Also Known As: thrombocytopenia and cardiomyopathy

These mice carry a spontaneous mutation at the Abcg5 locus characterized by macrothrombocytopenia, prolonged bleeding times, anemia, leukopenia, infertility, cardiomyopathy, and shortened life span.
The spontaneous mouse mutation 'thrombocytopenia and cardiomyopathy (Abcg5\textsuperscript{frac})' causes macrothrombocytopenia, prolonged bleeding times, anemia, leukopenia, infertility, cardiomyopathy, and shortened life span. Homozygotes show a 20-fold decrease in platelet numbers and a 3-fold increase in platelet size with structural alterations and functional impairments in activation and aggregation. Megakaryocytes in homozygous mice are present in increased numbers, have poorly developed demarcation membrane systems, and have decreased polyploidy. The thrombocytopenia is not intrinsic to defects at the level of hematopoietic progenitor cells but is associated with a microenvironmental abnormality. The Abcg5\textsuperscript{frac} mutation maps to mouse chromosome 17, syntenic with human chromosome 2p21-22. A G to A mutation in exon 10 of the adenosine triphosphate (ATP)-binding cassette subfamily G, member 5 gene, alters a tryptophan codon (UGG) to a premature stop codon (UAG). Crosses with mice doubly transgenic for the human ABCG5 and ABCG8 genes rescued platelet counts and volumes. ABCG5 and ABCG8 form a functional complex that limits dietary phytosterol accumulation. Phytosterolemia in homozygous mice confirmed a functional defect in the ABCG5/ABCG8 transport system. The Abcg5\textsuperscript{frac} mutation provides a new clinically significant animal model for human phytosterolemia and provides a new means for studying the role of phytosterols in hematologic diseases and testing therapeutic interventions.
Genotyping Protocols
Pyrosequencing: Abcg5
Genotyping resources and troubleshooting

Citation
When using the thrombocytopenia and cardiomyopathy mouse strain in a publication, please cite the originating article(s) and include JAX stock #001830 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
Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

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<tr>
<th>SERVICE/PRODUCT</th>
<th>DESCRIPTION</th>
<th>PRICE</th>
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<tr>
<td>Cryo Recovery</td>
<td>Heterozygous or wildtype for trac</td>
<td>$2,854.50</td>
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Terms are granted by individual review and stated on the customer invoice(s) and account statement. These transactions are payable in U.S. currency within the granted terms. Payment for services, products, shipping containers, and shipping costs that are rendered are expected within the payment terms indicated on the invoice or stated by contract. Invoices and account balances in
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