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CASE 2018-3

Submitted By:
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Clinical History:
2 year old male with mild global developmental delay, frequent falls, and oropharyngeal dysphagia. Neurologic exam revealed a positive Gowers' sign and abnormal gait consistent with a compensated Trendelenburg. His family history was positive for a maternal uncle with unknown neuromuscular disease requiring the use of a wheelchair since age 12. The serum CK was elevated in the range of 1300-1400 U/L.

Genetic testing prior to muscle biopsy included a non-diagnostic microarray, normal *DMD* del/dup testing and complete sequencing, normal GAA enzymatic assay, and negative congenital hypotonia next generation sequencing panel.

Material Submitted:
H&E stained cryosection of muscle

Points for Discussion:
1. Differential diagnosis
2. Approach to diagnostic testing