Canadian Association of Neuropathologists
L'Association Canadienne des Neuropathologistes

CASE No 11

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This 30 year-old woman died of complications of her myopathy. She had been hypotonic since birth, had severe generalised muscle atrophy and weakness, facial weakness, mild scoliosis, and normal cardiac function. Limb weakness was both proximal and distal. She walked independently with a steppage gait. Intelligence was normal. Her disease was slowly progressive.

There was no family history of neuromuscular disease. Chromosome analysis showed normal female karyotype 46, XX. Genetic classification of her disease had not been possible. She underwent a skeletal muscle biopsy at 12 years of age.

Autopsy findings included dolichocephaly but otherwise unremarkable brain and spinal cord. Skeletal muscle was sampled.

Material submitted:
Photographs of muscle biopsy at 12 years of age.
Slides (2) of skeletal muscle at autopsy: hematoxylin & eosin, toluidine blue.

Questions: What is the diagnosis of the muscle biopsy at 12 years of age?
Is the diagnosis at autopsy the same as the original diagnosis or modified?