Teaching NeuroImages: Amyloid myopathy

Not your usual suspects

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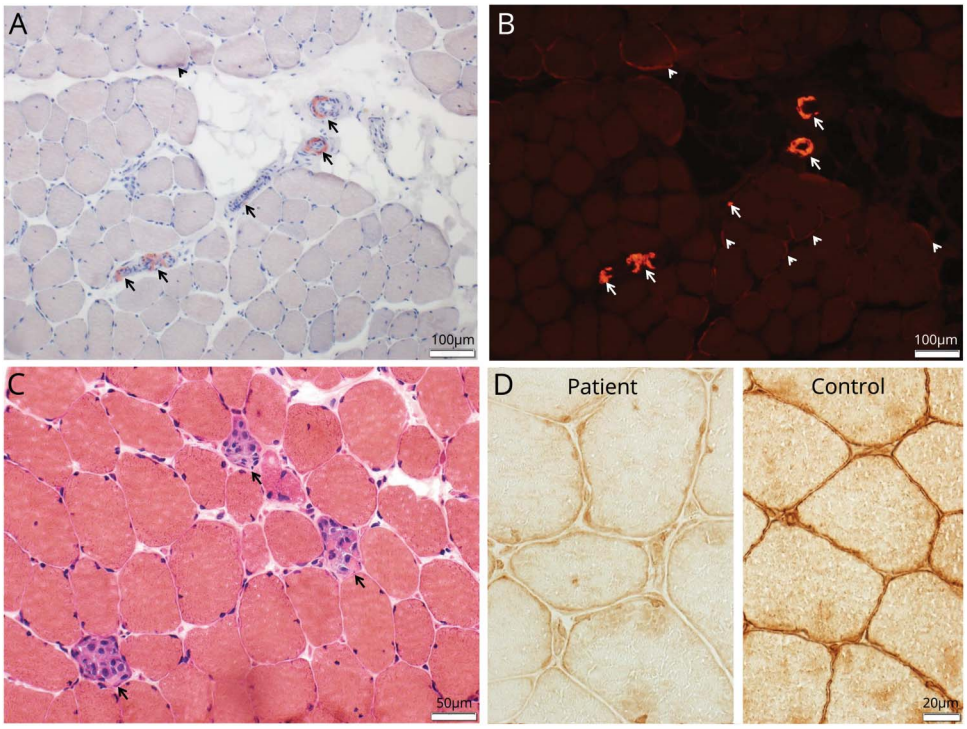
Figure Interstitial amyloid deposits involving skeletal muscle, necrotic muscle fibers, and markedly reduced dysferlin immunoreactivity



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 *ANO5*).[2] Longstanding symptoms, young age at onset, calf atrophy, markedly elevated CK, and lack of systemic involvement are suggestive of muscular dystrophies.[2]

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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
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.

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