Teaching NeuroImages: Symmetric Deep Cerebellar White Matter T2 and SWI Hypointense Lesions in a Case of Cerebrotendinous Xanthomatosis

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Case summary
A 39-year-old man presented with worsening slurred speech, difficult walking, and falls for 2 years. Examination found severe ataxia and enlargement of Achilles tendons. Brain CT demonstrated modest hyperdensity in cerebellar hypodense lesions. T2WI revealed hypointensity in deep cerebellar white matter, which were more evident on SWI. MRS suggested lipid storage and mitochondrial dysfunction (Figure). The diagnosis of cerebrotendinous xanthomatosis (CTX) was confirmed by gene screening of CYP27A1.

Typical imaging finding of CTX is T2WI hyperintensity in dentate nucleus. Cerebellar hypointensity is occasionally seen in late stage. It indicates deposition of hemosiderin, micro-hemorrhages, which may be secondary to cerebellar vacuolation.

Appendix 1. Authors

<table>
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References:

Figure legend
Figure. Enlargement of Achilles tendons (A, B). Brain CT showed modest hyperdensity in cerebellar hypodense lesions (C). T2WI (D) revealed hyperintensity with spots of hypointensity, which were more evident on SWI (E, F). MRS demonstrated decrease in NAA (N-acetylaspartate) and increase in Lip (lipid) and Lac (lactate) peaks (G).
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