Excessive brain iron accumulation in spinocerebellar ataxia type 17

Figure MRI in spinocerebellar ataxia type 17

(A) T1-weighted MRI shows cerebellar atrophy. (B) Susceptibility-weighted imaging shows hypointensities in (a) dentate nucleus, (b) red nucleus, and (c) globus pallidus.

A 60-year-old man had a 3-year history of cerebellar ataxia and dementia, without a family history. T1-weighted MRI showed cerebellar atrophy (figure, A). Susceptibility-weighted images (SWI) revealed hypointensities of the basal ganglia and mesencephalic and cerebellar nuclei (figure, B, a–c), suggesting neurodegeneration with brain iron accumulation.¹ Serum copper, iron, ferritin, transferrin, and ceruloplasmin levels were normal. Genetic testing revealed a CAG/CAA repeat expansion of 1 allele with 44 repeats (normal range 25–42), within the reduced penetrance range (43–48 repeats) in the TATA box binding protein (*TBP*) gene.² In patients with cerebellar atrophy with hypointensities of subcortical and cerebellar nuclei in SWI or gradient echo imaging, diagnostic considerations should include spinocerebellar ataxia 17.

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