The Jackson Laboratory

B6N.Cg-Tg(DUX4*)1Maar/J

Stock No: **027991** | D4Z4-2.5

Congenic, Transgenic

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

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2.5 mice, along with the control strain (D4Z4-12.5 transgenic mice carrying a normal sized, non-pathogenic allele; Stock No. 028012), may be useful in studying facioscapulohumeral dystrophy.

Donating Investigator

Silvere M van der Maarel, Leiden University Medical Center (LUMC)

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GENETIC OVERVIEW

Genetic Background **Generation**

Tg(DUX4*)1Maar

Allele Type

Transgenic (Inserted expressed sequence, Humanized sequence)

VIEW GENETICS

RESEARCH APPLICATIONS

Neurobiology Research

VIEW ALL RESEARCH APPLICATIO

BASE PRICE

Starting at:

$2,854.50 Domestic price Cryo Recovery

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Details

Detailed Description

Facioscapulohumeral dystrophy (FSHD) is an autosomal dominant muscular dystrophy predominantly affecting facial and upper extremity muscles. Hearing loss and retinopathy are frequently observed extramuscular features. FSHD1 is caused by contraction of the D4Z4 macrosatellite repeat to 10 or less D4Z4 units in the subtelomeric region of 4q on the 4A161 haplotype. DUX4 is more efficiently silenced with increasing D4Z4 copy number: human FSHD1 is associated with 1-10 D4Z4 repeat units, whereas the unaffected population has 11-100 D4Z4 repeat units. When expressed in skeletal muscle, the DUX4 transcription factor activates genes normally expressed in the germline (essentially inducing a stem cell program in the postmitotic muscle cell) and also suppresses several genes involved in the innate immune response.

The D4Z4-2.5 transgene contains a FSHD1-sized D4Z4 repeat derived from a human FSHD1 individual (haplotype 4A161; containing 2.5 D4Z4 repeat units followed by the pLAM *DUX4* exon 3 with polyA signal). Hemizygous D4Z4-2.5 transgenic mice recapitulate important epigenetic and DUX4 expression attributes seen in human FSHD1; including high DUX4 expression levels in the germline, incomplete epigenetic repression of DUX4 in somatic tissue, and FSHD-specific variegated DUX4 expression in sporadic muscle nuclei associated with D4Z4 chromatin relaxation.

The phenotype listed below compares the D4Z4-2.5 transgenic mice carrying a D4Z4 genomic region from a contracted pathogenic FSHD1 allele (Stock No. 027991) with the D4Z4-12.5 transgenic mice carrying a normal sized, non-pathogenic allele (Stock No. 028012). Unless noted otherwise, hemizygous mice are described.

The fewer (2.5) D4Z4 repeat units in D4Z4-2.5 mice causes inefficient DUX4 repression in somatic tissue (bodywide hypomethylation of D4Z4) and manifests in features of human FSHD1. In contrast, D4Z4-12.5 mice have a D4Z4 repeat array (12.5) that maintains somatic tissue epigenetic silencing of DUX4 at a low enough level sufficient to preclude FSHD1 phenotype in humans.

D4Z4-2.5 mice have abundant levels of DUX4 mRNA in germ line tissues (most notably in testis), in ES cells and in early developmental stages. In D4Z4-12.5 mice, the pattern is similar but at lower levels.

In adult somatic tissue from D4Z4-2.5 mice, DUX4 mRNA expression is low and variegated in all analyzed skeletal muscles, and a variegated expression pattern of DUX4 protein in skeletal muscle nuclei is observed. In adult somatic tissue from D4Z4-12.5 mice, DUX4 transcripts are reproducibly detected only in the tibialis anterior and pectoralis muscles, whereas all other somatic tissues do not show reproducible DUX4 expression.

D4Z4-2.5 mice exhibit chromatin relaxation of the D4Z4 repeats compared to D4Z4-12.5 mice.

In both D4Z4-2.5 and D4Z4-12.5 mice, the overall morphology/histology of the limb and some head muscles appear normal. Hemizygous mice are viable and fertile. The donating investigator also reports that homozygous mice are viable and fertile, and that the number of D4Z4 repeats within each line stays the same through several generations of breeding (*i.e.*, the D4Z4 repeat unit is stable).

Approximately half of the D4Z4-2.5 mice develop eye abnormalities (keratitis) around 8-12 weeks of age.

Development

Expression Data

Control Suggestions

Selected References

Genetics

Tg(DUX4*)1Maar

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

CONTACT TECHNICAL SUPPORT

Genotyping Protocols

Standard PCR:Tg(DUX4*)1Maar
Genotyping resources and troubleshooting

Breeding Considerations

When maintaining a live colony, hemizygous mice may be bred to wildtype (noncarrier) siblings or to C57BL/6NJ inbred mice (Stock No. 005304). The donating investigator reports that homozygous mice are viable and fertile, and that the number of D4Z4 repeats stays the same through several generations of breeding (*i.e.*, the D4Z4 repeat unit is stable).

Additional Breeding and Husbandry Support

Mating System
Hemizygote x Hemizygote

Citation
When using the D4Z4-2.5 mouse strain in a publication, please cite the originating article(s) and include JAX stock #027991 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 | **Internatic**

Pricing effective for USA, Canada and Mexico shipping destinations

CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
Cryo Recovery	Hemizygous or Non Carrier for Tg(DUX4*)1Maar	$2,854.50

RELATED PRODUCTS AND SERVICES

Frozen Mouse Embryo	B6N.Cg-Tg(DUX4*)1Maar/J	$2595.00

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