

Frontal lobes white matter abnormalities mimicking cystic leukodystrophy in Wilson's disease

Anormalidades na substância branca frontal mimetizando leucodistrofia cística na doença de Wilson

Flavio Moura Rezende Filho¹, Eva Rocha¹, Livia Almeida Dutra¹, José Luiz Pedroso¹, Orlando G. P. Barsottini¹

An 18-year-old male presented with a three-year history of slurred speech, gait impairment, seizures and progressive neurological deterioration. Brain MRI depicted bilateral hyperintense T2-signal in the basal ganglia and white matter abnormalities with a cystic appearance in the frontal lobes. Ophthalmological evaluation disclosed Kayser-Fleisher rings (Figure). Ceruloplasmin was low and urinary copper was increased, and Wilson's disease was diagnosed.

Wilson's disease is an autosomal recessive disorder. Typical neurological features include akinetic-rigid syndrome, tremor, ataxia and dystonia^{1,2}. Neuroimaging usually shows signal abnormalities in the globus pallidus, putamen, caudate nucleus, thalamus and cerebral peduncles^{1,2}. Frontal white matter involvement mimicking leukodystrophy with cystic evolution is a rare presentation^{3,4}.

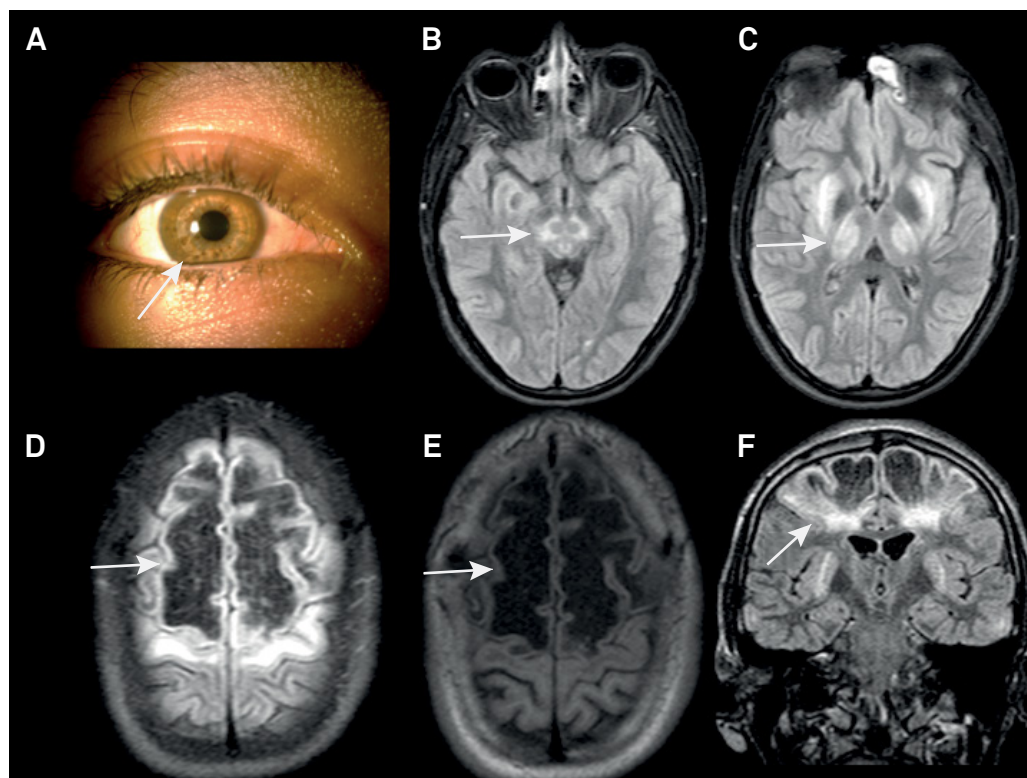


Figure. A: Kayser-Fleischer ring (arrow). B and C: Axial FLAIR-weighted brain MRI shows a giant Panda sign and hyperintense signals in the basal ganglia. D, E and F: marked bilateral frontal leukoencephalopathy with cystic lesions.

¹Universidade Federal de São Paulo, Departamento de Neurologia e Neurocirurgia, Divisão de Neurologia Geral, São Paulo SP, Brasil.

Correspondence: Orlando Barsottini; Departamento de Neurologia e Neurocirurgia, UNIFESP; Rua Napoleão de Barros, 715; 04023-900 São Paulo SP, Brasil; E-mail: orlandobarsottini@gmail.com

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