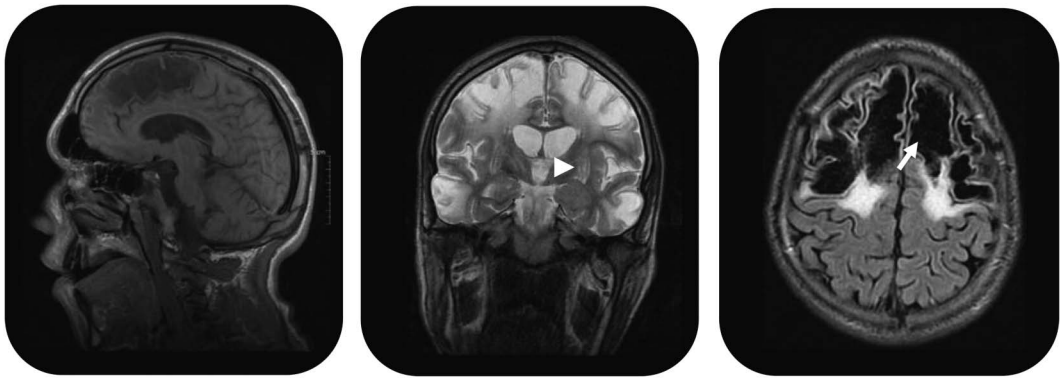
Extensive striatal, cortical, and white matter brain MRI abnormalities in Wilson disease

Figure White matter, cortical, and striatal abnormalities (brain MRI)



Diffuse bilateral white matter, cortical, and striatal (arrowhead) abnormalities, with cystic aspect (arrow) in frontal region (fluid-attenuated inversion recovery).

A 16-year-old boy presented with progressive dysarthria and gait and behavior disorders. The diagnosis of Wilson disease was made, based on Kayser-Fleischer rings, hypocupremia, hypoceruloplasminemia, and increased 24-hour urinary copper, and confirmed by molecular analysis (homozygous state, p.[Glu1382*]; [Glu1382*]). Brain MRI demonstrated diffuse bilateral cortical and subcortical abnormalities (figure). Chelator therapy (D-penicillamine) produced partial improvement, although the patient developed epileptic seizures, presumably due to the cortical involvement. Wilson disease with extensive cortical-subcortical lesions is rare,[1,2] but should be considered as a possible etiology of diffuse leukoencephalopathy with cystic evolution.

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