Neurodegeneration with brain iron accumulation in aceruloplasminemia

A 55-year-old African Canadian man with insulin-dependent diabetes mellitus and alcohol abuse presented with diabetic ketoacidosis. Progressive cognitive decline over the previous 5 years resulted in long-term care placement. Aside from pigmentary retinopathy, general examination was unremarkable. MRI demonstrated iron accumulation in the brain (figure 1) and liver (figure 2A). Ceruloplasmin, a ferroxidase enzyme important in iron homeostasis, was undetectable and associated with low serum iron, low serum copper, and 10-fold increase in serum ferritin. Liver biopsy confirmed increased hepatocyte iron storage (figure 2B). Aceruloplasminemia was diagnosed. Iron chelation was not administered given advanced dementia at presentation.

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AUTHOR CONTRIBUTIONS
Natalie E. Parks analyzed and interpreted material for inclusion in this case report and drafted and revised the manuscript. Robert A. Vandorpe analyzed and interpreted material for inclusion in this case report and revised the manuscript. Jeremy J. Moeller analyzed and interpreted material for inclusion in this case report and revised the manuscript.

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Photomicrograph courtesy of Dr. Erica Schollenberg (Department of Pathology, Dalhousie University, Halifax, Nova Scotia, Canada).

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REFERENCES
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