Brain and liver iron accumulation in aceruloplasminemia

A 68-year-old woman with diabetes, anemia, and retinal degeneration developed mild cognitive impairment and subtle gait ataxia. Her parents were nonconsanguineous, and her sister had diabetes. \( T2^*\)-weighted brain MRI showed symmetric hypointensity in the striatum, thalamus (A, D), red nucleus (B, E), and dentate nucleus (C, F), and on the brain surface (A-F).

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in the ceruloplasmin gene, confirming the diagnosis of aceruloplasminemia. This case highlights the importance of T2*-weighted MRI in diagnosing aceruloplasminemia, even in patients with minimal neurologic symptoms.

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Study funding: No targeted funding reported.

Disclosures: The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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Koji Fujita, Yusuke Osaki, Masafumi Harada, et al.
Neurology 2013;81;2145-2146
DOI 10.1212/01.wnl.0000437304.30227.bd

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