

## 63<sup>rd</sup> ANNUAL DIAGNOSTIC SLIDE SESSION 2022

### CASE 2022 #5

#### Submitted by:

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#### Clinical History:

A 60-year-old woman was admitted to an outside hospital with severe generalized weakness and one year of dyspnea that progressed over two to three weeks to acute respiratory failure requiring intubation. A muscle biopsy of the left biceps was performed.

The patient was then transferred for a higher level of care and a more detailed medical history revealed that she had delayed motor milestones with ambulation at 18 months and scoliosis since childhood. She also described longstanding, very slowly progressive generalized weakness while remaining able to ambulate for short distances. On examination, she had facial, neck flexor, and limb weakness that was predominantly proximal in the upper limbs and both proximal and distal in the lower limbs. She had pes cavus and contractures at the wrists and ankles limiting dorsiflexion. EMG showed evidence of a severe diffuse myopathy with fibrillation potentials only in the vastus medialis. CK value was normal.

#### Material submitted:

H&E frozen section of left biceps muscle

#### Points for discussion:

1. Differential diagnosis
2. Ancillary studies
3. Genetic and acquired etiologies related to the differential diagnosis