Excessive brain iron accumulation in spinocerebellar ataxia type 17

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. 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. 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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