

Teaching NeuroImage: Symmetric Deep Cerebellar White Matter T2 and Susceptibility-Weighted Imaging Hypointense Lesions in a Case of Cerebrotendinous Xanthomatosis

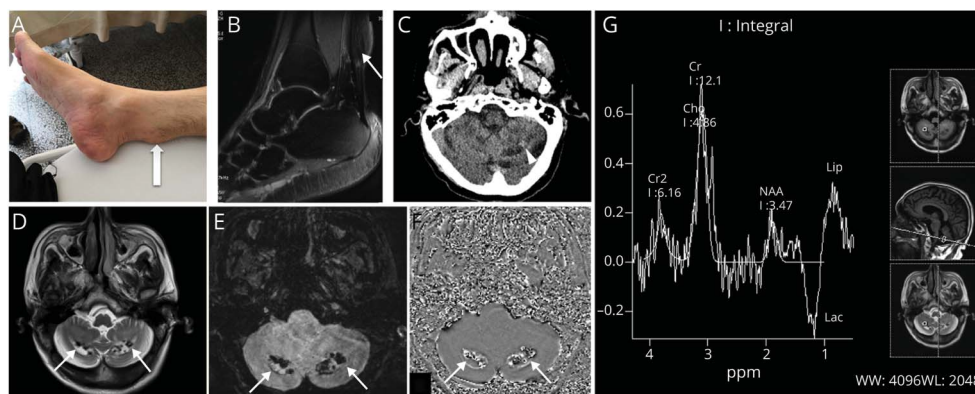
Yue Zhang, MD, PhD,* Yi-Min Sun, MD, PhD,* and Haiqing Li, MD, PhD

Neurology® 2021;97:e968-e969. doi:10.1212/WNL.00000000000012154

Correspondence

Dr. Li
lihaiqing@fudan.edu.cn

Figure Clinical, Imaging, and Magnetic Resonance Spectroscopy (MRS) Findings



(A, B) Enlargement of Achilles tendon. (C) Brain CT shows modest hyperdensity in cerebellar hypodense lesions. (D) T2-weighted imaging reveals hyperintensity with spots of hypointensity, which were more evident on susceptibility-weighted imaging (E, F). (G) MRS demonstrates decrease in *N*-acetylaspartate (NAA) and increase in lipid (Lip) and lactate (Lac) peaks.

A 39-year-old man presented with worsening slurred speech, difficulty walking, and falls for 2 years. Examination found severe ataxia and enlargement of Achilles tendons. Brain CT demonstrated modest hyperdensity in cerebellar hypodense lesions. T2-weighted imaging revealed hypointensity in deep cerebellar white matter, more evident on susceptibility-weighted imaging. Magnetic resonance spectroscopy suggested lipid storage and mitochondrial dysfunction. The diagnosis of cerebrotendinous xanthomatosis (CTX) was confirmed by gene screening of *CYP27A1* (Figure).

The typical imaging finding of CTX is T2-weighted imaging hyperintensity in dentate nucleus. Cerebellar hypointensity is occasionally seen in the late stage and indicates deposition of hemosiderin and microhemorrhages,¹ which may be secondary to cerebellar vacuolation.²

Study Funding

The authors report no targeted funding.

MORE ONLINE

Teaching slides

links.lww.com/WNL/B408

*These authors contributed equally.

From the Departments of Neurology (Y.Z., Y.-M. Sun) and Radiology (H.L.), Huashan Hospital, Fudan University, Shanghai, China.

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](https://www.neurology.org/N) for full disclosures.

Appendix Authors

Name	Location	Contribution
Yue Zhang, MD, PhD	Department of Neurology, Huashan Hospital, Fudan University, Shanghai, China	Patient management, analysis of the radiologic data, preparation of the draft manuscript
Yi-Min Sun, MD, PhD	Department of Neurology, Huashan Hospital, Fudan University, Shanghai, China	Critical review of manuscript, language modification

Appendix *(continued)*

Name	Location	Contribution
Haiqing Li, MD, PhD	Department of Radiology, Huashan Hospital, Fudan University, Shanghai, China	Concept and design of the study, analysis of the radiologic data, critical review, final approval of the manuscript to be published

References

1. Barkhof F, Verrips A, Wesseling P, et al. Cerebrotendinous xanthomatosis: the spectrum of imaging findings and the correlation with neuropathologic findings. *Radiology*. 2000;217(3):869-876.
2. Mignarri A, Dotti MT, Federico A, et al. The spectrum of magnetic resonance findings in cerebrotendinous xanthomatosis: redefinition and evidence of new markers of disease progression. *J Neurol*. 2017;264(5):862-874.

Neurology[®]

Teaching NeuroImage: Symmetric Deep Cerebellar White Matter T2 and Susceptibility-Weighted Imaging Hypointense Lesions in a Case of Cerebrotendinous Xanthomatosis

Yue Zhang, Yi-Min Sun and Haiqing Li

Neurology 2021;97:e968-e969 Published Online before print May 4, 2021

DOI 10.1212/WNL.0000000000012154

This information is current as of May 4, 2021

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/97/9/e968.full
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/97/9/e968.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): MRI http://n.neurology.org/cgi/collection/mri
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2021 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

