Teaching NeuroImages: Slowly progressive hypertrophic brachial plexopathy due to *SEPT9* mutation

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A 51-year-old man had slowly progressive muscle wasting, weakness, and paresthesia of the right upper limb since age 21 years. His deceased mother had a single episode of painful right upper limb paresis. Examination showed severe amyotrophy and hypoesthesia of the right upper limb. Neuroimaging studies disclosed marked involvement of the right brachial plexus (figures 1 and 2). Neurophysiologic studies disclosed severe right brachial plexopathy. Gene panel for inherited neuropathies disclosed pathogenic variant c.278C>T (p.Ser93Phe) in the *SEPT9* gene,[1,2] confirming the diagnosis of hereditary neuralgic amyotrophy.[1,2] *SEPT9*-related disorders can present as autosomal dominant recurrent or progressive hypertrophic brachial plexus neuropathy.[1,2]

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Full consent was obtained from the patient for the case report. This study was approved by our institutional ethics committee (CEP-UNIFESP/HSP).

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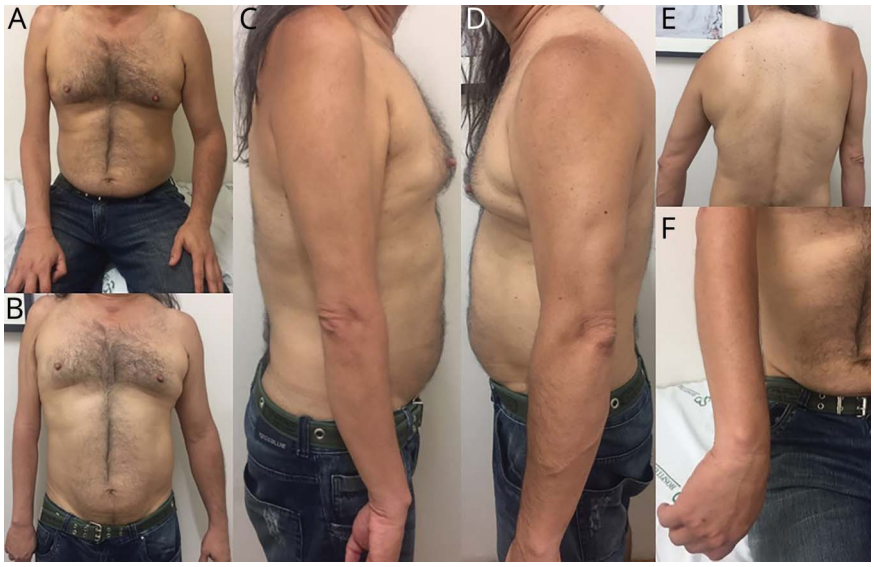
Figure 1 Examination in *SEPT9*-related brachial plexus neuropathy



Evaluation shows marked amyotrophy of the right upper limb (A and B), mainly in the scapular girdle (C–E), arm, and (F) minor extent in the forearm and hand.

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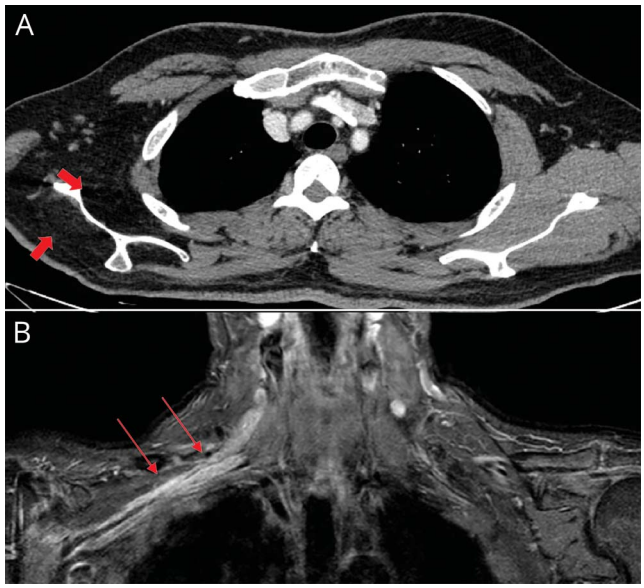
Figure 2 Muscle and brachial plexus imaging studies



(A) Chest CT scan shows marked global amyotrophy and liposubstitution (wide red arrows) of the right scapular girdle muscle groups. (B) Coronal MRI of the brachial plexus shows asymmetric hyperintense signal and hypertrophy of right brachial plexus anterior roots, trunks, and cords in short tau inversion recovery sequence (red arrows).

Disclosure

The authors report no relevant disclosures. Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Location	Contribution
Paulo Victor Sgobbi de Souza, MD	Federal University of São Paulo (UNIFESP), Brazil	Case report project (conception, organization, execution), writing of the first draft, review and critique
Eduardo Augusto Gonçalves, MD	Federal University of São Paulo (UNIFESP), Brazil	Case report project (conception, organization, execution), writing of the first draft, review and critique
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