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
Teaching NeuroImage: Axial muscle atrophy in adult-onset Pompe disease
Bashar Katirji, Vita Kesner, Rana B. Hejal, et al.
Neurology 2008;70;e36
DOI 10.1212/01.wnl.0000304251.98684.69

This information is current as of March 3, 2008

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/70/10/e36.full

References
This article cites 2 articles, 2 of which you can access for free at:
http://n.neurology.org/content/70/10/e36.full#ref-list-1

Subspecialty Collections
This article, along with others on similar topics, appears in the following collection(s):
CT
http://n.neurology.org/cgi/collection/ct
Muscle disease
http://n.neurology.org/cgi/collection/muscle_disease

Permissions & Licensing
Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
http://www.neurology.org/about/about_the_journal#permissions

Reprints
Information about ordering reprints can be found online:
http://n.neurology.org/subscribers/advertise

Neurology ® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.