

VETERINARY TECHNICAL DATASHEET

X-Linked Myotubular Myopathy



Mutation Found In :Labrador Retriever

Disorder Type

- Muscle

Disease Severity

- Severe

Background

X-linked myotubular myopathy (XLMTM) is an inherited disorder that affects a myotubular protein involved in cellular transport particularly in the muscle cell and was discovered in Labrador Retrievers. The disorder is characterized by early-onset pelvic limb weakness, progressing into an inability to move. Dogs with this disease generally present with muscle weakness, eating difficulties, respiratory distress, and delayed motor milestones. The disease follows an X-linked mode of inheritance.

Key Signs

- Progressive muscle weakness
- Muscle atrophy
- Absence of patellar reflexes
- Inability to rise and walk
- Difficulty chewing and swallowing

Clinical Description

The clinical signs of X-linked myotubular myopathy can be seen in puppies as young as 10 to 19 weeks of age with early signs of muscle atrophy being apparent earlier in some cases. Pelvic limb weakness is typically observed as one of the first signs. Affected dogs also lack patellar reflexes. X-linked myotubular myopathy is characterized by rapidly progressing muscle weakness and muscle atrophy. Affected dog won't be able to rise and move unassisted within a few weeks of the onset of clinical signs and may also have difficulties chewing and swallowing. There is no cure for the condition.

Mode of Inheritance

- X-linked

Gene Name

- MTM1

Next Steps

Affected puppies are usually euthanized on welfare grounds because of the severity of the condition.

References

Beggs AH, Böhm J, Snead E, Kozlowski M, Maurer M, Minor K, Childers MK, Taylor SM, Hitte C, Mickelson JR, Guo LT, Mizisin AP, Buj-Bello A, Tired L, Laporte J, Shelton GD. MTM1 mutation associated with X-linked myotubular myopathy in Labrador Retrievers. Proc Natl Acad Sci U S A 107:14697-702, 2010.

Snead ECR, Taylor SM, van der Kooij M, Cosford K, Beggs AH, Shelton GD. Clinical Phenotype of XLinked Myotubular Myopathy in Labrador Retriever Puppies. J Vet Intern Med. 2015 Jan-Feb; 29(1): 254–260.