VETERINARY TECHNICAL DATASHEET

Myotubular Myopathy; mutation originally found in Rottweiler

Mutation Found In :Rottweiler

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 cells. Two mutations causing myotubular myopathy have been discovered and this mutation is responsible for the disease in the Rottweiler. The disorder is characterized by early-onset pelvic limb weakness, progressing into an inability to move. The disease follows an X-linked mode of inheritance. Given males only have one X chromosome, the condition is seen most commonly in males as a single affected copy will cause the condition; females require two copies to exhibit the condition.

Key Signs

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

Clinical Description

The clinical signs of X-linked myotubular myopathy can be seen in puppies as young as 10-19 weeks of age. Pelvic limb weakness is typically observed as one of the first signs. Affected dogs also lack patellar reflexes. XLMTM 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. Excessive autophagy and prominent autophagic vacuoles are seen upon histopathology.

Mode of Inheritance

- X-linked

Gene Name

- MTM1

Next Steps

As the disorder is progressive, the welfare of affected dogs should be monitored closely. Owners of affected dogs should be advised that their dog will require assistance with movement and that clinical signs are likely to progress rapidly.

References

Shelton GD, Rider BE, Child G, Tzannes S, Guo LT, Moghadaszadeh B, Troiano EC, Haase B, Wade CM, Beggs AH. X-linked myotubular myopathy in Rottweiler dogs is caused by a missense mutation in Exon 11 of the MTM1 gene. Skelet Muscle. 2015 Jan 27;5(1):1.