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Case Report

Atypical Presentation of Parkinsonism in a Pediatric Patient with Wilson's Disease: A Case Report

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ABSTRACT

Wilson's Disease (WD) is an autosomal recessive disorder involving mutations of the ATP7B gene on chromosome 13, characterized by impaired hepatic copper transport, which results in copper accumulation particularly in the liver and brain. Neurological manifestations such as Parkinsonism can occur in advanced stages of the disease but is rare in pediatric patients. A 10-year-old female presented with complaints of inappropriate laughter and slowness in carrying out daily activities. She also had a history of tremors, rigidity, bradykinesia, and dystonic portray of the neck and upper limbs for 2 months. Neurological examination revealed cogwheel rigidity, resting tremors, facial hypomimia, and dystonic portraying of both upper limbs. The patient had no family history of movement disorders. Laboratory investigations, including low serum ceruloplasmin levels and high 24 hr urinary copper excretion, confirmed the diagnosis of Wilson's disease. Ocular examination revealed the presence of Kayser-Fleischer (KF) rings, a common manifestation of Wilson's disease. Brain Magnetic Resonance Imaging (MRI) demonstrated characteristic findings of basal ganglia hyper-intensities. The patient was treated with Tab Trihexyphenidyl (THP), Vitamin C and Zinc supplementation for 5 days, following which she was discharged with a comprehensive follow-up plan. This case emphasizes the atypical presentation of Parkinsonism in a pediatric patient with Wilson's disease. It underscores the importance of considering Wilson's disease as a potential etiology in young patients presenting with movement disorders, even in the absence of family history. Early diagnosis and treatment are necessary to prevent irreversible neurological damage. Long-term monitoring and follow-up are necessary to ensure treatment efficacy and prevent disease complications.

Keywords: Wilson's disease, Hepatolenticular Degeneration, Parkinsonian Disorders, KF rings.

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INTRODUCTION

Wilson's Disease (WD), also known as hepatolenticular degeneration, is an autosomal recessive disorder of copper metabolism characterized by impaired biliary excretion of copper, leading to its accumulation in various organs, including liver, cornea and brain.[1-3] Copper is an essential nutrient involved in various physiological processes, including mitochondrial respiration, melanin biosynthesis, dopamine metabolism, iron hemostasis, antioxidant defense, connective tissue formation and peptide amidation.[1,4]

First described by Samuel Kinnier Wilson over a century ago, WD arises due to mutations in the copper transporting gene ATP7B, located on human chromosome 13.[1,2,4] These mutations lead to impaired copper transport and its subsequent accumulation.[1] The disease presents with hallmark features of liver disease, neurologic symptoms and presence of Kayser-Fleischer corneal rings, which result from excessive copper deposition.[2] Owing to its diverse clinical manifestations, WD has been referred to as "the great masquerader."[2] While hepatic impairment and cognitive dysfunction are well-known features, Parkinsonism is an uncommon presentation, especially in pediatric patients. We present a case of a 10-year-old female patient with atypical Parkinsonism as the initial manifestation of Wilson's disease.

CASE DESCRIPTION

We present a case of a 10-year-old female patient who sought medical attention with complaints of inappropriate laughter and slowness in daily activities. The parents reported a progressive onset of symptoms over past two months, including worsening tremors, rigidity, bradykinesia and dystonic movements involving neck and upper limbs. There was no known family history of movement disorders or similar symptoms.

Physical examination revealed cogwheel rigidity, resting tremors, facial hypomimia and dystonic movements affecting both upper

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Table 1: Treatment chart.

Brand name	Generic name	Dose	Frequency	D1	D2	D3	D4	D5
Tab Pacitane	Trihexyphenidyl	2mg	1-0-0	+	+	+	+	+
Cap Becosules	Vit B1+Vit B2+Vit B3+Vit B6+ Vit B7+Vit B9+Vit B12+Vit B5+Vitamin C.	10 mg+10 mg+100 mg+3 mg+100 mcg+ 1.5 mg+15 mcg+50 mg+ 150 mg.	1-0-1	+	+	+	+	+
Tab Combifiz D3	Ascorbic acid+Lycopene+Vitamin D3+Zinc sulphate.	400 mg+ 1 mcg+ 25 mcg+ 10 mg.	0-0-1		+	+	+	+

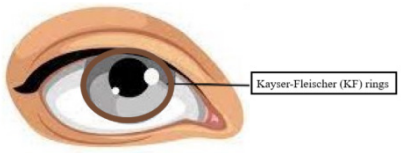

Figure 1: Kayser-Fleischer (KF) rings.

limbs. These clinical features raised concerns for a possible movement disorder secondary to WD.[5]

Laboratory investigations carried out revealed a low level of serum ceruloplasmin, a copper-binding protein essential for copper transport in the body. Furthermore, analysis of 24 hr urinary copper excretion revealed a significantly elevated level, consistent with the diagnosis of WD.[2,4] Ocular examination using slit-lamp confirmed the presence of Kayser-Fleischer (KF) rings, which are golden-brown pigmented rings around the cornea resulting from copper deposition (Figure 1). The presence of KF rings further supported the diagnosis of WD.[2,4] Brain Magnetic Resonance Imaging (MRI) was conducted to assess the extent of neurological involvement. The MRI revealed characteristic findings of hyper-intensities in the basal ganglia on T2-weighted images, a hallmark feature of WD affecting the central nervous system.[2,4]

Prompt management was initiated to alleviate the patient's symptoms and prevent further progression of disease. The treatment regimen included Tab Trihexyphenidyl (THP), Vitamin C and Zinc supplementation for 5 days. Detailed treatment is shown in Table 1. During hospitalization, a gradual improvement in tremors, rigidity and bradykinesia was observed. Following the improvement in symptoms, the patient was discharged with a comprehensive follow-up plan.

DISCUSSION

The presented case highlights the atypical manifestation of Parkinsonism secondary to WD in a 10-year old female patient. WD is an autosomal recessive disorder caused by mutations in the ATP7B gene, leading to impaired copper transport and subsequent copper accumulation in various organs, particularly the liver, cornea and brain.[1,3] While WD is a rare genetic disorder, early recognition and appropriate management are crucial to prevent irreversible neurological damage.

The patient in this case exhibited a combination of neurological symptoms, including inappropriate laughter, slowness in daily activities, tremors, rigidity, bradykinesia and dystonic movements affecting the neck and upper limbs. Such atypical presentation of Parkinsonism in a pediatric patient is relatively rare, underscoring the need for heightened awareness among healthcare professionals to consider WD as a potential etiology in young patients presenting with movement disorders.

Timely and accurate diagnosis is crucial in WD, as it allows for the initiation of appropriate treatment to prevent further copper accumulation and subsequent organ damage. The diagnosis of WD was confirmed through laboratory investigations, including low serum ceruloplasmin levels and elevated 24 hr urinary copper excretion, consistent with impaired copper metabolism. Ocular examination with a slit lamp revealed Kayser-Fleischer rings, further supporting the diagnosis. Brain MRI showed characteristic hyper-intensities in the basal ganglia, which are commonly observed in patients with neurological involvement of WD. In this case, the patient was promptly started on Tab Trihexyphenidyl (THP), Vitamin C and Zinc supplementation, which led to a gradual improvement in her symptoms during hospitalization.

CONCLUSION

This case report emphasizes the atypical presentation of Parkinsonism in a 10-year-old female patient with WD. WD is a rare autosomal recessive disorder characterized by impaired copper transport and subsequent copper accumulation, leading to severe hepatic and neurologic disease. While the disease typically presents with hepatic and neurologic symptoms in pediatric patients, the occurrence of Parkinsonism is relatively rare, highlighting the importance of considering WD as a potential etiology in young patients presenting with movement disorders. Early diagnosis and appropriate management are crucial to prevent irreversible neurological damage and improve patient outcomes. Long-term monitoring is essential to ensure treatment efficacy and prevent disease progression. The presented case highlights the significance of considering WD

in the differential diagnosis of movement disorders in pediatric patients. Vigilance among the healthcare providers and ongoing research efforts are crucial in improving the care and prognosis of individuals affected by this rare genetic disorder.

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CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

DECLARATION OF PATIENT CONSENT

The authors declare that patient consent was taken for the publication.

ABBREVIATIONS

KF rings: Kayser-Fleischer rings; **MRI:** Magnetic Resonance Imaging; **THP:** Trihexyphenidyl; **WD:** Wilson's disease.

SUMMARY

Wilson's Disease (WD) is a rare genetic disorder marked by impaired metabolism of copper as a result of mutations in the ATP7B gene, which leads to copper accumulation in several organs. We report a case of a 10-year-old female patient, who had presentation Parkinsonism as a manifestation of WD. Despite no family history, she displayed neurological symptoms including tremors, stiffness, and dystonic movements. Low serum ceruloplasmin, increased urine copper excretion, KF rings, and hyper-intensities in the basal ganglia on MRI all supported the diagnosis. It emphasizes the significance of considering WD into account in young patients with movement disorders since prompt therapy initiation and long-term monitoring are essential to prevent irreversible neurological damage.

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