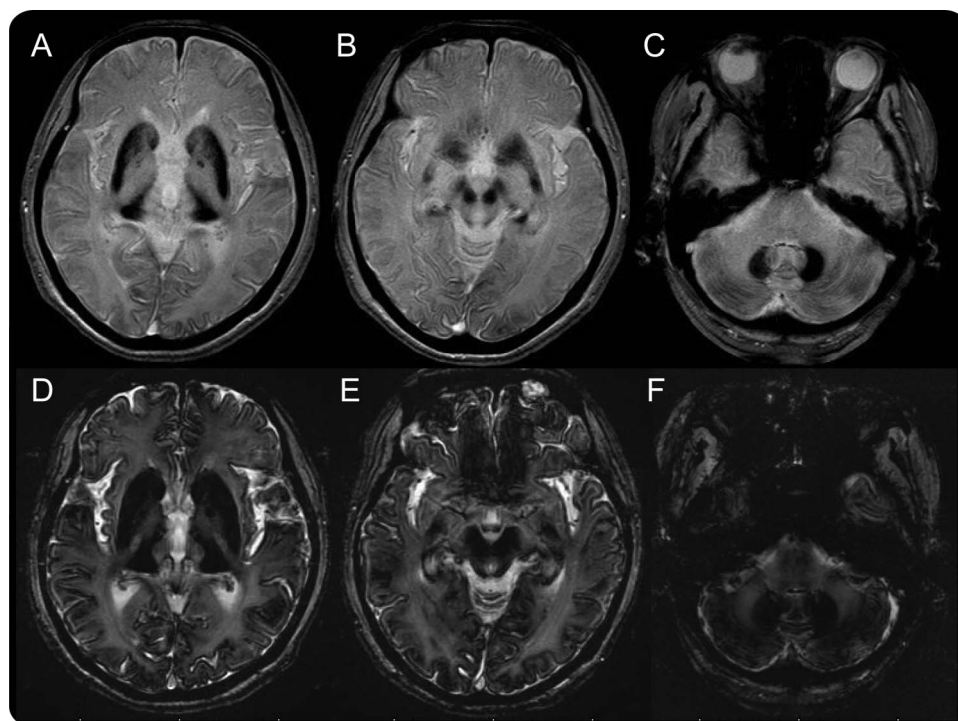


Brain and liver iron accumulation in aceruloplasminemia

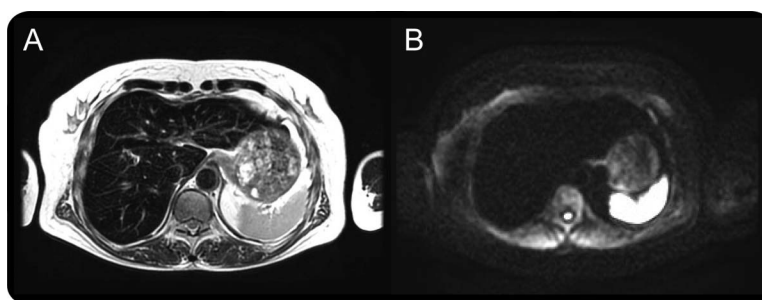
Figure 1 Brain MRI



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/mm²; 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.

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