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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