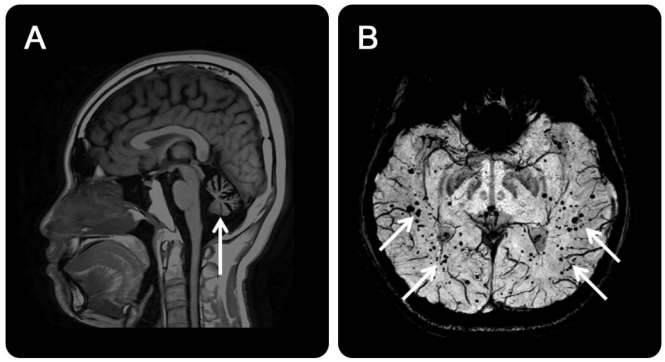
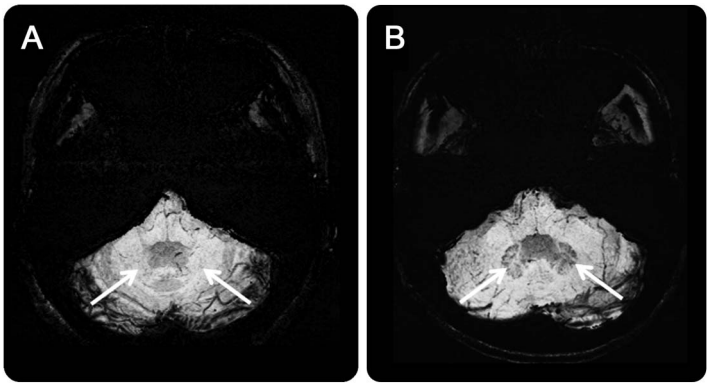
Cerebral microbleeds and iron depletion of dentate nuclei in ataxia-telangiectasia

Figure 1 Cerebellar atrophy and cerebral microbleeds in ataxia-telangiectasia



(A) T1-weighted MRI shows cerebellar atrophy (arrow). (B) Susceptibility-weighted images (SWI) show punctate signal voids scattering throughout the normal-sized cerebrum (arrows), suggesting microbleeds with hemosiderin deposits, which are related to telangiectatic vessels present in the patient with ataxia-telangiectasia.

Figure 2 Iron depletion of dentate nuclei in ataxia-telangiectasia



(A) Susceptibility-weighted images (SWI) reveal an absence of hypointensity of the iron signal in the dentate nuclei of the patient with ataxia-telangiectasia (arrows) compared to an age- and sex-matched normal control participant (B), whereas the dentate nuclei with a typical dark signal related to iron deposits are remarkably visible in SWI (arrows).

A 27-year-old man had been diagnosed with ataxia-telangiectasia at age 13 years. He had ocular telangiectasia and motor ataxia, with incoordination of head and eyes in lateral gaze. Laboratory data revealed deficiency of immunoglobulin and elevated α-fetoprotein. MRI demonstrated cerebellar atrophy and cerebral microbleeds[1] (figure 1); the dentate nuclei had deficient iron signals, which otherwise should be visible as hypointensities in susceptibility-weighted imaging (figure 2). Iron depletion in dentate nuclei is a novel finding and could be explained by the blockage of axonal iron transport in the olivocerebello-olivary loop.[2]

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