Teaching NeuroImages: Amyloid myopathy

Not your usual suspects

Marcus V. Pinto, MD, MS, Jennifer A. Tracy, MD, and Teerin Liewluck, MD

Neurology® 2020;95:e1290-e1291. doi:10.1212/WNL.00000000000010107

Correspondence

Dr. Pinto
pinto.marcus@mayo.edu

Figure Interstitial amyloid deposits involving skeletal muscle, necrotic muscle fibers, and markedly reduced dysferlin immunoreactivity

Frozen sections of left vastus medialis muscle show (A, B) congophilic deposits in perimysial blood vessels (arrows) and in adjacent endomysium encasing muscle fibers (arrowheads) on a Congo red stained section viewed under (A) light microscopy and (B) rhodamine optics. (C) Hematoxylin & eosin-stained section shows necrotic fibers replaced by macrophages (arrows). (D) Sarcolemmal dysferlin immunoreactivity is markedly reduced on patient's muscle fibers compared to the control section.

A 31-year-old man presented with a 6-year history of slowly progressive calf atrophy and weakness. EMG showed distal myopathy with fibrillation potentials. Creatine kinase (CK) was 2,848 U/L (normal <310). Muscle biopsy showed necrotic fibers and interstitial amyloid deposits (figure). The search for extramuscular amyloidosis was unrevealing. Next-generation sequencing identified known pathogenic c.6124C>T and novel c.145-1G>A variants in the dysferlin gene (*DYSF*). Intramuscular interstitial amyloid deposits can occur in systemic amyloidosis (AL or ATTR)¹ or less commonly in muscular dystrophies (*DYSF* and *ANOS*).² Longstanding symptoms, young age at onset, calf atrophy, markedly elevated CK, and lack of systemic involvement are suggestive of muscular dystrophies.²

MORE ONLINE

→Teaching slides
links.lww.com/WNL/B144

From the Department of Neurology, Mayo Clinic, Rochester, MN.

Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

Study funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

Appendix Authors

Name	Location	Contribution
Marcus V. Pinto, MD, MS	Mayo Clinic, Rochester, MN	Data collection, drafting of final manuscript, approval and critical review of final form

Appendix *(continued)*

Name	Location	Contribution
Jennifer A. Tracy, MD	Mayo Clinic, Rochester, MN	Data collection, approval and critical review of final form
Teerin Liewluck, MD	Mayo Clinic, Rochester, MN	Data collection, drafting of final manuscript, approval and critical review of final form

References

1. Prayson RA. Amyloid myopathy: clinicopathologic study of 16 cases. *Hum Pathol* 1998;29:463–468.
2. Liewluck T, Milone M. Characterization of isolated amyloid myopathy. *Eur J Neurol* 2017;24:1437–1445.

Neurology®

Teaching NeuroImages: Amyloid myopathy: Not your usual suspects

Marcus V. Pinto, Jennifer A. Tracy and Teerin Liewluck

Neurology 2020;95:e1290-e1291 Published Online before print July 1, 2020

DOI 10.1212/WNL.0000000000010107

This information is current as of July 1, 2020

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/95/9/e1290.full
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/95/9/e1290.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): 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 © 2020 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.