64TH ANNUAL DIAGNOSTIC SLIDE SESSION 2023

CASE 2023-7

Submitted by:
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Clinical History:
A 48-year-old man with a "Marfanoid" body habitus (tall, with long arms and pectus carinatum) and a thin sharp face presented with right heart failure and mixed hypoxic/hypercapnic respiratory failure several months following an asymptomatic COVID infection. Although the patient denied muscle weakness, he reported longstanding difficulties reaching things overhead and jumping. There was no family history of a neuromuscular disorder. Physical examination showed slightly weakened facial muscles, mild bilateral tongue weakness, bilateral scapular winging, high steppage gait, inability to heel walk, positive Trendelenburg sign, and reduced muscle bulk in legs, pectoralis, deltoid, triceps, and biceps muscles. The pulmonary function tests showed a restrictive pattern. The severity of hypercapnia raised the possibility that his respiratory failure had neuromuscular etiology, so the neurology and genetics teams were consulted. Electrodiagnostic testing provided evidence for a myopathic process without membrane irritability, affecting both the upper and lower extremities. CK levels were within normal limits. The patient was evaluated for Marfan syndrome when he was 14 years old, but the tests done at the time were unrevealing. Genetics recommended a muscle biopsy to guide the further genetic testing strategy, and a deltoid muscle biopsy was performed.

Material submitted:
1. One (1) H&E-stained cryosection
2. One (1) modified Gomori trichrome-stained cryosection

Points for discussion:
1. Differential diagnosis.
2. Ancillary studies and molecular findings.