

VETERINARY TECHNICAL DATASHEET

Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier



Mutation Found In :Norfolk Terrier

Disorder Type

- Muscle

Disease Severity

- Severe

Background

Dystrophin-deficient muscular dystrophy (Duchenne muscular dystrophy) is a progressive, hereditary disorder leading to muscular dysfunction. The disease is caused by the deficiency of dystrophin that protects muscles from damage during muscle contraction. A form of the disease has been described in Norfolk terriers.

Key Signs

- Progressive muscular weakness
- Respiratory dysfunction
- Cardiomyopathy

Clinical Description

Clinical signs of the disease appear at a young age. Affected puppies show progressive muscular weakness, respiratory problems, and have cardiomyopathy. The prognosis of the disease is poor.

Mode of Inheritance

- X-linked

Gene Name

- Dystrophin

Next Steps

Glucocorticoid therapy and dantrolene therapy may provide temporary improvement. However, because of the severity of the clinical signs, affected puppies are usually euthanized at a young age on welfare grounds.

References

Jenkins C, Forman O. Identification of a novel frameshift mutation in the DMD gene as the cause of muscular dystrophy in a Norfolk terrier dog. *Canine Genetics and Epidemiology*;2:7, 2015.