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CASE 2017 - 8

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Clinical History:

This infant boy was born by C-section at 41 weeks gestation after a normal pregnancy, and his 2 month well child exam was normal.

However by 3 months of age, his parents had observed him moving less and having difficulty holding his head up. On exam, he had significant hypotonia and weakness, greater proximally than distally. An MRI showed diffuse, mildly increased T2 signal in his muscles. Electromyography (EMG) showed fibrillations and low amplitude motor units with polyphasic waves consistent with myopathy. Nerve conduction and neuromuscular junction studies were normal. His weakness progressed, leading to difficulty with eating and respiration; he had episodes of aspiration. His serum creatinine kinase levels ranged from 3170 to 4944 U/L. There is no family history of neuromuscular disease. He was hospitalized multiple times, became dependent on tube feedings and respiratory support, and passed away at 6 months of age.

A right quadriceps muscle biopsy was performed at 4 months of age.

Material submitted:

H&E stained cryosection of muscle

Points for discussion:

1. Differential diagnosis
2. Approach to diagnostic testing