Teaching NeuroImages: Spinal xanthomatosis

A misdiagnosed, treatable cause of progressive myelopathy

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Figure 1 Photograph of the patient's Achilles tendons

Enlargement of Achilles tendons bilaterally was suggestive of tendon xanthomas.

A 57-year-old woman presented with a 20-year history of progressive lower extremity weakness, spasticity, and proprioception deficits. She was given a diagnosis of primary progressive multiple sclerosis at age 38. Her Achilles tendons were enlarged (figure 1). Brain MRI was normal. Spine MRI demonstrated T2-hyperintense signal involving the posterior and lateral columns (figure 2). Serum cholestanol level was elevated. *CYP27A1* gene sequencing revealed 2 pathogenic variants, c.1183C>T(p.Arg395Cys) and c.410G>A(p.Arg137Gln), confirming the diagnosis of cerebrotendinous xanthomatosis (CTX). Spinal xanthomatosis is a rare variant of CTX presenting with progressive corticospinal and posterior column signs.[1] Early treatment with chenodeoxycholic acid may improve outcomes.[2]

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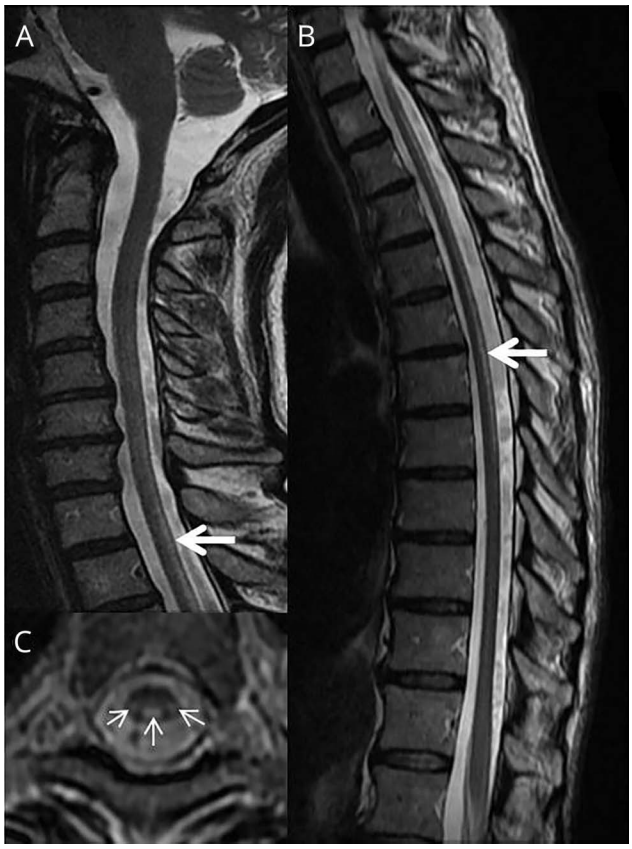
Figure 2 Spinal MRI



T2-weighted cervical sagittal (A), thoracic sagittal (B), and axial (C) images demonstrate T2-hyperintense signal abnormality involving the posterior and lateral columns from C2 to T8–T9.

Appendix Authors

Name	Location	Contribution
Cristina Valencia-Sanchez, MD	Mayo Clinic, Phoenix, AZ	Case concept and design, acquisition and interpretation of data, manuscript writing
Dean M. Wingerchuk, MD	Mayo Clinic, Phoenix, AZ	Case concept and design, interpretation of data, critical revision of manuscript for intellectual content
Rhadika Dhamija, MD	Mayo Clinic, Phoenix, AZ	Case concept and design, acquisition and interpretation of data, critical revision of manuscript for intellectual content

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