

57th ANNUAL DIAGNOSTIC SLIDE SESSION 2016

CASE 2016-1

Submitted by:

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Clinical History:

A 9-year-old boy has a past medical history significant for cerebral palsy, inturning right foot, and a left clubfoot that underwent surgery at 3 years of age. He was crawling before the surgery, was able to walk with a walker afterwards, and ultimately walk independently. His parents report a 6-month history of increasing weakness that is both proximal and distal on examination. He has difficulty rising from the floor and uses a Gower-type maneuver by pulling up on a chair. He shows a steppage gait that is slightly wide based. Stretch reflexes are absent. His serum creatine kinase (CK) is 2800 IU/L. Electromyography (EMG) shows complex repetitive discharges in the gastrocnemius. There is no family history of neuromuscular problems.

A left quadriceps muscle biopsy is performed.

Material submitted:

H&E stained cryosection of muscle

Points for discussion:

- 1) Approach to diagnostic testing
- 2) Differential diagnosis