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

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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

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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

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