CASE 2012-1

SUBMITTED BY:
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CLINICAL PRESENTATION:
L. C. is a 7 year-old Chinese boy with mild scoliosis who 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. While in China, 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.