BEYOND THE BASAL GANGLIA: RARE CORTICAL INVOLVEMENT IN A CASE OF WILSON'S DISEASE

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BACKGROUND

- Wilson's disease is an AR disorder caused by defective copper metabolism.
- Neurological manifestations usually appear later and involve dystonia, dysarthria, and parkinsonism.
- *Typical MRI* findings include basal ganglia and brainstem hyperintensities, but cortical lesions are rarely reported.
- Recognizing atypical imaging features is vital for early diagnosis and management to prevent irreversible neurological damage

AIMS

- The aim of this report is to highlight the *occurrence of rare cortical and subcortical involvement* in Wilson's disease.
- It seeks to demonstrate how advanced neuroimaging can reveal atypical patterns extending beyond the basal ganglia.
- This case also aims to expand current *understanding of the neuropathological* mechanisms involved in WD.

MATERIALS AND METHODS

A focused literature review was conducted using PubMed to identify reports describing cortical imaging findings in Wilson's disease. Search terms included "Wilson's disease," "MRI," "cortical involvement," and "white matter lesions."

Relevant studies were screened for case descriptions and series detailing cortical or subcortical abnormalities.

Additionally, we present a case of a 16-year-old boy with Wilson's disease showing rare cortical and subcortical MRI abnormalities.

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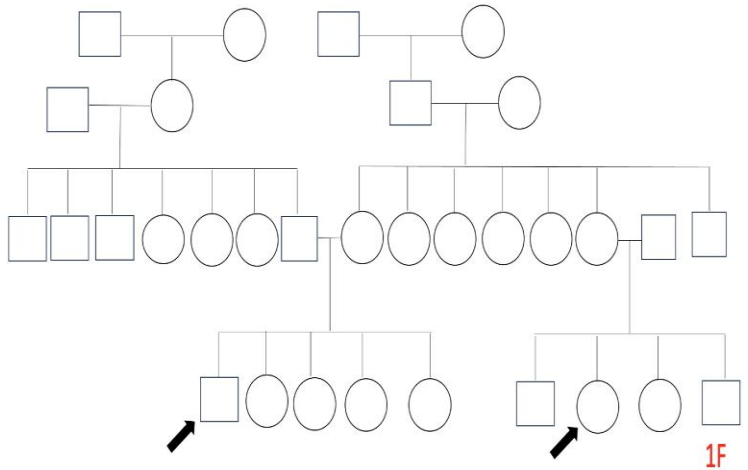

Reference	Year	Study Type / Population	MRI Findings	Key Observations
King et al., AJR 1996	1996	Case series	Cortical signal abnormalities in 59%	Early report highlighting frequent cortical signal changes
Grover et al., Indian J Radiol Imaging 2006	2006	Case report (14-year-old boy)	T2 hyperintensities in cortical gray & subcortical white matter (frontal, parietal, temporal)	Demonstrated extensive involvement; suggested cytotoxic edema or copper-induced inflammation
Sinha et al., Neuroradiology 2006	2006	100 patients	Cortical signal abnormalities in 9%	One of the largest Indian cohorts; cortical lesions uncommon
Ranjan et al., Clin Neurol Neurosurg 2015	2015	68 patients	Cortical signal abnormalities in 26.5%	Higher cortical involvement; correlated with worse neurological outcomes
Present case, 2024	2024	Single case (16-year-old boy)	Symmetrical T2 & FLAIR hyperintensities in basal ganglia + cortical/subcortical frontoparietal changes; DWI restriction	Rare cortical involvement and gyral diffusion restriction; likely copper-induced cytotoxic edema

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DISCUSSION

Neuroimaging Patterns: Typical MRI findings include symmetric hyperintensities in deep gray matter (putamen, caudate, thalamus), brainstem, and sometimes white matter. Cortical involvement is rare, reported in 9–26.5% of cases.

Cortical Diffusion Restriction: Rare gyral DWI hyperintensity with low ADC reflects cytotoxic edema due to copper-induced neuronal injury, inflammation, and cell death, rather than ischemia—highlighting a unique neuroimaging manifestation.

Clinical Significance: WD can involve extensive gray and white matter beyond basal ganglia. Recognizing cortical and subcortical lesions broadens diagnostic awareness and underscores the value of advanced MRI for assessing neurological involvement.

CONCLUSION

- This case underscores that Wilson's disease may present with extensive gray and white matter lesions beyond the basal ganglia and brainstem.
- Cortical diffusion restriction is an unusual but significant manifestation of WD.
- Awareness of such imaging patterns can aid in timely diagnosis and guide effective treatment strategies.
- The report broadens the neuroimaging spectrum and enhances understanding of WD's neuropathological diversity.

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