A 38-year-old man presented with a 2-year history of dyspnea resulting in hypercarbic respiratory failure. CT scan of the chest displayed atrophy of the thoracic paraspinal and chest wall muscles (figure). He had neck and abdominal muscle weakness with normal limbs strength. Creatine kinase was increased (704 IU/L; normal 0 to 240). Electromyography showed insertional myotonic discharges in paraspinal muscles without voluntary motor unit activity. Biopsy of the deltoid muscle demonstrated scarce lysosomal glycogen accumulation. Acid-alpha-glucosidase activity in a dried blood spot was reduced (0.59 to 3.88 pmol/punch/hour; normal: 10.00 to 48.96).

Adult-onset Pompe disease is a rare, potentially treatable glycogen storage disorder.1 It may be overlooked because of the preferential involvement of the axial muscles resulting in truncal muscle weakness and respiratory failure.2

REFERENCES
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