

53rd ANNUAL DIAGNOSTIC SLIDE SESSION 2012

CASE 2012-1

Submitted by:

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Clinical Presentation:

A 7-year-old boy with mild scoliosis was evaluated in the neuromuscular clinic for fine and gross motor difficulties. He cannot run, jump, or throw a ball. He also has difficulty with stairs, buttons, and zippers. Regarding his developmental milestones, he began walking at 16 months. At 18 months, he had progressively more falls. Outside of his motor difficulties, there are no other concerns. He is cognitively normal. He had a brain MRI which was normal and an electromyogram that showed myopathic features. He has no family history of nerve or muscle abnormalities.

On physical examination, he was non-dysmorphic in appearance. Cardiac exam revealed a regular rate and rhythm and no murmurs. His cranial nerves were intact bilaterally. He had normal muscle bulk, though tone was mildly decreased. He had mild weakness, proximal greater than distal. He had Gowers' sign. Deep tendon reflexes were present but decreased throughout. Sensory examination was within normal limits. A biopsy of the right vastus lateralis muscle was obtained. Histopathologic examination revealed abnormalities. The tissue was then sent for genetic testing.

Material submitted:

Three Virtual sections of right vastus lateralis muscle

One H&E-stained section

One toluidine blue-stained section

One Gomori trichrome-stained section

Points for discussion:

1. Discuss the differential diagnosis of congenital myopathies.
2. Review genetic alterations in various congenital myopathies.