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Wilson's disease: Atypical imaging features

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CASE STUDY

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ABSTRACT

Wilson's disease is a genetic movement disorder with characteristic clinical and imaging features. We report a 17-year-old boy who presented with sialorrhea, hypophonic speech, paraparesis with repeated falls and recurrent seizures along with cognitive decline. He had bilateral Kayser Fleischer rings. Other than the typical features of Wilson's disease in cranial MRI, there were extensive white matter signal abnormalities (T2 and FLAIR hyperintensities) and gyriform contrast enhancement which are rare imaging features in Wilson's disease. A high index of suspicion is required to diagnose Wilson's disease when atypical imaging features are present.

Key Words
Wilson's disease, hepato-lenticular degeneration, copper storage disorders, neuroimaging, MRI

Implications for Practice:

1. What is known about this subject?
The radiological picture of Wilson's disease is unique with MRI typically showing lesions in basal ganglia, thalamus and brainstem as well in cortex and subcortical white matter. Unusual imaging features can be present in Wilson's disease.

2. What new information is offered in this case study?
We report a case of Wilson's disease who had unique radiological features like extensive white matter abnormalities and gyral contrast enhancement in cranial MRI

3. What are the implications for research, policy, or practice?
This case study highlights that atypical imaging features can occur in Wilson's disease and a high index of suspicion is necessary to detect this treatable disease.

Background

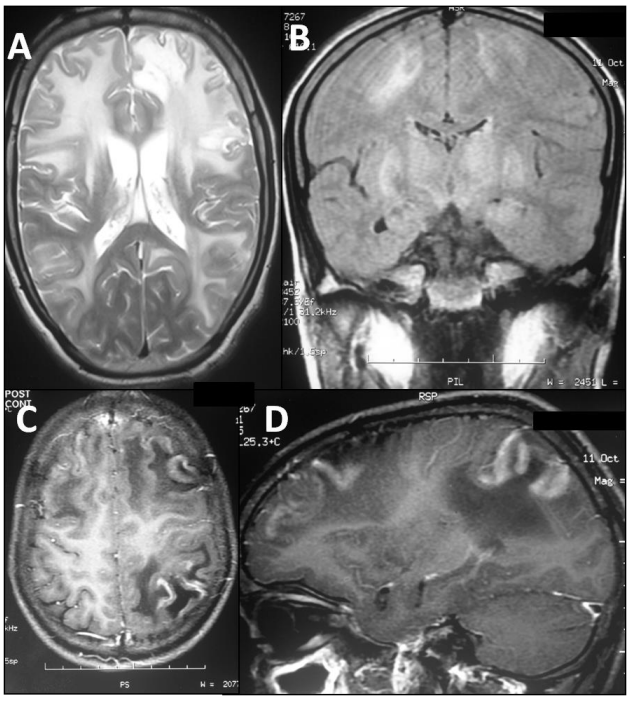
Conventionally, diagnosis of Wilson's disease (WD) rests on demonstration of characteristic biochemical abnormalities (low serum ceruloplasmin, high 24hr urinary copper, increased free serum copper) and Kayser Fleischer rings.[1] Magnetic resonance imaging (MRI) of brain is a useful alternative tool in diagnosis of WD, especially so as KF ring may be absent and serum ceruloplasmin and other copper parameters may be spuriously normal. The MRI picture of WD is unique with characteristic lesions in basal ganglia, thalami and brainstem.[1] With increasing use of MRI in WD during the last two decades, it is not uncommon to encounter rare radiological manifestations of WD. Here we report a patient with rare clinical presentation and radiological findings.

Case details

A 17-year-old previously healthy boy presented with sialorrhea and hypophonic speech since two years, paraparesis with repeated falls since one and a half year years and recurrent seizures along with cognitive decline since one year. He was bed bound with urinary and faecal incontinence since last six months. The parents did not report any behavioural disturbance during the course of illness. Family history was non-contributory. He was diagnosed as a case of leukodystrophy at a primary health centre and managed conservatively. On examination he was

conscious though bedbound and without any meaningful communication. There was severe spasticity in all four limbs with brisk deep tendon reflexes and bilateral extensor plantar responses. He had bilateral Kayser Fleischer rings. He was moving all four limbs. There were no fasciculation's or flexor spasms. Pupils were equal and reacting to light. Cerebellar and sensory examinations could not be performed. MRI done at another centre about 4 months earlier showed lesions in bilateral putamina, bilateral thalami and brainstem, which were consistent with clinical diagnosis of WD. However, the most glaring feature on MRI was extensive white matter signal abnormalities (T2 and FLAIR hyperintensities) and gyriform contrast enhancement (Figure 1 A-D). There was no history of hypoxia or recent seizures. His biochemical abnormalities (serum ceruloplasmin - 6.2µg/dL, 24hr urine copper - 130µg per day) were consistent with diagnosis of WD. The urine and serum tandem mass spectroscopy were normal. He was started on Zinc and d-Penicillamine and at last follow up (1 year after presentation), he has shown mild improvement and though still bedbound, he is currently able to tell his daily body needs.

Figure 1A: T2W Axial MRI Brain images showing severe T2 hyperintensities, cavitation and gliosis with signal changes in outer rim of putamina, thalamus and tegmentum of midbrain
Figure 1B: MRI Brain coronal FLAIR images showing hyperintensities in bilateral basal ganglia and thalami
Figure 1C and 1D: Gad-enhanced MRI brain showing gyriform contrast enhancement



Discussion

Magnetic resonance imaging (MRI) is a useful tool in diagnosis of WD with a sensitivity approaching 100 per cent in symptomatic patients. The putamina are involved most commonly (86 per cent) with a distinctive lateral rim of high signal intensity on T2 weighted images. Other commonly involved regions include ventrolateral thalami (54 per cent), caudate (45 per cent), globus pallidus (41 per cent), midbrain (82 per cent), pons (77 per cent), cortical white matter and cerebellum.[2]

Though, there are a few case reports of extensive white matter abnormalities in WD, the exact pathogenesis of white matter lesions of WD is still far from clear.[2-6] It is still unknown as to why only a few patients develop massive white matter lesions. In few reported cases of WD with extensive white matter abnormalities, concentration of copper was higher in frontal lobe than in the putamen indicating that neurotoxicity of unbound copper storage on white matter is one of the main factors.[4] However reason for this deposition of copper is still unknown.

The other interesting MRI finding in our patient was gyral contrast enhancement. There is only one similar case report in literature where Gd enhanced MRI of brain revealed enhancement of cerebral cortex and cortico-medullary junction in WD.[5] It was hypothesised in that report that excessive deposition of copper in vascular wall leads to vascular leakage and contrast enhancement. However in the absence of biopsy proof in our case this hypothesis is difficult to confirm or refute.

The possibility of a co-existent leucodystrophy or inborn error of metabolism (IEM) cannot be ruled out without brain biopsy. But tandem mass spectroscopy was normal in our patient which can rule out significant number of disorders of IEM. The clinical features and course were nevertheless not suggestive of a leucodystrophy.

Conclusion

Our patient is unique as it showed two rare MRI observations (extensive white matter abnormalities and gyral contrast enhancement), in addition to features characteristic of WD. Recognition of these changes can help in early management of this genetic disorder. Our report highlights the importance of a high index of clinical suspicion for WD, careful observation of MRI findings and a proper clinic-radiological correlation.

The exact cause and why only some patients show this unique set of findings remains unanswered and future studies are required to answer these questions.

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PEER REVIEW

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CONFLICTS OF INTEREST

The authors declare that they have no competing interests.

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PATIENT CONSENT

The authors, *Vishnu VY, Modi M, Goyal MK, Vyas V, Lal V* declare that:

1. They have obtained written, informed consent for the publication of the details relating to the patient(s) in this report.
2. All possible steps have been taken to safeguard the identity of the patient(s).
3. This submission is compliant with the requirements of local research ethics committees.