VETERINARY TECHNICAL DATASHEET

Nemaline Myopathy; mutation originally found in American Bulldog

Mutation Found In :Bulldog (American)

Disorder Type

- Muscle

Disease Severity

- Mild/moderate

Background

Nemaline myopathy is a rare muscular disorder that is characterized by nemaline rod bodies inside muscle fibers. Nemaline myopathy can be either hereditary or acquired and there are different forms of the disorder. Here we describe the rare hereditary nemaline myopathy that has been found in the American Bulldog breed.

Key Signs

- Muscle weakness
- Exercise intolerance
- Tremors

Clinical Description

Clinical signs are first noticed around 2 months of age. Affected puppies are able to move but have generalized muscle atrophy and tremors leading to exercise intolerance. The clinical signs are relatively non-progressive. The disease should be a differential diagnosis based on breed history and clinical presentation when other possible causes have been ruled out. Serum creatine kinase levels are elevated in affected dogs and EMG and MNCV testing show typical changes. The diagnosis is confirmed by muscle biopsy.

Mode of Inheritance

- autosomal recessive

Gene Name

- NEB

Next Steps

There is no curative treatment for the disorder. Treatment is supportive care.

References

Evans J, Cox M, Huska J, Li F, Gaitero L, Guo L, Casal M, Granzier H, Shelton D, Clark LA. Exome sequencing reveals a nebulin nonsense mutation in a dog model of nemaline myopathy. Mamm Genome (2016) 27:495–502. DOI 10.1007/s00335-016-9644-9.