NEUROIMAGES

Diagnostic challenge

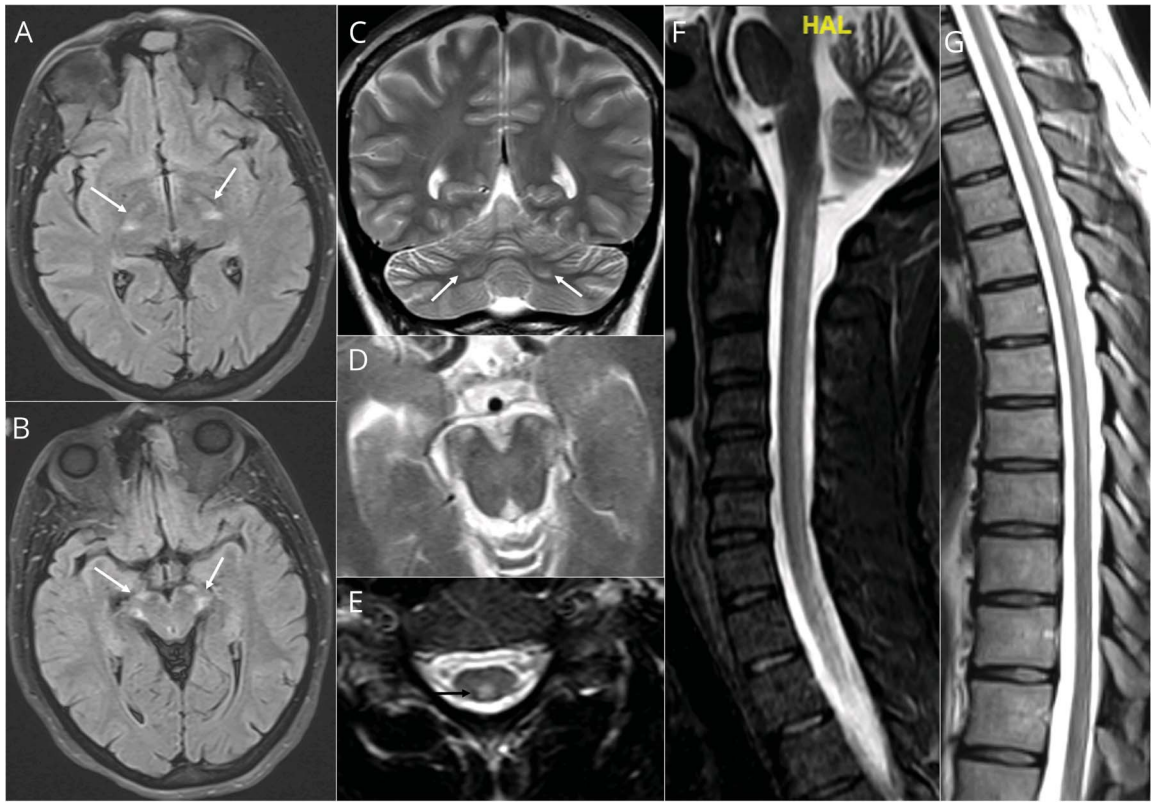
A case of late-onset spinal form cerebrotendinous xanthomatosis

Dilek Mutlu, MD, Aslı Tuncer, MD, Rahsan Gocmen, MD, Gul Yalcin-Cakmakli, MD, Serap Saygı, MD, and Bulent Elibol, MD, PhD

Correspondence
Dr. Elibol
bulentelibol@gmail.com

Neurology® 2019;92:438-439. doi:10.1212/WNL.0000000000007015

Figure Imaging



Axial fluid-attenuated inversion recovery (A, B) and T2-weighted (D) MRI show bilateral symmetrical hyperintensity in internal capsule and crus cerebri; T2-weighted images demonstrate longitudinally extensive hyperintensity within dorsal columns (E–G) and mild hyperintensity in both dentate nuclei (C) (arrows), strongly suggesting radiologic diagnosis of the spinal form of cerebrotendinous xanthomatosis.

A 42-year-old woman was admitted with lower extremity weakness slowly progressing over 10 years. Examination showed spastic paraparesis, deep sensory deficit on legs, and mild intentional tremor of hands. Extensive serum and CSF analyses were unremarkable. Bilateral symmetrical internal capsule, crus cerebri, and longitudinal spinal cord lesions and particularly slight involvement of dentate nuclei on MRI (figure), together with history of parental consanguinity, suggested spinal form of cerebrotendinous xanthomatosis, despite absence of all cardinal systemic features, including xanthomas.[1] Diagnosis was revealed by high plasma cholestanol level (15 μg/mL, normal range 0.45–3.75) and novel homozygous mutation (p.L524R [c.1571T > G]) on *CYP27A1* gene by DNA sequencing. Chenodeoxycholic acid treatment was started.

Study funding

No targeted funding reported.

From the Departments of Neurology (D.M., A.T., G.Y.-C., S.S., B.E.) and Radiology (R.G.), Faculty of Medicine, Hacettepe University, Sihhiye, Ankara, Turkey.

Go to 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

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

Appendix Authors

Name	Location	Role	Contribution
Dilek Mutlu, MD	Hacettepe University, Ankara	Author	Examined the patient, planned the evaluation for differential diagnosis, wrote the manuscript
Aslı Tuncer, MD	Hacettepe University, Ankara	Author	Examined the patient, planned the evaluation for differential diagnosis, revised the manuscript
Rahsan Gocmen, MD	Hacettepe University, Ankara	Author	Interpreted the brain and spinal cord MRI, revised the manuscript, prepared the figure
Gul Yalcin-Cakmakli, MD	Hacettepe University, Ankara	Author	Examined the patient, wrote and revised the manuscript
Serap Saygı, MD	Hacettepe University, Ankara	Author	Planned the evaluation for differential diagnosis, revised the manuscript
Bülent Elibol, MD, PhD	Hacettepe University, Ankara	Author	Examined and diagnosed the patient, wrote and revised the manuscript

Reference

1. Pilo-de-la-Fuente B, Jimenez-Escrig A, Lorenzo JR, et al. Cerebrotendinous xanthomatosis in Spain: clinical, prognostic, and genetic survey. Eur J Neurol 2011;18:1203–1211.

Diagnostic challenge: A case of late-onset spinal form cerebrotendinous xanthomatosis

Dilek Mutlu, Asli Tuncer, Rahsan Gocmen, et al.

Neurology 2019;92;438-439
DOI 10.1212/WNL.0000000000007015