61st ANNUAL DIAGNOSTIC SLIDE SESSION 2020

CASE 2020 - 8

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Clinical history:

38-year-old female with the onset of symptoms approximately 10 months prior to biopsy, beginning with persistent post-exercise soreness in her legs. A PCP noted an elevated CK and she was referred to a rheumatologist. The patient showed a brief improvement on empiric steroids and hydroxychloroquine but then showed further decline. Methotrexate was added but discontinued because of mouth ulcers. She reported normal motor development and history of being athletic and hiking without any issues.

She experienced gradual progression of her weakness, with progression to her arms, trouble raising her arms, as well as bulbar symptoms of fatigue with chewing and jaw pain. Neurologic exam showed progressive proximal muscle weakness more prominent in legs than arms. EMG was normal. Neuromuscular specialists recommended a muscle biopsy, followed by high dose prednisone, mycophenolate mofetil, and IVIG, with possible mild improvement of symptoms.

Laboratory evaluations revealed negative or normal myositis panel, LFTs with AST/ALT of 106/91, and positive ANA and anti-DNA. The CK varied from 288 to 1089. The muscle biopsy showed reduced skeletal muscle carnitine levels, and normal acid and neutral maltase and CPT2 activities.

Material submitted: A cryosection of quadriceps muscle stained with H&E, combined anti-slow and fast myosin dual IHC for fiber typing (brown fibers=Type 1, red fibers=Type 2), and an electron micrograph.

Points for discussion:
1. Diagnosis
2. Therapeutic implications