Overview

Also Known As: TS2-neo, G406R

The TS2-neo mutation has the G406R mutation associated with severe Timothy Syndrome (TS2) and an inverted neomycin resistance cassette, all inserted at the end of exon 8 of the CaV1.2 L-type calcium channel locus (Cacna1c). Heterozygous TS2-neo mice exhibit several characteristics of autism spectrum disorder, including repetitive/perseverative stereotypical behavior, decreased social interaction, decreased communication/vocalization, enhanced fear memory and self-injurious scratching. These mice are useful in studying the autism spectrum disorder (ASD) characteristics of Timothy Syndrome.
The TS2-neo allele has a G406R mutation associated with severe Timothy Syndrome (TS2) and an inverted neomycin resistance cassette, all inserted at the end of exon 8 of the CaV1.2 L-type calcium channel locus (Cacna1c). In similar G406R mutant mice without a neo cassette, expression of the G406R mutation-bearing channel causes death in both heterozygotes and homozygotes. Mice homozygous for the TS2-neo allele are also lethal. However, a single copy of the TS2-neo allele results in 75% lower G406R mutation-bearing channel expression in brain and no expression of the G406R mutation-bearing channel in heart (suggesting
transcriptional interference from the neomycin cassette). The attenuated expression diminishes the deleterious effects of mutant L-type channel overactivity and allows heterozygous TS2-neo animals to be viable and fertile. Heterozygous TS2-neo mice exhibit distinct traits strikingly reminiscent of the entire core triad of autism spectrum disorder (ASD): repetitive/perseverative behavior, impaired social behavior, and impaired communication. TS2-neo mice also display enhanced fear memory (reminiscent of persistent memories or abnormal fear associated with ASD), as well as self-injurious scratching.

Genetics

*Cacna1c<sup>tm2ltl</sup>*

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

Genotyping Protocols

Standard PCR: *Cacna1c* alternate1
Standard PCR: *Generic LacZ* Melt Curve Analysis
Probe: *Generic LacZ* Probe
Probe: *Cacna1c* Probe
Genotyping resources and troubleshooting

Breeding Considerations

Homozygous animals are lethal. When maintaining a live colony, heterozygous mice are bred with wildtype mice from the colony or with C57BL/6J inbred mice (Stock No. 000664).

Additional Breeding and Husbandry Support

Citation

When using the TS2-neo mouse strain in a publication, please cite the originating article(s) and include JAX stock #019547 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.

<table>
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<tr>
<th>Cryo Recovery</th>
<th>Heterozygous or wildtype for Cacna1c&lt;tm2Itl&gt;</th>
<th>$2,854.50</th>
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</thead>
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| Frozen Mouse Embryo | B6.Cg-Cacna1c<tm2Itl>/J Frozen Embryos | $2595.00 |

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