

**B6;129S-Fdxr<sup>m1J</sup> Otop2<sup>m1J</sup>/GrsrJ**Stock No: **026096** | stiffened hindquarters Spontaneous Mutation

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This spontaneous mutant develops a mature onset stiffening of the hindquarters, mitochondrial defects and retinal ganglion cell degeneration, providing a model for mitochondriopathy with optic atrophy.

[R E A D M O R E +](#)

## GENETIC OVERVIEW

**Genetic Background****Generation***Fdxr<sup>m1J</sup>***Alele Type**

Spontaneous (Hypomorph)

**Gene Symbol***Fdxr***Gene Name**

ferredoxin reductase

*Otop2<sup>m1J</sup>***Alele Type**

Spontaneous

**Gene Symbol***Otop2***Gene Name**

otopetrin 2

[V I E W G E N E T I C S](#)

## RESEARCH APPLICATIONS

Neurobiology Research

Internal/Organ Research

Developmental Biology Research

[V I E W A L L R E S E A R C H A P P L I C A T I O N S](#)

## BASE PRICE

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\$2,854.50 Domestic price Cryo Recovery

V I E W   P R I C E   L I S T

### Details

#### Detailed Description

The stiffened hindquarters phenotype presents at approximately 9 weeks of age, but has presented as early as 7 weeks of age or as late as almost 12 weeks of age. The hind legs become stiffened and are held much wider than normal and the hips and proximal tail also appear stiffened. The gait becomes rigid and sometimes hop-like in the hindquarters although the front legs appear to move in the normal alternating manner without noticeable stiffness. This stiffening of the hindquarters is progressive, but does not become crippling and homozygotes are able to move reasonably, access the food hopper and water bottle, and reproduce. Both males and females will reproduce after phenotypic onset and homozygous females have taken care of their litters, but homozygous females are not used to maintain this strain. One female homozygote at 6 weeks of age and one female homozygote at five and a half weeks of age, along with aged matched controls, were assessed for their ability to swim and both were able to swim. Two distinct point mutations were identified in the region of Chromosome 11 to which this mutation mapped: a C to T SNV at Chromosome 11 position 115,269,462 bp (GRCm38) resulting in the codon change cGg/cAg in ferredoxin reductase (*Fdxr*) and the resulting amino acid change R389Q; and a G to A change at Chromosome 11 position 115,332,175 bp (GRCm38) in the 5-prime UTR of otopetrin 2 (*Otop2*) with unknown consequence. Mitochondriopathy with optic atrophy has been attributed to the ferredoxin reductase mutation. Ferredoxin NADP reductase activity is reduced in homozygotes, most severely in muscle and brain but also significantly reduced in liver and heart. Retinal ganglion cell degeneration is evident at 6 months of age when approximately half of the retinal ganglion cell layer nuclei are lost and transmission electron micrographs of these cells show empty cytoplasm, shrunken nuclei, aggregated chromatin, thinned myelin sheaths, and damaged mitochondria in their axons.

#### Development

#### Control Suggestions

#### Selected References

### Genetics

#### *Fdxr*<sup>m1J</sup>

#### *Otop2*<sup>m1J</sup>

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## – Disease/Phenotype

[+ Disease Terms](#)

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[+ Research Areas By Phenotype](#)

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[+ Mammalian Phenotype Terms by Genotype](#)

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[+ References](#)

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## – Technical Support

C O N T A C T   T E C H N I C A L   S U P P O R T

### Genotyping Protocols

Probe:[Fdxr EP Alternate1](#)

End Point Analysis:[Otop3-EP](#)

Sanger sequencing:[Fdxr](#)

Sanger sequencing:[Otop3](#)

[Genotyping resources and troubleshooting](#)

### Breeding Considerations

Although they can reproduce, female homozygotes are not used for breeding. Instead this strain is maintained by intercrossing heterozygous females with either homozygous or heterozygous males.

[Additional Breeding and Husbandry Support](#)

### Citation

When using the stiffened hindquarters mouse strain in a publication, please [cite the originating article\(s\)](#) and include JAX stock #026096 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](#)*

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## ☰ Pricing & Availability



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Pricing effective for USA, Canada and Mexico shipping destinations

#### CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
<a href="#">Cryo Recovery</a>	Heterozygous for Fdxr, Heterozygous for, Otop3	\$2,854.50

#### RELATED PRODUCTS AND SERVICES

<a href="#">Frozen Mouse Embryo</a>	B6;129S-Fdxr<m1J> Otop2<m1J>/GrsrJ Frozen Embryos	\$2595.00
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Email: [TechTran@jax.org](mailto:TechTran@jax.org)

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