B6.129(Cg)-Dp(5Gtf2i-Gtf2ird1)1Lro/JcrwJ

Stock No: **027792** | Gtf2i duplication (Gtf2i ^{Dup})

Congenic, Duplication

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

PLACE ORDER

l). 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

READ MORE +

GENETIC OVERVIEW

Genetic Background	Generation

Dp(5Gtf2i-Gtf2ird1)1Lro

Allele Type	Gene Symbol	Gene Name
Targeted (Not Applicable)	Dp(5Gtf2i-Gtf2ird1)1Lro	duplication, Chr 5, L Osborne 1

VIEW GENETICS

RESEARCH APPLICATIONS

Developmental Biology Research
Neurobiology Research
Cell Biology Research

VIEW ALL RESEARCH APPLICATIO

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

VIEW PRICE LIST

Details

Detailed Description

The Gtf2i duplication allele (Gtf2i^{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$^{+/+}$), hemizygous (Gtf2i$^{Dup/+}$) and homozygous (Gtf2i$^{Dup/Dup}$) 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$^{Dup/+}$ mice exhibit deficits in a cortically-dependent learning and memory task (novel object recognition). Ca2+ influx via neuronal TRPC3 channels is decreased in Gtf2i$^{Dup/+}$ (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).

Development

Control Suggestions

Selected References

Genetics

Dp(5Gtf2i-Gtf2ird1)1Lro

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

CONTACT 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 ($Gtf2i^{Dup/+}$) 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 ($Gtf2i^{Dup}$) 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

Cryo Recovery

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

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	Heterozygous or wildtype for Dp(5Gtf2i-Gtf2ird1)1Lro	$2,854.50

RELATED PRODUCTS AND SERVICES

Frozen Mouse Embryo	B6.129(Cg)-Dp(5Gtf2i-Gtf2ird1)1Lro/JcrwJ Frozen Embryo	$2595.00

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.

THE JACKSON LABORATORY'S GENOTYPE PROMISE

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.

Terms Of Use

TERMS OF USE

General Terms and Conditions

QUESTIONS ABOUT TERMS OF USE

ADDITIONAL USE RESTRICTIONS APPLY

Use of MICE by companies or for-profit entities requires a license prior to shipping.

LICENSING INFORMATION

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

Related Strains

All

By Allele

By Gene

By Collection

Leading the search for

TOMORROW'S CURES