Teaching Neuro*Image*

Cerebrotendinous xanthomatosis

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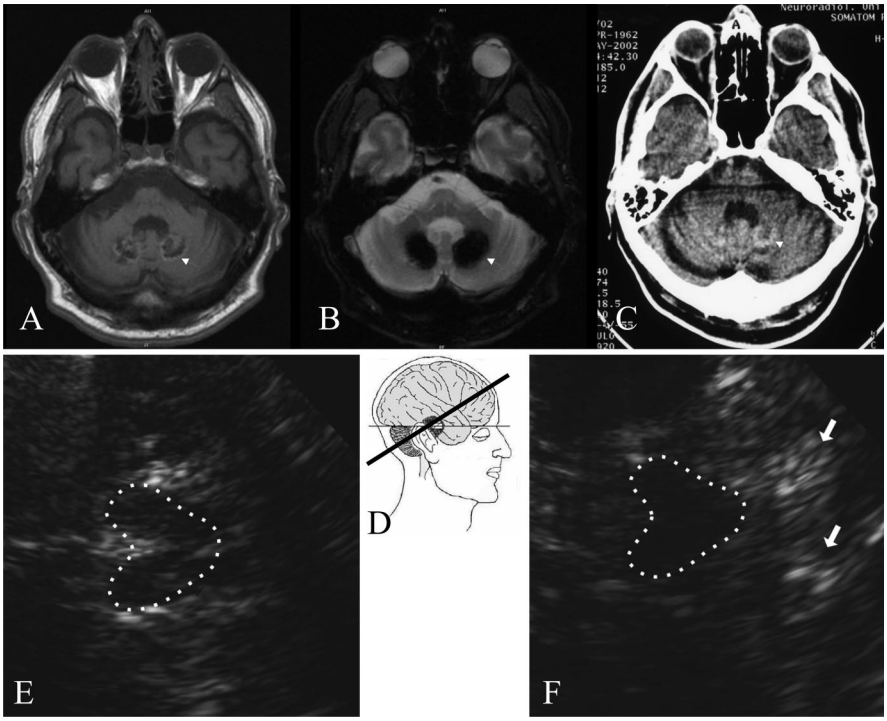

Figure. MRI revealed T1 hypointensities with hyperintense rim (A) with corresponding hyperdensity on CT (C) suggestive of a mixture of calcification and hemosiderin deposits. On T2 images (B) both regions are indistinguishable hypointense due to susceptibility effects. Transcranial sonography showed in the fourth-ventricular scanning plane (D) marked hyperechogenicity of the dentate nucleus (arrows) behind the mesencephalic brainstem (dotted lines) (F) compared to a control (E).*

A 43-year-old man developed progressive ataxia since 21 years of age followed by dementia, spasticity, epilepsy, and neuropathy. MRI, CT, and transcranial sonography revealed pronounced calcification and hemosiderin deposits of the dentate nucleus not affecting the basal ganglia (figure).[1] CSF was normal and disorders of copper and calcium metabolism were excluded. Cholestanol and abnormal bile alcohols in plasma were increased, suggestive of 27-hydroxylase (CYP27) deficiency, establishing the diagnosis of cerebrotendinous xanthomatosis. However, xanthomas were missing, as in about 30% of patients.[2] Chronic diarrhea was present since infancy and juvenile cataracts were removed at age 22. Relation of these early manifestations to neurologic disease was missed and supplementation with chenodeoxycholic acid was not started before age 42. Treatment improved diarrhea and stabilized the disease but did not alleviate neurologic symptoms, as in other cases.[2]

References

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