

# Teaching NeuroImages: Neuromyopathy in a patient with hereditary transthyretin Thr60Ala amyloidosis

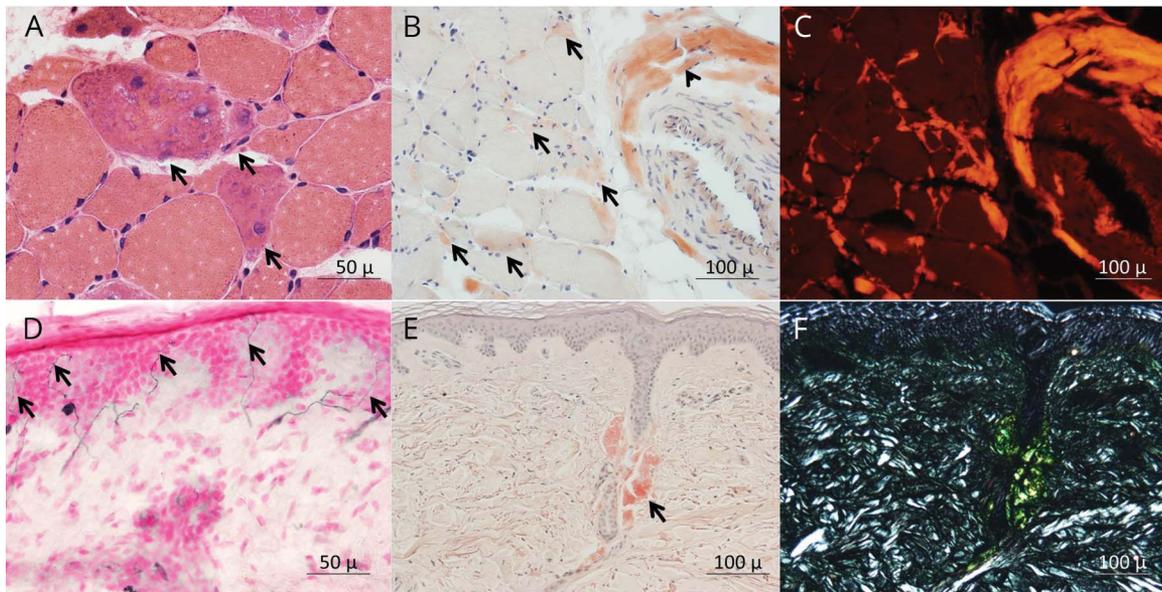
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**Figure** Amyloid deposits in skeletal muscle and skin in a patient with hATTR amyloidosis



(A–C) Frozen sections of right gluteus medius muscle show (A) regenerating muscle fibers on hematoxylin & eosin stained-section and (B) extensive congophilic amorphous deposits adjacent to a large perimysial vessel (arrowhead) and in the endomysium surrounding many muscle fibers (arrows) on a Congo red–stained section (C) with red fluorescence viewed under rhodamine optics. (D–F) Skin punch biopsy of the right dorsal foot shows (D) numerous epidermal nerve fibers (arrows) with a mean intraepidermal nerve fiber density of 2.0 fibers/mm ( $n > 0.2$  fibers/mm) on PGP 9.5 and (E) large congophilic amorphous deposits around a dermal vessel on Congo red preparation (F) with apple-green birefringence viewed under polarized light.

A 69-year-old man with hereditary transthyretin (hATTR) Thr60Ala amyloid cardiomyopathy presented with a 2-year history of proximal greater than distal weakness and pan-modality sensation loss in the lower extremities. Nerve conduction studies/EMG revealed a proximal myopathy and length-dependent axonal polyneuropathy. Muscle biopsy was diagnostic of amyloid myopathy (figure) and skin biopsy showed large amyloid deposits but normal intraepidermal nerve fiber density (figure). Myopathy is rare in hATTR amyloidosis and concomitant polyneuropathy is always present.<sup>1</sup> Skin biopsy is less invasive than nerve biopsy and has good sensitivity for amyloid diagnosis.<sup>2</sup> hATTR amyloidosis is treatable and should be included in the differential diagnosis of a neuromyopathy.

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Go to [Neurology.org/N](https://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.

## Disclosure

M. Pinto, J. Tracy, M. Grogan, and M. Mauermann report no disclosures relevant to the manuscript. Go to [Neurology.org/N](http://Neurology.org/N) for full disclosures.

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2. Ebenezer GJ, Liu Y, Judge DP, et al. Cutaneous nerve biomarkers in transthyretin familial amyloid polyneuropathy. *Ann Neurol* 2017;82:44–56.

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## Appendix Authors

Name	Location	Role	Contribution
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