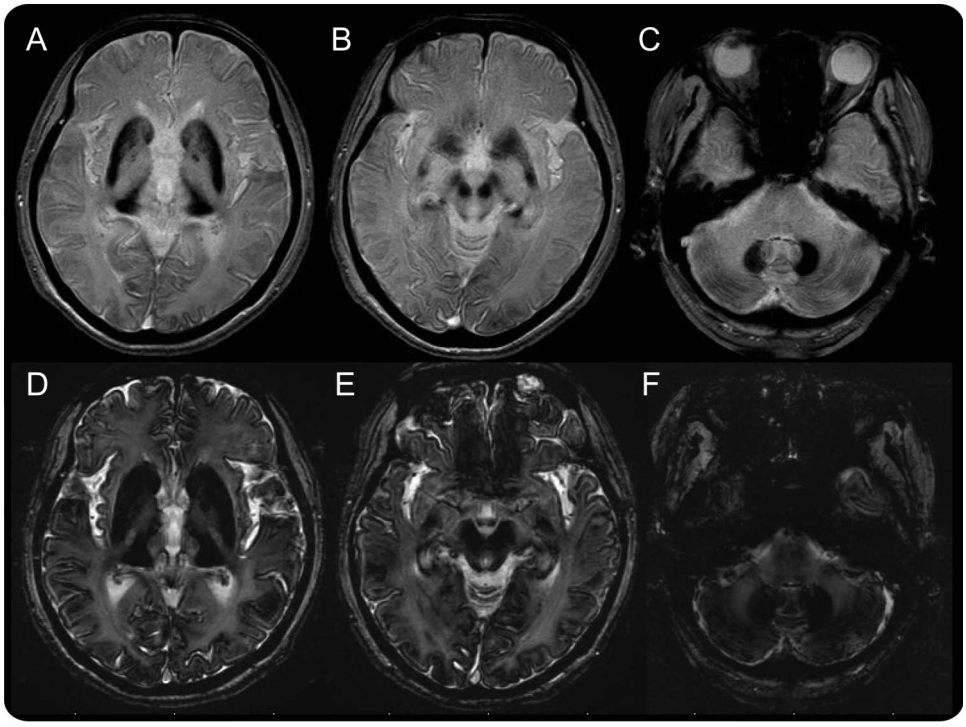
Brain and liver iron accumulation in aceruloplasminemia

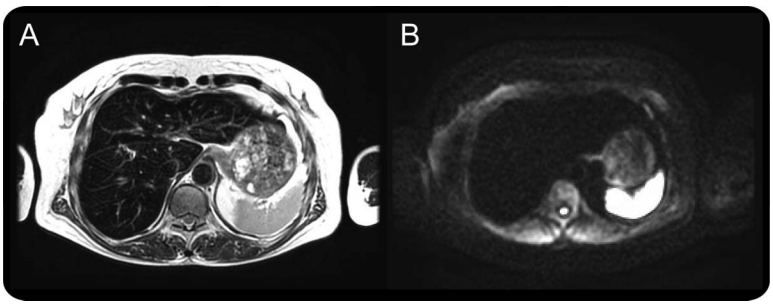
Figure 1 Brain MRI



1.5T (A–C) and 3T (D–F) T2*-weighted MRI shows hypointensity in the striatum, thalamus (A, D), red nucleus (B, E), and dentate nucleus (C, F), and on the brain surface (A–F).

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, red nucleus, and dentate nucleus, and on the brain surface, more prominently on high-field imaging (figure 1). Liver MRI demonstrated marked signal attenuation (figure 2). Serum ceruloplasmin was undetectable, and genetic testing revealed a homozygous mutation (p.W858X)

Figure 2 Liver MRI



1.5T T2-weighted (A) and diffusion-weighted (b = 800 seconds/mm^2; B) liver MRI.

in the ceruloplasmin gene, confirming the diagnosis of aceruloplasminemia.[1] This case highlights the importance of T2*-weighted MRI in diagnosing aceruloplasminemia, even in patients with minimal neurologic symptoms.

Koji Fujita, MD, Yusuke Osaki, MD, Masafumi Harada, MD, Satoshi Kono, MD, Hiroaki Miyajima, MD, Yuishin Izumi, MD, Ryuji Kaji, MD

From the Department of Clinical Neuroscience (K.F., Y.O., Y.I., R.K.) and Radiology (M.H.), Institute of Health Biosciences, The University of Tokushima Graduate School, Tokushima, Japan; and the First Department of Medicine (S.K., H.M.), Hamamatsu University School of Medicine, Hamamatsu, Japan.

Author contributions: Dr. Fujita: design/conceptualization of the study, acquisition of data, analysis/interpretation of the data, drafting/revising the manuscript. Dr. Osaki: acquisition of data, drafting/revising the manuscript. Dr. Harada: acquisition of data, analysis/interpretation of the data, drafting/revising the manuscript. Dr. Kono: acquisition of data, analysis/interpretation of the data, drafting/revising the manuscript. Dr. Miyajima: acquisition of data, analysis/interpretation of the data, drafting/revising the manuscript. Dr. Izumi: drafting/revising the manuscript. Dr. Kaji: drafting/revising the manuscript.

Study funding: No targeted funding reported.

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

Correspondence to Dr. Fujita: kof@clin.med.tokushima-u.ac.jp

1. Kono S. Aceruloplasminemia. Curr Drug Targets 2012;13:1190–1199.

Brain and liver iron accumulation in aceruloplasminemia

Koji Fujita, Yusuke Osaki, Masafumi Harada, et al.

Neurology 2013;81;2145-2146
DOI 10.1212/01.wnl.0000437304.30227.bd