**Overview**

Also Known As: hyperspin

This 123.6 kb intragenic deletion appears to include a regulatory sequence that controls Dlx5 expression at least in the otocyst so this mutant provides a model of split-hand/foot malformation 1 (SHFM1) without craniofacial defects and a model with which to dissect the regulation of the genomic region that includes Dlx5 and Slc25a13.
GENETIC OVERVIEW

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<table>
<thead>
<tr>
<th>Genetic Background</th>
<th>Generation</th>
</tr>
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<tbody>
<tr>
<td>00664 C57BL/6J</td>
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**Slc25a13<sup>hspn</sup>**

<table>
<thead>
<tr>
<th>Allele Type</th>
<th>Gene Symbol</th>
<th>Gene Name</th>
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<tbody>
<tr>
<td>Spontaneous (Null/Knockout,</td>
<td>Slc25a13</td>
<td>solute carrier family 25 (mitochondrial carrier, adenine</td>
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<tr>
<td>Dominant negative)</td>
<td></td>
<td>nucleotide translocator), member 13</td>
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**Marker(s)**

<table>
<thead>
<tr>
<th>Marker Symbol</th>
<th>Marker Name</th>
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<tr>
<td>Dlx5</td>
<td>distal-less homeobox 5</td>
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</table>

RESEARCH APPLICATIONS

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Developmental Biology Research
Sensorineural Research

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

The hyperspin mutation is a spontaneous 123.6 kb deletion that begins in intron 3 of *Slc25a13* and ends in exon 17, an interval that includes an enhancer of *Dlx5*. This deletion includes the putative enhancer elements hs2313 and hs1642, and overlaps the regions corresponding to the predicted intervals of split-hand/foot malformation (SHFM) and split-hand/foot malformation with hearing loss (SHFM & HL) sub-regions of the SHFM1 locus, but not the split-hand/foot malformation with hearing loss and craniofacial abnormalities (SHFM & HL & CF) sub-region. Hyperspin homozygotes exhibit the phenotypes indicative of a vestibular defect, rapid circling, head shaking, and failure to orient in water, but null alleles in *Slc25a13* have not been found to cause vestibular defects. At 4 to 5 weeks of age homozygotes were found to be deaf by ABR analysis. At 5 weeks of age the inner ears of homozygotes are small...
and malformed, the cochlea has too few turns and appears swollen, and the three semicircular canals are absent, reduced, or malformed. At embryonic day 15.5 the endolymphatic duct and sac and the superior and posterior semicircular canals are missing, the cochlear duct is wider and has fewer coils than normal, and most homozygotes also lack the lateral semicircular canal and utricle. Scanning electron microscopy of the basal and middle turns of the cochlea at 4 weeks of age revealed only occasional missing hair bundles, but the apical turn had an additional forth and sometimes fifth row of outer hair cells and abnormal hair bundle orientations. Only approximately 30% of female and 50% of male hyperspin homozygotes survive to wean age. Hyperspin homozygotes have reduced otocyst expression but not branchial arch expression of Dlx5, a gene more than 600 kb away on Chromosome 6, and null alleles of Dlx5 have been found to cause perinatal death, dysmorphic inner ears and other craniofacial abnormalities. While hyperspin heterozygotes have a normal phenotype fewer than 10% of Slc25a13$^{hspn}$ Dlx5$^+/Slc25a13^+$ Dlx5$^{tm1(creERT2)zh}$ transchromosomal heterozygotes survive to wean age, a more severe phenotype than that of hyperspin homozygotes and further proof of the presence of a Dlx5 modifier within the hyperspin deletion. These double heterozygotes display circling and head bobbing, are deaf by ABR, and have malformed inner ears similar to that found in hyperspin homozygotes.

To identify the sequence within the hyperspin deletion that regulates Dlx5 otic expression, endonuclease-mediated deletions were generated of hs2313 in intron 14, hs1642 in intron 3, and intron 3 through 4. None of these caused the phenotype found in hyperspin homozygotes, even when Dlx5$^{tm1(creERT2)zh}$ transchromosomal heterozygotes were generated.
Genotyping Protocols
Genotyping resources and troubleshooting

Citation
When using the hyperspin mouse strain in a publication, please cite the originating article(s) and include JAX stock #005679 in your Materials and Methods section.

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>
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<tr>
<td>Cryo Recovery</td>
<td>Heterozygous for Slc25a13&lt;hspn&gt;</td>
<td>$2,854.50</td>
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