Stenotic nasopharyngeal dysgenesis and suspect muscular dystrophy in a Jack Russell Terrier dog (*Canis lupus familiaris*)

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Introduction

Stenotic nasopharyngeal dysgenesis is a rare condition described in dogs characterised by abnormal development of the nasopharynx, with thickening of the soft palate and irregular attachment to the pharyngeal wall. Muscular dystrophies are a group of inherited, degenerative myopathies that are typically noninflammatory and often X-linked. Here we describe the macroscopic and histopathological findings in a case of stenotic nasopharyngeal dysgenesis associated with suspect muscular dystrophy (MD) in a 4-year and 7-month-old, male neutered, Jack Russell Terrier dog.

Material and Methods

A complete post-mortem examination was carried out and samples of soft palate, oesophagus, and diaphragm were formalin-fixed and routinely processed for histopathology. Five-micron sections were stained with H&E and Masson's trichrome stain.

Results

The dog presented with a chronic history of inability to swallow, dysphagia with episodes of cyanosis while eating, and poor body condition. Biochemistry revealed markedly increased levels of creatine kinase. CT scan revealed nasopharyngeal dysgenesis (**Figure 1**) associated with multifocal, oesophageal and diaphragmatic thickening, which were confirmed during post-mortem examination (**Figure 2**). The remaining organs and body systems were macroscopically unremarkable.

On histopathology, the oesophagus, diaphragm, and soft palate showed changes consistent with a chronic degenerative myopathy, characterised by myofibre atrophy, degeneration and loss, with replacement by coalescing bands of mature collagen with variable numbers of admixed fibroblasts (fibrosis), and mature adipose tissue (fibrofatty replacement). Multifocal myofibre hypertrophy, regeneration, and mineralization were also present (**Figures 3-6**). These changes are compatible with a form of MD.

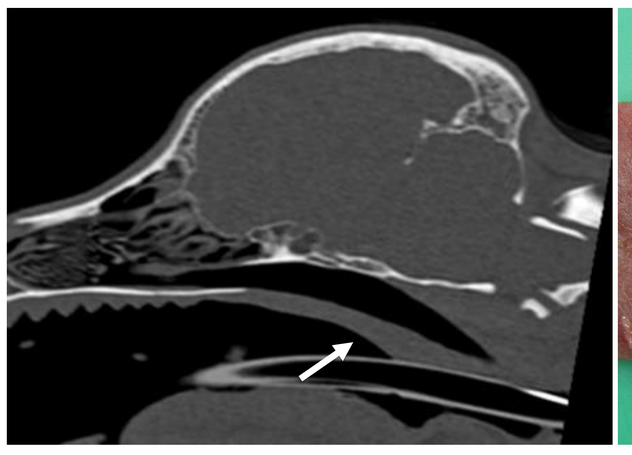

Figure 1. CT scan. Abnormal nasopharynx with absence of communication between nasopharynx and oropharynx due to abnormally long palatopharyngeal muscles in continuity with the oesophagus (arrow).

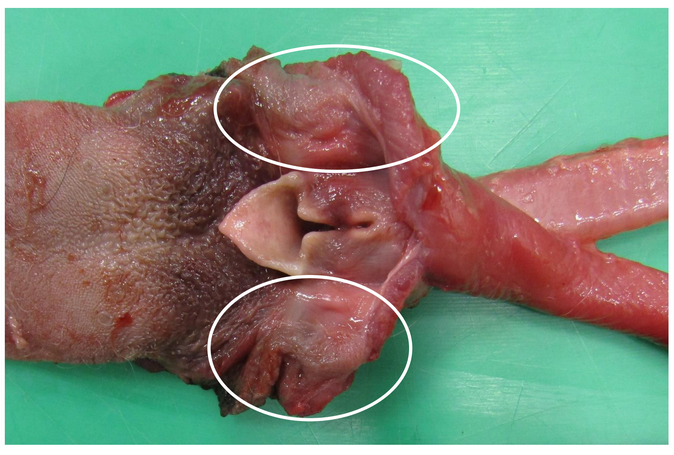

Figure 2. Opened oropharynx/laryngopharynx. Abnormal soft tissue directly continuing from the soft palate into the oesophagus, resting over the midline over the glottis (cut in half and circled).

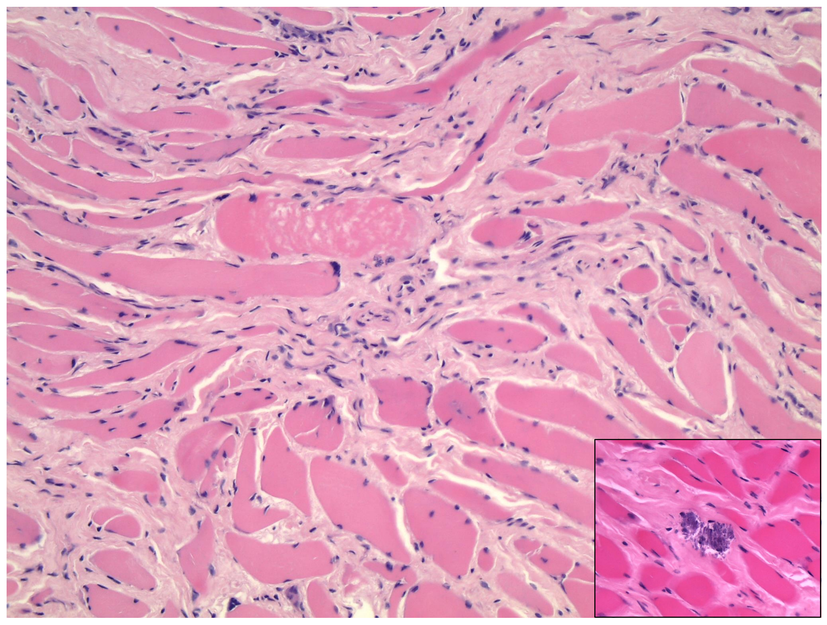

Figure 3. Oesophagus. Myofibre atrophy, degeneration and loss, with fibrosis and prominent satellite cells. Inset: Myofibre mineralisation. H&E, 10x.

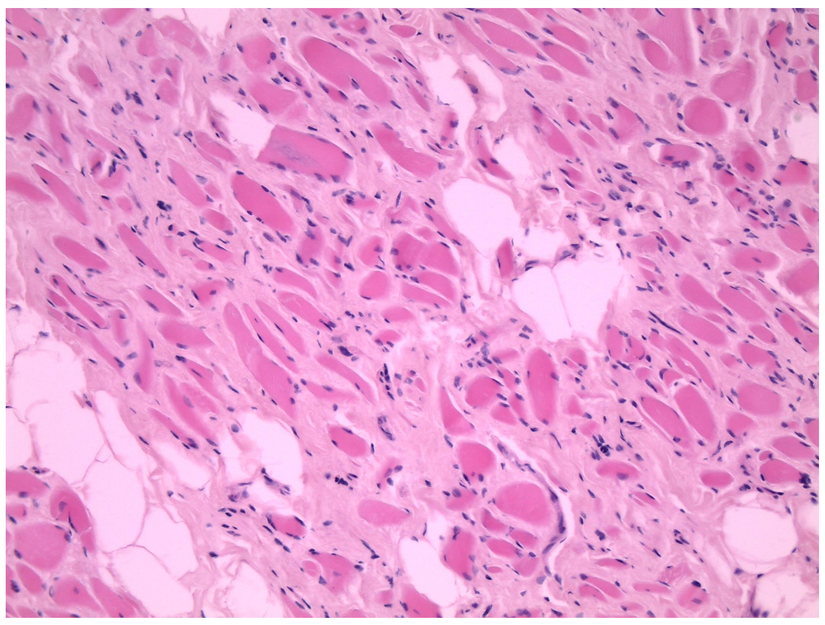

Figure 4. Diaphragm. Prominent myofibre atrophy with fibrosis and fibrofatty replacement. H&E, 10x.

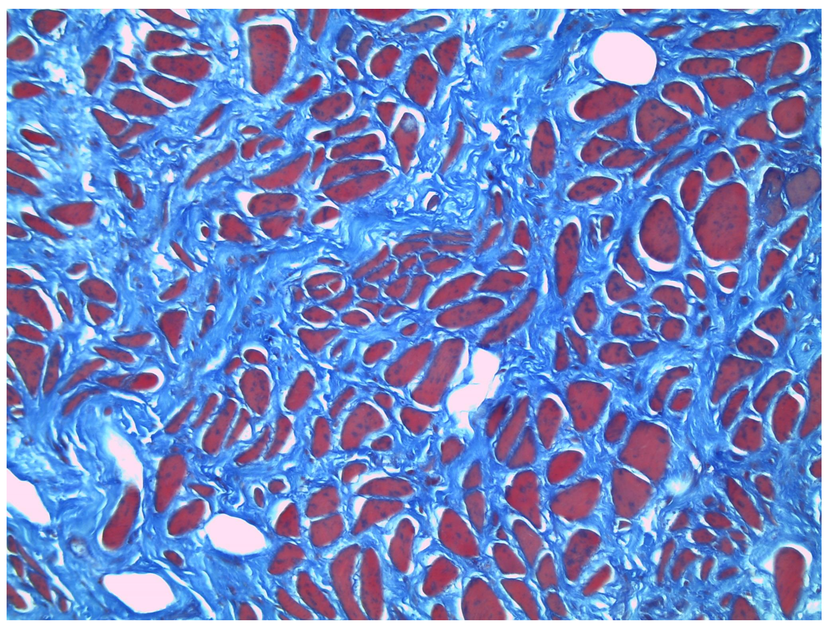

Figure 5. Oesophagus. Prominent myofibre atrophy and loss with multifocal, severe fibrosis. Masson's Trichrome, 10x.

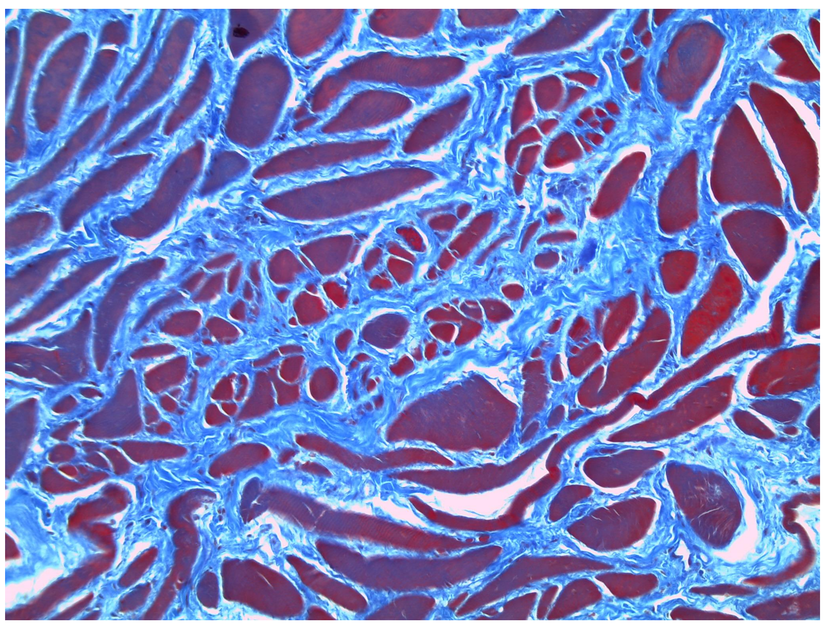

Figure 6. Diaphragm. Prominent myofibre atrophy and loss with multifocal, severe fibrosis. Masson's Trichrome, 10x.

Conclusions

Clinical, macroscopic, and histopathological findings are consistent with a case of stenotic nasopharyngeal dysgenesis accompanied by a form of MD. Caudal nasopharyngeal stenosis with obliteration of the communication between the nasopharynx and oropharynx was observed secondary to hypertrophic and abnormal palatopharyngeal muscles in continuity with the oesophagus. Although the developmental pathophysiology of this disorder is still unclear, primary palatopharyngeal dysgenesis is considered most likely.

Additionally, there was histological evidence of a MD. MD has been described in different dog breeds as inherited, degenerative myopathies related to defective muscular proteins, such as dystrophin and sarcoglycan.

To the authors' knowledge, this is the first report of nasopharyngeal dysgenesis associated with MD in dogs.

References

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