The Gtf2i duplication allele (Gtf2i) has one additional copy of a functional mouse general transcription factor 2I gene (TFII-I). These mice model characteristics of 7q11.23 duplication syndrome (Dup7q11.23), and may be useful for studying the cellular mechanisms underlying GTF2I-related neurodevelopmental disorders and molecular separation-anxiety pathways.

Donating Investigator
Lucy R Osborne, University of Toronto

### GENETIC OVERVIEW

**Genetic Background**

<table>
<thead>
<tr>
<th>Dp(5Gtf2i-Gtf2ird1)1Lro</th>
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<tbody>
<tr>
<td>Allele Type</td>
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**Generation**

<table>
<thead>
<tr>
<th>Dp(5Gtf2i-Gtf2ird1)1Lro</th>
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</thead>
<tbody>
<tr>
<td>Gene Symbol</td>
</tr>
<tr>
<td>Gene Name</td>
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### RESEARCH APPLICATIONS

- Developmental Biology Research
- Neurobiology Research
- Cell Biology Research

**VIEW GENETICS**

**VIEW ALL RESEARCH APPLICATIONS**
The Gtf2i duplication allele (Gtf2i\textsuperscript{Dup}; Dp(5Gtf2i-Gtf2ird1)1Lro) has one additional copy of a functional mouse Gtf2i gene (and a non-functional Gtf2ird1 allele) inserted centromeric to the endogenous general transcription factor 2I locus (GTF2I; encoding protein TFII-I).

TFII-I is a transcriptional activator that regulates neuronal maturation and intracellular Ca2+ levels (through the TRPC3 channel), and is highly expressed in the prenatal and postnatal developing brain. The human chromosome 7 locus, 7q11.23, is a 1.5-1.8 Mbp region susceptible to genomic rearrangement through non-allelic homologous recombination. This region contains ~25 protein coding genes; including GTF2I. 7q11.23 duplication syndrome (Dup7q11.23) and 7q11.23 deletion syndrome (Williams-Beuren Syndrome [WBS]) are neurodevelopmental disorders with unique/contrasting cognitive and behavioral profiles. In humans, Dup7q11.23 is associated with speech and language delay, anxiety (both social and nonsocial) and autism. Some individuals exhibit macrocephaly and increased brain volume, as well as cortical dysplasia. Approximately 30% of 4- to 12-year-olds with Dup7q11.23 may meet diagnostic criteria for a separation-anxiety disorder.

Mice that are wildtype (Gtf2i\textsuperscript{+/+}), hemizygous (Gtf2i\textsuperscript{Dup/+}) and homozygous (Gtf2i\textsuperscript{Dup/Dup}) for the Gtf2i duplication allele harbor a total of two, three and four Gtf2i copies, respectively. When compared to mouse pups with one Gtf2i copy (heterozygous for a knockout allele) or two Gtf2i copies (wildtype), pups with additional Gtf2i copies (hemizygous and homozygous mice) show significantly increased maternal separation-induced anxiety as measured by ultrasonic vocalizations. Copy number alterations of GTF2I affect cortical neuronal morphology/maturation, novel object recognition and agonist-induced calcium entry (calcium signaling via the TRPC3 channel). Specifically, Gtf2i\textsuperscript{Dup/+} mice exhibit deficits in a cortically-dependent learning and memory task (novel object recognition). Ca2+ influx via neuronal TRPC3 channels is decreased in Gtf2i\textsuperscript{Dup/+} (whereas it is increased in mice heterozygous for a knockout allele).

The donating investigator reports hemizygous and homozygous mice are viable and fertile with healthy nutritional status. When maintaining their colony by breeding wildtype (noncarrier) females with hemizygous males, they report the mice are good breeders with no reduced survival. They also report no GTF2IRD1 expression from the additional copy of the Gtf2ird1 allele (i.e., the additional copy of Gtf2ird1 is a functional null).
Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

Genotyping Protocols
Standard PCR: Dp(5Gtf2i-Gtf2ird1)1Lro-Alternate 1
Genotyping resources and troubleshooting

Breeding Considerations
When maintaining our live colony, wildtype (noncarrier) females from the colony or C57BL/6J inbred females (Stock No. 000664) may be bred with hemizygous (Gtf2i<sup>Dup</sup>) males.

Of note, the donating investigator reports hemizygous and homozygous mice are viable and fertile with healthy nutritional status. When maintaining their colony by breeding wildtype (noncarrier) females with hemizygous males, they report the mice are good breeders with no reduced survival.

Additional Breeding and Husbandry Support
Mating System
Noncarrier x Hemizygote

Citation
When using the Gtf2i duplication (Gtf2i<sup>Dup</sup>) mouse strain in a publication, please cite the originating article(s) and include JAX stock #027792 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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<th>Domestic</th>
<th>International</th>
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<td>Pricing effective for USA, Canada and Mexico shipping destinations</td>
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Related Products and Services

| Frozen Mouse Embryo | B6.129(Cg)-Dp(5Gtf2i-Gtf2ird1)1Lro/JcrwJ Frozen Embryo | $2,595.00 |

Payment Terms and Conditions

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 arrears of stated terms may result in The Jackson Laboratory pursuing collection activities including but not limited to outside agencies and court filings.

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The Jackson Laboratory has rigorous genetic quality control and mutant gene genotyping programs to ensure the genetic background of JAX® Mice strains as well as the genotypes of strains with identified molecular mutations. JAX® Mice strains are only made available to researchers after meeting our standards. However, the phenotype of each strain may not be fully characterized and/or captured in the strain data sheets. Therefore, we cannot guarantee a strain’s phenotype will meet all expectations. To ensure that JAX® Mice will meet the needs of individual research projects or when requesting a strain that is new to your research, we suggest ordering and performing tests on a small number of mice to determine suitability for your particular project. We do not guarantee breeding performance and therefore suggest that investigators order more than one breeding pair to avoid delays in their research.

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Terms Of Use

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Phone: 207-288-6470
Email: TechTran@jax.org

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