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

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

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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].

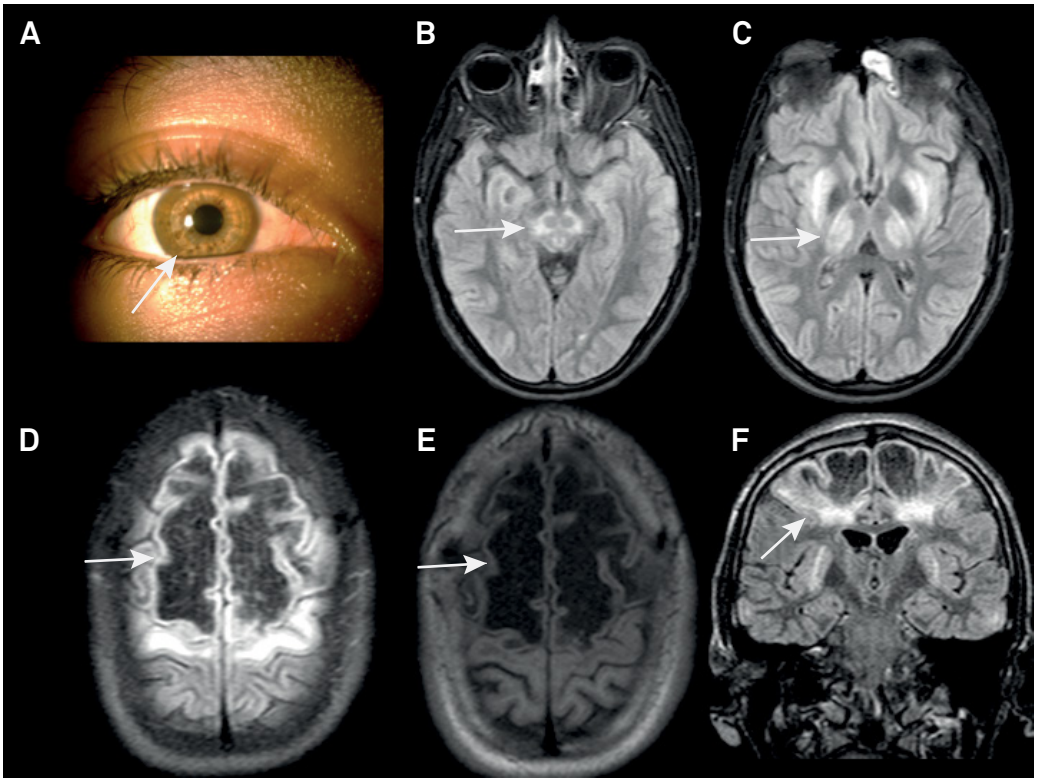

Figure. A. Kayser-Fleischer ring (arrow). B and C: Axial FLAIR-weighted brain MRI shows a giant Panda sign and hyperintense signs in the basal ganglia. D, E and F: marked bilateral frontal leukoencephalopathy with cystic lesions.

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