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, Lívia 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}.

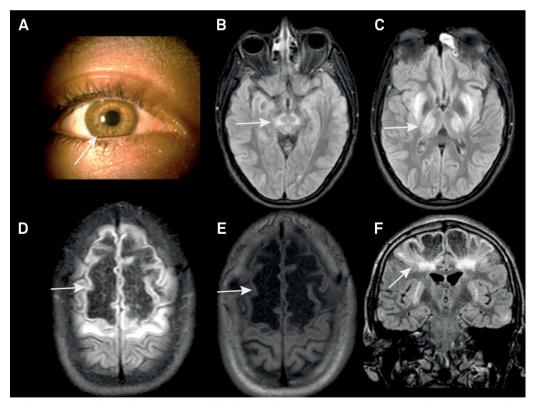


Figure. A. Kayser-Fleischer ring (arrow). B and C: Axial FLAIR-weighted brain MRI shows a giant Panda sign and hyperintense signs in the basal ganglia. D, E and F: marked bilateral frontal leukoencephalopathy with cystic lesions.

Conflict of interest: There is no conflict of interest to declare.

Received 05 July 2016; Accepted 08 November 2016.



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

References

- Bandmann O, Weiss KH, Kaler SG. Wilson's disease and other neurological copper disorders. Lancet Neurol. 2015;14(1):103-13. https://doi.org/10.1016/S1474-4422(14)70190-5
- Machado A, Chien HF, Deguti MM, Cançado E, Azevedo RS, Scaff M et al. Neurological manifestations in Wilson's disease: report of 119 cases. Mov Disord. 2006;21(12):2192-6. https://doi.org/10.1002/mds.21170
- Hedera P, Brewer GJ, Fink JK.. White matter changes in Wilson disease. Arch Neurol. 2002;59(5):866-7.
- 4. Trocello JM, Woimant F, El Balkhi S, Guichard JP, Poupon J, Chappuis P, Feillet F. Extensive striatal, cortical, and white matter brain MRI abnormalities in Wilson disease. Neurology. 2013;81(17):1557. https://doi.org/10.1212/WNL.0b013e3182a95883