CAST;B6–Gars\textsuperscript{Nmf249}/JRwb

Stock No: 017540 | Gars\textsuperscript{P278KY}

Spontaneous Mutation

AVAILABILITY VARIES

Also Known As: B6;CAST-Gars\textsuperscript{Nmf249}/JRwb, Gars\textsuperscript{P278KY}

CAST;B6-Gars\textsuperscript{Nmf249}/JRwb mice have a severe axonal neuropathy of both sensory and motor axons. Mutant mice have abnormal neuromuscular junction morphology and impaired transmission, reduced nerve conduction velocities, and a loss of large-diameter peripheral axons, without defects in myelination. These mice may be useful in studies of inherited Charcot-Marie-Tooth peripheral neuropathies, type 2D (CMT2D).

GENETIC OVERVIEW

<table>
<thead>
<tr>
<th>Genetic Background</th>
<th>Generation</th>
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| Gars\textsuperscript{Nmf249} |

- **Allele Type:** Spontaneous
- **Gene Symbol:** Gars
- **Gene Name:** glycyl-tRNA synthetase

RESEARCH APPLICATIONS

Developmental Biology Research
Neurobiology Research

BASE PRICE

Starting at:

$1.00 Domestic price for female
Details

Detailed Description

NMF249 mice have a distal neuropathy in motor neurons with retracting axons (Figure 1) and poorly innervated or denervated neuromuscular junctions (Figure 2). Visualized through immunohistochemistry of neuromuscular junctions; green labeling indicates the presence of neurofilament protein in axons (SMI31) and synaptic vesicle proteins at motor terminals (SV2); red labeling indicates ACh receptors visualized with rhodamine-labeled alpha-bungarotoxin; yellow color reveals the overlay of the presynaptic nerve with the motor terminal arborization. Large diameter myelinated fibers are absent and a significant number of unidentified cells is evident in the sciatic nerve of affected mice (Figure 3). No defects have been observed in the CNS.

On the mixed CAST;B6 background, motor nerves have a 26% reduction in axons, neuromuscular junctions are partially denervated and nerve conduction velocity is substantially reduced.

On the C57BL/6 background, heterozygous mice are smaller than their littermates and develop an unsteady gait about 3 weeks of age (average 3.5 weeks of age +/− 0.5, n=22). The mice fail to thrive and die between 4-8 weeks of age. Homozygotes die before birth. Males and females are affected and do not live long enough to mate normally. Due to the difficulties in maintaining the strain, it is outcrossed to CAST/Ei every few generations.

Although the NMF249 mutation arose in an ENU mutagenized family, it is a spontaneous dominant mutation. This strain may be useful for studying Charcot-Marie-Tooth disease type 2D.

Development

Genetics

Gars<sup>tm269</sup>

Disease/Phenotype

Disease Terms

Research Areas By Genotype

Mammalian Phenotype Terms by Genotype

References

Technical Support
Genotyping Protocols
Genotyping resources and troubleshooting

Breeding Considerations
Homozygous mice die shortly after birth. The strain is maintained by mating heterozygotes to C57BL/6J (Stock No. 000664) mice for several backcross generations until breeding declines, and then backcrossing to CAST/EiJ (Stock No. 000928).

Additional Breeding and Husbandry Support

Mating System
Heterozygote to C57BL/6 for several backcross generations until breeding declines, then outcross to CAST/E

Citation
When using this Genetically Engineered mouse strain in a publication, please cite the originating article(s) and include JAX stock #017540 in your Materials and Methods section.

Facility Barrier Level Descriptions
A6 (Intermediate)

Pricing & Availability

Availability Varies

Domestic International
Pricing effective for USA, Canada and Mexico shipping destinations

<table>
<thead>
<tr>
<th>AGE</th>
<th>SEX</th>
<th>GENOTYPE</th>
<th>PRICE</th>
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</thead>
<tbody>
<tr>
<td>Approx 4-8 weeks</td>
<td>Female</td>
<td>Heterozygous for GarsNmf249</td>
<td>$1.00</td>
</tr>
<tr>
<td></td>
<td>Male</td>
<td>Heterozygous for GarsNmf249</td>
<td>$1.00</td>
</tr>
</tbody>
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