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 Saygi, MD, and Bulent Elibol, MD, PhD


Correspondence
Dr. Elibol
bulenteilibol@gmail.com

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. 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.

Reference

Appendix Authors

<table>
<thead>
<tr>
<th>Name</th>
<th>Location</th>
<th>Role</th>
<th>Contribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dilek Mutlu, MD</td>
<td>Hacettepe University, Ankara</td>
<td>Author</td>
<td>Examined the patient, planned the evaluation for differential diagnosis, wrote the manuscript</td>
</tr>
<tr>
<td>Asli Tuncer, MD</td>
<td>Hacettepe University, Ankara</td>
<td>Author</td>
<td>Examined the patient, planned the evaluation for differential diagnosis, revised the manuscript</td>
</tr>
<tr>
<td>Rahsan Gocmen, MD</td>
<td>Hacettepe University, Ankara</td>
<td>Author</td>
<td>Interpreted the brain and spinal cord MRI, revised the manuscript, prepared the figure</td>
</tr>
<tr>
<td>Gul Yalcin-Cakmakli, MD</td>
<td>Hacettepe University, Ankara</td>
<td>Author</td>
<td>Examined the patient, wrote and revised the manuscript</td>
</tr>
<tr>
<td>Serap Saygi, MD</td>
<td>Hacettepe University, Ankara</td>
<td>Author</td>
<td>Planned the evaluation for differential diagnosis, revised the manuscript</td>
</tr>
<tr>
<td>Bülent Elibol, MD, PhD</td>
<td>Hacettepe University, Ankara</td>
<td>Author</td>
<td>Examined and diagnosed the patient, wrote and revised the manuscript</td>
</tr>
</tbody>
</table>

Subspecialty Alerts by E-mail!

Customize your online journal experience by signing up for e-mail alerts related to your subspecialty or area of interest. Access this free service by clicking on the “My Alerts” link on the home page. An extensive list of subspecialties, methods, and study design choices will be available for you to choose from—allowing you priority alerts to cutting-edge research in your field!

The AAN Has Your Back!

Every day, the AAN is fighting for you. From actively lobbying members of Congress for common sense legislation, to meeting with regulators to demonstrate the value of neurology and reduce regulatory hassles, the Academy is forcefully countering any threats to your profession and patient access to care. Learn more at AAN.com/policy-and-guidelines/advocacy, read the bimonthly Capitol Hill Report and monthly AANnews® member magazine, and respond to Advocacy Action Alert emails when we invite you to share your voice with Congress. Get into the conversation at #AANAdvocacy.
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

This information is current as of February 25, 2019