</td>
</tr>
</tbody>
</table>

### RESEARCH APPLICATIONS

Cell Biology Research  
Metabolism Research  
Cardiovascular Research  
Developmental Biology Research

### BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

### 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 have 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
Genotyping Protocols
Standard PCR: Mthfr<sup>tm1Rzn</sup>
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<sup>tm1Rzn</sup>/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.
We will fulfill your order by providing at least two carriers for each strain ordered. The total number, sex, and genotypes provided will vary, although typically 8 or more animals are provided. Please check genotypes which will be recovered. While the genotypes of all animals produced will be communicated to you prior to scheduling shipment, the genotypes of animals provided may not reflect the mating scheme and genotypes described in the strain description. Animals are typically ready to ship in 11-14 weeks. If a second recovery is required to produce the minimum number of animals, then delivery time would increase to approximately 25 weeks. If we fail to produce animals of the correct genotype, you will not be charged. We cannot guarantee the reproductive success of mice shipped to your facility. If the mice are lost after the first three days (post-arrival) or do not produce progeny at your facility, a new order and fee will be necessary.

Cryorecovery to establish a Dedicated Supply for greater quantities of mice. Mice recovered can be used to establish a dedicated colony to contractually supply you mice according to your requirements. Price by quotation.

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