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Case Report

Neuroradiological Manifestations of Pediatric Wilson Disease: A Case Report

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Abstract

Wilson disease is a rare autosomal recessive disorder of copper metabolism resulting in excessive copper accumulation, primarily in the liver and brain. Neurological involvement is commonly associated with characteristic magnetic resonance imaging (MRI) findings, particularly affecting the basal ganglia. We report the case of an 11-year-old boy with Wilson disease who underwent brain MRI for neurological evaluation. Imaging revealed bilateral and symmetrical signal abnormalities involving the lentiform and caudate nuclei, characterized by hyperintensity on T2-weighted and fluid-attenuated inversion recovery (FLAIR) sequences, with corresponding hypointensity on T2*-weighted images. These findings were consistent with basal ganglia involvement secondary to Wilson disease. This case highlights the characteristic neuroradiological manifestations of pediatric Wilson disease and underscores the pivotal role of brain MRI in detecting central nervous system involvement, supporting diagnosis, and facilitating appropriate clinical management.

Keywords: *Wilson Disease; Magnetic Resonance Imaging; Brain MRI; Basal Ganglia; Lentiform Nucleus; Caudate Nucleus*

Introduction

Wilson disease is a rare autosomal recessive disorder of copper metabolism caused by mutations in the *ATP7B* gene, resulting in impaired biliary copper excretion and progressive copper accumulation in various organs, particularly the liver and brain. The disease most commonly presents during childhood or adolescence with hepatic, neurological, or psychiatric manifestations. Early diagnosis and prompt treatment are essential to prevent irreversible neurological damage and improve long-term outcomes [1,2].

Neurological involvement is frequently associated with characteristic magnetic resonance imaging (MRI) findings, predominantly affecting the basal ganglia. Typical abnormalities include bilateral and symmetrical signal changes involving the putamen, globus pallidus, and caudate nuclei, reflecting copper deposition and secondary neurodegeneration. Brain MRI therefore plays a crucial role in assessing central nervous system involvement and supporting the diagnosis of Wilson disease [3,4].

Herein, we report the case of an 11-year-old boy with Wilson disease presenting with characteristic neuroradiological manifestations on brain MRI, emphasizing the importance of MRI in the evaluation of children with neurological involvement.

Case Report

An 11-year-old boy with a known diagnosis of Wilson disease presented with progressive neurological symptoms, including tremor, dysarthria, dystonia, and gait disturbance. Brain magnetic resonance imaging (MRI) was performed to assess central nervous system involvement. MRI demonstrated bilateral and symmetrical hyperintense signal abnormalities involving the lentiform and caudate nuclei on T2-weighted and fluid-attenuated inversion recovery (FLAIR) images, with corresponding hypointense signal changes on T2*-weighted images. These findings were consistent with the characteristic neuroradiological manifestations of Wilson disease.

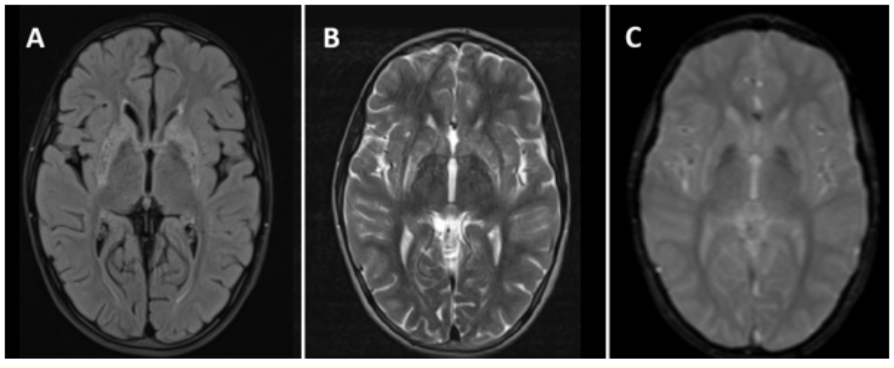

Figure 1: *Characteristic brain MRI findings in pediatric Wilson disease.*

(A) Axial fluid-attenuated inversion recovery (FLAIR) image demonstrating bilateral and symmetrical hyperintense signal abnormalities involving the lentiform and caudate nuclei. (B) Axial T2-weighted image confirms the corresponding bilateral and symmetrical hyperintense signal abnormalities. (C) Axial T2-weighted image demonstrates corresponding hypointense signal changes in the lentiform and caudate nuclei.*

Discussion

Wilson disease is a rare autosomal recessive disorder caused by mutations in the *ATP7B* gene, resulting in impaired biliary copper excretion and subsequent copper accumulation in multiple organs, particularly the liver and the central nervous system. Although hepatic manifestations often predominate during childhood, neurological involvement generally appears later and is associated with copper deposition in the basal ganglia, brainstem, and cerebellum. Early recognition of neurological involvement is essential because timely initiation of chelation therapy may prevent irreversible neurological damage and improve clinical outcomes [1,2].

Clinical manifestations of Wilson disease are highly variable and depend on the organs predominantly affected by copper accumulation. In children, hepatic manifestations are often the initial presentation, whereas neurological symptoms generally develop later during the course of the disease. The most common neurological manifestations include tremor, dysarthria, dystonia, parkinsonism, gait disturbances, and psychiatric or cognitive abnormalities. The presence of Kayser-Fleischer rings further supports the diagnosis in patients with neurological involvement [2,3].

Magnetic resonance imaging (MRI) is the imaging modality of choice for evaluating neurological involvement in Wilson disease. The most common MRI findings include bilateral and symmetrical signal abnormalities affecting the putamen, globus pallidus, caudate nuclei, and thalami. These lesions typically appear hyperintense on T2-weighted and FLAIR sequences, reflecting edema, gliosis, demyelination, and tissue degeneration induced by copper deposition. Signal alterations on susceptibility-weighted or T2*-weighted images may also be observed owing to paramagnetic mineral deposition and chronic tissue injury [3,4].

In the present case, brain MRI demonstrated bilateral and symmetrical hyperintense signal abnormalities involving the lentiform and caudate nuclei on T2-weighted and FLAIR images, with corresponding hypointense signal changes on T2*-weighted imaging. These findings are characteristic of basal ganglia involvement in Wilson disease and correlate well with the neuroimaging patterns described in the literature. The absence of significant mass effect or focal enhancement further supports a metabolic rather than a structural or inflammatory etiology [3,4].

Although the MRI findings in Wilson disease are not entirely specific, their characteristic distribution, particularly in the basal ganglia, combined with the appropriate clinical and laboratory context, strongly supports the diagnosis. MRI also plays an important role in evaluating disease severity, monitoring therapeutic response, and detecting disease progression during follow-up [2,5].

This case highlights the characteristic neuroradiological manifestations of pediatric Wilson disease and emphasizes the important role of brain MRI in identifying central nervous system involvement. Familiarity with these imaging features enables radiologists to suggest the diagnosis promptly and contributes to early multidisciplinary management, thereby improving patient outcomes.

Conclusion

Brain MRI plays a pivotal role in the evaluation of neurological involvement in Wilson disease by demonstrating characteristic bilateral and symmetrical basal ganglia abnormalities. Recognition of these neuroradiological features is essential for supporting the diagnosis, assessing the extent of central nervous system involvement, and guiding appropriate clinical management. Early identification of these imaging findings, particularly in children, may facilitate timely therapeutic intervention and help prevent irreversible neurological damage. This case underscores the importance of MRI as a valuable diagnostic tool in pediatric Wilson disease and highlights the need for increased awareness of its characteristic imaging manifestations among radiologists and clinicians.

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Conflict of Interest

The authors declare no conflict of interest.

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