These spontaneous Dmd<sup>mdx</sup> mutant mice do not express dystrophin and may be useful for studying Duchenne muscular dystrophy.

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The X-linked dystrophin gene (Dmd) is highly expressed in muscle cells and encodes a cytoskeletal protein which localizes to the inner face of the sarcolemma. Dystrophin molecules bind to cytoskeletal F-actin and transmembrane beta-dystroglycan as part of a complex, multimolecular unit that mediates signaling between the intracellular cytoskeleton and the extracellular matrix. The structure and localization also suggest that dystrophin is important for stabilizing the plasma membrane, particularly during contraction. The mdx mutation of Dmd is recessive and heterozygous females are visually indistinguishable from wild-type mice. Females homozygous and males hemizygous for the Dmd^{mdx} allele retain a normal lifespan and can survive up to two years. Like human patients who suffer from one of the most common neuromuscular diseases, Duchenne muscular dystrophy (DMD), the Dmd^{mdx} mutants do not express dystrophin and therefore have been routinely used as an animal model of the disease even though the resultant myopathology is much less severe compared to the human disease course.

Muscle from Dmd^{mdx} mutants is histologically normal early in postnatal development, but starting around 3 weeks muscle necrosis develops with some visible muscle weakness. Biochemical analysis of related pathologies includes elevated serum creatine kinase and pyruvate kinase levels, along with an accumulation of macrophages, both early markers of muscle degeneration. While skeletal limb muscles are characterized by a persistent and progressive degeneration and necrosis, this is offset by a regenerative response activated by satellite cells and muscle hypertrophy. The regenerating fibers are morphologically typified by small-caliber centrally nucleated fibers; nevertheless, the mice assume normal behavior. The muscles of Dmd^{mdx} mutants have an overall reduction in elasticity, making them more susceptible to injury due to lengthening-activation. Interestingly, the mutant leg muscles were found to initially develop normally, but the differentiation of regenerated myotubes into both fast and slow fiber types was significantly inhibited. The comparatively mild phenotype of the Dmd^{mdx} mice can, in part, be attributed to the compensatory function of the dystrophin-related protein utrophin, which is highly upregulated in regenerating muscle fibers in adult Dmd^{mdx} mutants. This functional redundancy was demonstrated in mice deficient for both of these sarcolemmal proteins where the observed muscular dystrophy was much more severe and
led to a premature death in the dystrophin/utrophin double mutants. Also, the muscle-specific transcription factor MYOD may also be involved in facilitating muscle regeneration in the mutant mice as Dmd<sup>mdx</sup> mice also lacking MYOD exhibit a more severe dystrophy of the muscles. In contrast to limb muscles, the diaphragm muscles of Dmd<sup>mdx</sup> mice do not undergo a significant regeneration phase such that the continuous dystrophy weakens these muscles with age. The specific twitch force, specific titanic force and maximum power are all reduced in the diaphragm of Dmd<sup>mdx</sup> mutants.

Auditory function of Dmd<sup>mdx</sup> mutants, as assessed by brainstem auditory evoked potentials, is altered leaving them more vulnerable to noise damage. In mouse cardiac myocytes, dystrophin colocalizes with L-type calcium channels; in Dmd<sup>mdx</sup> mutants, the inactivation of these channels is reduced and voltage-dependant activation shifts to more positive potentials, providing evidence that the protein normally regulates calcium channel activity in cardiac tissue. In brain areas associated with learning, memory and cognitive tasks, dystrophin and its isoforms have been detected within postsynaptic specializations. In the Dmd<sup>mdx</sup> mouse sympathetic superior cervical ganglion, postsynaptic nicotinic acetylcholine receptor complexes containing the alpha3 subtype are destabilized as assayed by immunocytochemical and immunoprecipitation techniques. That proper dystrophin function is linked in nervous tissue to synaptic ligand-gated ion channel organization raises intriguing possibilities regarding the pathologic mechanisms underlying the cognitive defects often seen in DMD patients. [reviewed by Watchko et al. 2002, Durbeej and Campbell 2002; Ahn and Kunkel 1993; Cook and Davisson 1991; Doolittle 1997; Monaco and Kunkel 1987; Tamura et al. 1993; Stevens and Faulkner 2000; Del Signore et al. 2002; Houzelstein et al. 1992; Sicinski et al. 1989; Deconinck et al. 1997; Grady et al. 1997; Earshaw et al. 2002; Chen et al. 2002; D'Souza et al. 1995; Lynch et al. 2001; Carretta et al. 2001; Sadeghi et al. 2002; Lidov et al. 1995]

Nuclear opacity (cataracts) can be seen in the lens of one day old mice. A slight anterior subcapsular opacity is seen by four days progressing to complete anterior subcapsular opacity in 150 day old mice.
Genotyping Protocols

End Point Analysis: Dmd

Genotyping resources and troubleshooting

Breeding Considerations

*This strain is a challenging breeder.*

**Additional Breeding and Husbandry Support**

**Mating System**

Homozygote x Hemizygote

**Appearance**

black, affected

**Related Genotype:** $a/a \text{Dmd}^{mdx}$ or $a/a \text{Dmd}^{mdx}/Y$

**Citation**

When using the mdx mouse strain in a publication, please cite the originating article(s) and include JAX stock #001801 in your Materials and Methods section.

**Animal Health Reports**

*Facility Barrier Level Descriptions*

- AX5 (Standard)
- RB07 (Maximum)

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**International**

*Please contact Customer Service for pricing and availability.*
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