A case of Central pontine myelinolysis-like lesion associated with Wilson disease: an atypical onset of hepatic or neurological Wilson disease?

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Background

Wilson disease (WD) is an inborn error of copper metabolism caused by a mutation in the copper transporting gene ATP7B on chromosome 13q14.3. This condition leads to copper deposition in liver, brain, kidneys, eyes, bones and blood tissues and may be responsible of hepatic dysfunction (40%) neurological (40%) and psychiatric (16%) disorders, associated with other rare manifestations (3%).

Objective

To report the case of a woman with central pontine myelinolysis (CPM)-like lesion associated with WD.

Case Report

A female patient presented with rapid progression to cirrhosis at the age of 67. The ATP7B gene was analyzed and a homozygous state of the -441/-427 mutation was found. Kayser-Fleischer rings were not seen on slit lamp examination. WD diagnosis was made and treatment with penicillamine was initiated, although currently taking trientine (900 mg/day). After 4 years, walking difficulties with postural instability and frequent risk of falls were reported and a brain MRI showed only the presence of "a triangular hyperintense signal on T2-weighted images involving the peduncle of the pons until the midbrain, similar to pontine myelinolysis" (Figure). Neurophysiological studies reported mild difficulties in stocking new informations, poor letter verbal fluencies and moderate impairment in picture naming tasks for nouns and actions. Laboratory findings revealed reduced blood copper level, increase of urinary copper excretion, hypoceruloplasminemia, slight increase of blood creatinine and hyperbilirubinemia. The other laboratory data were in normal limits. Neurological examination showed the presence of signs of pyramidal tract lesion with increased deep tendon reflexes and pathological reflexes such as Hoffman's sign.

Discussion and Conclusions

Findings of CPM-like at MRI in WD are uncommon and the univocal interpretation of the underlying physiopathological mechanism is difficult. In fact, CPM is usually detected in chronic alcoholism, malnutrition, and rapid correction of sodium in patients with severe hyponatremia. Currently, three patterns of CPM-like changes in WD have been described. Our patient showed the classic pattern; however, she did not exhibit any of the neurological symptoms which are the most frequently reported such as bulbar symptoms and drooling. Our case documents as CPM-like lesion may be present also in WD with hepatic injury as the first manifestation, but it could be also predictive of a neurological involvement.

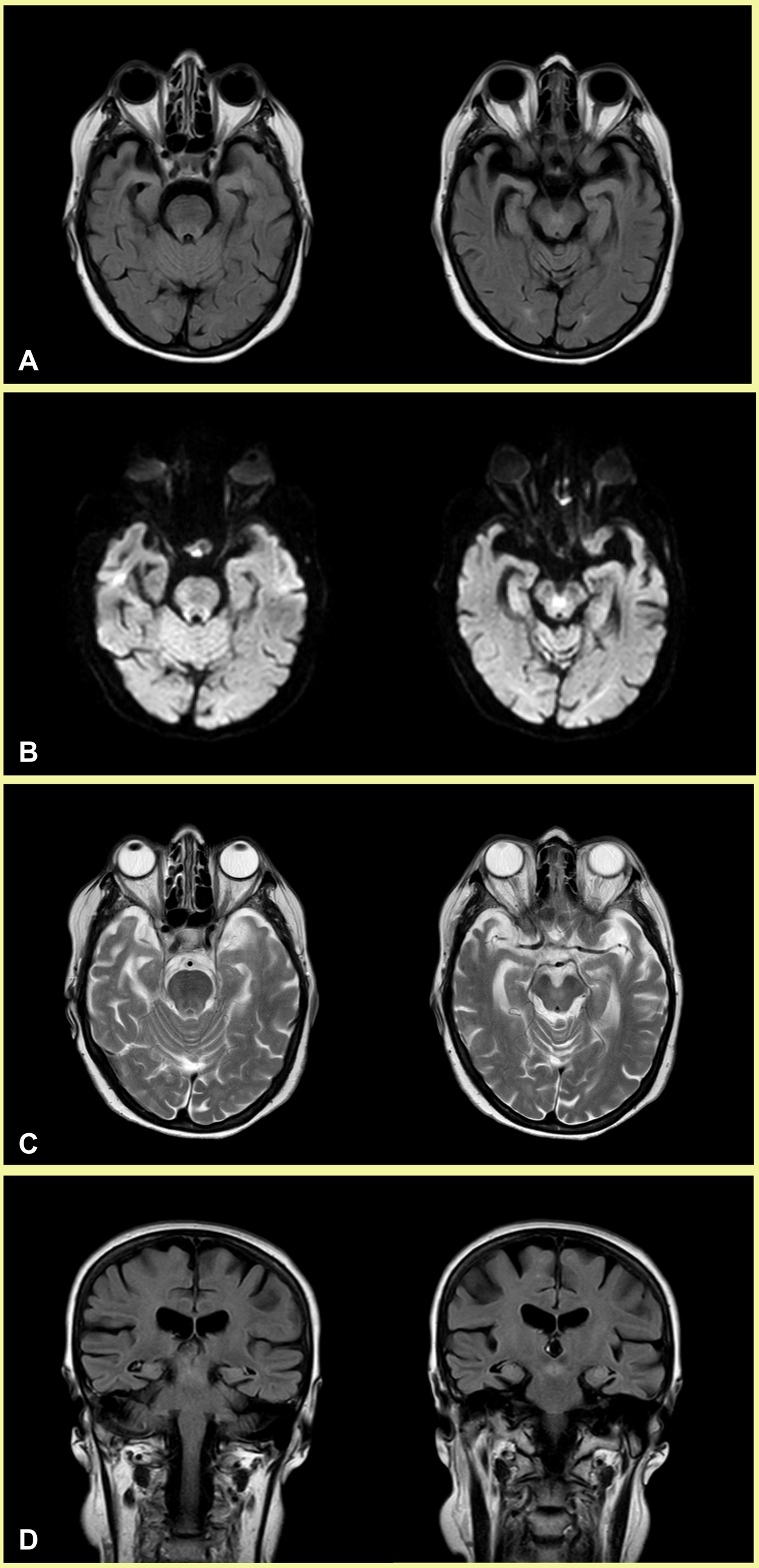

Figure. (A) FLAIR axial sequence; (B) DWI axial sequence; (C) T2 axial sequence; (D) FLAIR coronal sequence.

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