B6.B10Sn-Mctp1<sup>dwn</sup>/Kjn

**Stock No:** 009690 | deaf wanderer

- Congenic, Spontaneous Mutation

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**PLACE ORDER**

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

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**Overview**

**Also Known As:** deaf wanderer

This spontaneous intragenic deletion in Mctp1 also causes reduced expression of Nr2f1 in the developing cochlea, but not retina, indicative of a disruption in the long-range regulation of Nr2f1 transcription. This strain is valuable for understanding both the function of MCTP1 and the regulation and inner ear-specific function of NR2F1.
GENETIC OVERVIEW

<table>
<thead>
<tr>
<th>Genetic Background</th>
<th>Generation</th>
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<tbody>
<tr>
<td>00664 C57BL/6J</td>
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**Mctp1<sup>dwnd</sup>**

<table>
<thead>
<tr>
<th>Allele Type</th>
<th>Gene Symbol</th>
<th>Gene Name</th>
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<tbody>
<tr>
<td>Spontaneous (Modified</td>
<td>Mctp1</td>
<td>multiple C2 domains,</td>
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<tr>
<td>regulatory region)</td>
<td></td>
<td>transmembrane 1</td>
</tr>
</tbody>
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RESEARCH APPLICATIONS

BASE PRICE

Starting at:

VIEW PRICING OPTIONS

**Details**

**Detailed Description**

This spontaneous deletion in the sequence of Mctp1 encompasses exons 11-15, their intervening introns, 7,270 bp of intron 10-11 and 40,908 bp of intron 15-16. This deletion is expected to cause a frameshift and premature stop codon resulting in the loss of the third C2 domain and the two transmembrane helices of MCTP1. However, it is the long-range dysregulation of NR2F1 expression during inner ear development, and not Mctp1 inactivation, that is believed to be the primary cause of the outward phenotype of Mctp1<sup>dwnd</sup> mutant mice. Mice homozygous for the Mctp1 deaf wanderer mutation display moderate circling behavior due to inner ear defects, whereas Mctp1 knockout mice have normal inner ear anatomy and function. The cochlea of Mctp1<sup>dwnd</sup> mutant mice is smaller and shorter than normal, there are extra, disorganized cochlear inner hair cells near the base, an extra row of outer hair cells near the apex, the saccule is smaller than normal and fails to separate fully from the utricle, and the cochleosaccular duct is larger than normal. Auditory brainstem response analysis showed hearing impairment as early as 3 to 4 weeks of age, with thresholds 25-45 dB above those of controls.

RT-qPCR for exons 2-3 of Mctp1 in extractions from the E16.5 cochlear membranous labyrinth showed approximately 25% of normal levels of expression in deaf wanderer homozygotes. Additionally, RT-qPCR for Nr2f1 revealed a 50% reduction in expression in the cochlea of deaf wanderer homozygotes, but no change in expression in the retina, consistent with the loss of an auditory-specific enhancer region for Nr2f1 within the Mctp1 sequence deleted in deaf wanderer. Nr2f1 is approximately 1.4 Mb from Mctp1 on Chromosome 13. Mice homozygous for the Ming-Jer Tsai targeted disruption of Nr2f1, which deletes the amino terminus, DNA binding domain, and some of the ligand binding domain of NR2F1 (Tang et al. 2006), also have abnormal cochlear development with regions of supernumerary hair cells consistent with the phenotype of deaf wanderer homozygotes. However, all die neonatally with
no swallowing reflex, defects in the ninth cranial ganglion, and further axonal guidance and arborization defects elsewhere. Consistent with the deaf wanderer deletion modifying Nr2f1 expression, Mcpt1<sup>dwn</sup>+/+ Nr2f1<sup>tm1(KOMP)Mbo</sup> transheterozygotes have cochlear hair cell disorganization, excess inner hair cells, an extra row of outer hair cells in the apex region, malformed utricular and saccular macula, and hearing impairment consistent with that found in deaf wanderer homozygotes.

Genotyping Protocols

Breeding Considerations
A simple, PCR-based genotyping protocol is presented in Tarchini et al. 2018

Additional Breeding and Husbandry Support

Mating System
Homozygote x Homozygote

Citation
When using the deaf wanderer mouse strain in a publication, please cite the originating article(s) and include JAX stock #009690 in your Materials and Methods section.

Animal Health Reports

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.

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

| Related Products and Services | Frozen Mouse Embryo | B6.B10Sn-Mctp1<dwnd>/Kjn | $2595.00 |

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- By Gene
- By Collection

All Related Strains