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

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. Neuroimaging usually shows signal abnormalities in the globus pallidus, putamen, caudate nucleus, thalamus and cerebral peduncles. Frontal white matter involvement mimicking leukodystrophy with cystic evolution is a rare presentation.

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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References


