DDx: Do Not Forget Congenital Neuromuscular Disease

Muscular dystrophy (MD), a rare disease in cats, varies in clinical presentation. Dystrophin-deficient muscular dystrophy (DDMD) can only be definitively diagnosed through muscle biopsies and histopathology. Clinical signs of DDMD include gait abnormalities, appendicular muscle hypertrophy, regurgitation or dysphagia, and an enlarged tongue with white plaques caused by secondary mineralization of the tongue muscle. Bloodwork abnormalities include elevations of aspartate aminotransferase (AST), alanine aminotransferase (ALT), and creatine kinase (CK). Radiographs may show megaesophagus, diaphragmatic scalloping, adrenal calcification, and cardiomegaly.

A 7-month-old, male indoor domestic short-haired cat was presented for vomiting and open-mouth breathing at an emergency hospital. Physical examination revealed an enlarged tongue, abdominal discomfort, mild dehydration, increased bronchovesicular sounds, and tachypnea. The cat’s gait was unremarkable. Whole-body radiographs showed a diffuse megaesophagus, scalloped diaphragm, and hepatomegaly. Elevations in serum ALT, AST, and CK were identified. A presumptive diagnosis of feline MD was made, and the owners elected humane euthanasia because of poor prognosis.

Further testing confirmed DDMD caused by a novel gene mutation. Although MD is rare in cats, congenital neuromuscular disease should be considered in emergency cases. The authors recommend CK be added as part of the minimum database in emergency cases.

Commentary
This article reviews a novel genetic mutation and case presentation of DDMD, providing a good summary of the expected clinical presentation. Unique genetic mutations may lead to the absence of typical DDMD signs (eg, appendicular muscle hypertrophy). General practitioners should consider that although DDMD is rare, the common presentations for illness in this age group can overlap with DDMD clinical signs. As the condition is more common in young, male cats, signalment is an important consideration. CK levels should be included in sick cat or preoperative blood testing. Considerations for CK elevations include acute stress or struggling, but may also reflect conditions such DDMD. —Kelly St. Denis, DVM, DABVP (Feline)

Source

Grading Tumors

Soft tissue sarcoma (STS) represents 15% of all canine skin and subcutaneous tissue tumors. STS include fibrosarcomas, leiomyosarcomas, peripheral nerve sheath tumors, liposarcomas, and myxosarcomas. This study compared concordance of tumor grade between various pretreatment biopsy techniques with the gold standard of excisional biopsy. In this retrospective study, medical records of 70 dogs were reviewed. Needle core biopsy was performed in 19 cases, punch biopsy in 7 cases, and wedge biopsy in 44 cases. Tumors were histologically graded as I (27 cases), II (34 cases), or III (9 cases). Both pretreatment and excisional biopsy data were available for 68 of the 70 cases; a discordance rate of 41% was noted when grading using the 3-tier scheme, but only 13% when using a 2-tier (high grade vs low grade) grading system. Grade concordance was not significantly affected by the pretreatment biopsy method used.

Several biopsy methods were evaluated, and the method used did not appear to influence grading accuracy. This suggests that less aggressive biopsy techniques (eg, punch, TruCut), which can be performed under local anesthesia +/- sedation, are viable sampling methods. Usually a diagnosis of STS can be obtained with cytology, and biopsies are rarely needed. Treatment of choice for STS is wide surgical excision to achieve clean margins, and this should be pursued regardless of grade. In some high-grade tumor cases, pretreatment biopsy may be indicated. However, this study suggests that the biggest risk with biopsy grading is under-diagnosing and therefore under-treating high-grade tumors, which is hugely undesirable. —Cecilia Robat, DVM, DACVIM (Oncology)

Source