# Teaching NeuroImages: Spinal xanthomatosis

## A misdiagnosed, treatable cause of progressive myelopathy

Cristina Valencia-Sanchez, MD, Dean M. Wingerchuk, MD, and Radhika Dhamija, MD

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**Correspondence**
Dr. Dhamija
dhamija.radhika@mayo.edu

**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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## Disclosure

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

From the Neurology Department, Mayo Clinic, Phoenix, AZ.

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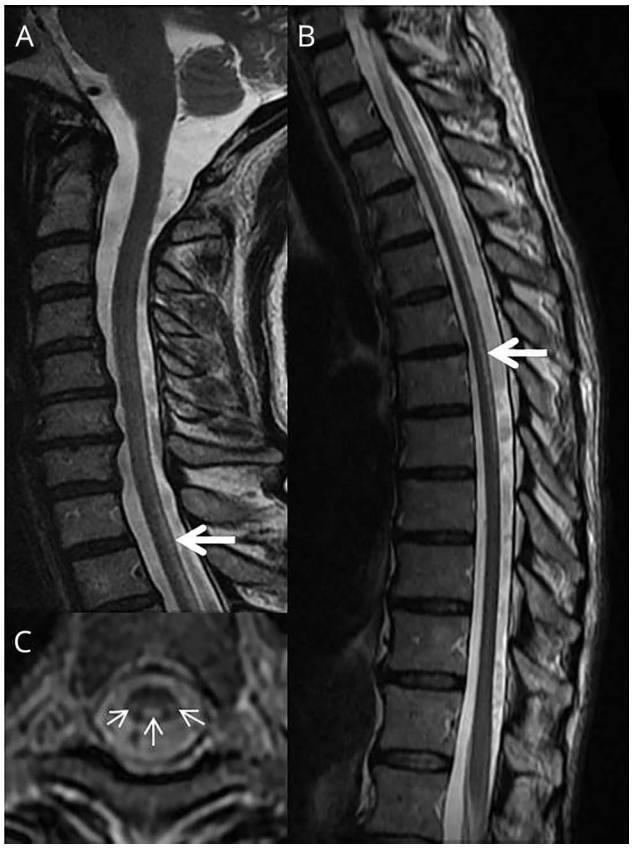
**Figure 2** Spinal MRI




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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