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# Neurological Wilson's Disease in Adolescents: A Case Series from South India

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**Abstract**

Wilson's disease is an autosomal recessive disorder that disrupts copper metabolism, leading to serious consequences in the liver and brain, including cirrhosis and the characteristic Kayser-Fleischer (KF) ring in the cornea. Our study focuses on children who presented with neurological symptoms, with or without hepatic involvement. While there have been a few reports from northern India, data on pediatric cases in southern India are sparse, highlighting the need for more comprehensive research in this region. In this series, we report on nine adolescent children diagnosed with neurological manifestations of Wilson's disease who presented to a tertiary care center. We collected relevant clinical histories, along with details from physical and neurological examinations, from the medical record system. This comprehensive data collection aimed to provide a clearer understanding of the clinical presentation and progression of neurological Wilson's disease in our patient cohort. Wilson's disease should be considered as one of the initial differential diagnoses even when there is only one neurological manifestation and without hepatic involvement in adolescents as well.

**Keywords:** Wilson's disease; Neurological manifestations; Kayser - Fleischer ring; adolescents; hepatomegaly

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## Introduction

Wilson's disease is an autosomal recessive disorder that disrupts copper metabolism, leading to serious consequences in the liver and brain, including cirrhosis and the characteristic Kayser-Fleischer (KF) ring in the cornea. First described in 1921, it is primarily caused by mutations in the *ATP7B* gene located on chromosome 13q14.[1]

Hepatic symptoms typically appear in childhood, but over time, neuropsychiatric symptoms become increasingly significant[2], with about 60% of patients developing neurological manifestations, commonly between ages 20 and 40.[3] While hepatic involvement is prevalent, there is limited data on patients with neurological symptoms alone. Common neurological presentations include dystonia, ataxia with postural and intentional tremors, and paediatric symptoms like hypokinesia, rigidity, and resting tremors.[3]

Diagnosis involves various tests, including complete blood count, liver function tests, low serum ceruloplasmin levels, and high urinary copper excretion, with KF rings confirmed via slit lamp examination. Neuroimaging can further support the diagnosis.[4]

Current management primarily includes pharmacological treatments like penicillamine, trientine, and zinc, although gene therapies using adenoviruses are being explored.

Our study focuses on children who presented with neurological symptoms, with or without hepatic involvement. While there have been a few reports from northern India, data on pediatric cases in southern India are sparse, highlighting the need for more comprehensive research in this region.

In this series, we report on nine adolescent children diagnosed with neurological manifestations of Wilson's disease who presented to a tertiary care centre in Puducherry between July 2015 and July 2023. There was a total of 14 adolescents diagnosed with Wilson's disease in our hospital. In that, only 9 of them had neurological manifestations and were included in this case series. We retrospectively collected relevant clinical histories, along with details from physical and neurological examinations, from the medical record system. Written informed consent for publication was obtained from parents, and the assent form was signed by the children.

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## Case Report

### *CASE 1*

*Clinical Features:*

Drooling, slurring, and slowing of speech (1 year), dystonia, dysarthria, tremor, micrographia, intermittent right-thumb dystonia, KF ring, hepatomegaly.

*Key Investigations:*

Abnormal LFT; normal blood counts; serum ceruloplasmin 11 mg/dl; 24-hr urinary copper 144 mcg/day.

USG: coarse liver texture, splenomegaly.

*MRI Findings:*

Bilateral hyperintensity in the caudate nucleus and putamen. But Face of the Giant Panda" or "Miniature Panda" signs were not observed

*Leipzig Score:*

7 (diagnostic of Wilson disease).

*Treatment:*

D- Penicillamine, Calcium, and Zinc supplementation.

### *CASE 2*

*Clinical Features:*

Difficulty in eating, chewing, writing, walking, buttoning; increased toe grip (5 months); worsening over 2 months; dystonia, KF ring, hypertonia, knee hyperextension, abnormal ankle jerk; family history (sibling with WD). No hepatic symptoms.

*Key Investigations:*

USG liver: chronic liver disease; serum ceruloplasmin 9 mg/dl; 24-hr urinary copper 264 mcg/day.

*MRI Findings:*

Basal ganglia hyperintensities

*Leipzig Score:*

7.

*Treatment:*

Penicillamine, Pacitane, Pyridoxine, Zinc supplementation.

### *CASE 3*

*Clinical Features:*

Slurred speech and abnormal movements (2 years); KF ring; hepatosplenomegaly; family history (brother died with jaundice).

*Key Investigations:*

Leucopenia and thrombocytopenia; serum ceruloplasmin 13 mg/dl; 24-hr urinary copper 114 mcg/day.

*MRI Findings:*

Basal ganglia hyperintensities

*Leipzig Score:*

6.

*Treatment:*

Because of Leucopenia and thrombocytopenia, Only Zinc was started.

### *CASE 4*

*Clinical Features:*

Blurring of vision (3 years); limb stiffness (18 days); tremors (15 days); dysphagia (8 days); inability to sit/walk (5 days); involuntary eye closure (2 days); neck pain (1 day). No hepatic symptoms.

Exam: KF ring, dystonia, spasticity of all limbs, flexed posture.

*Key Investigations:*

Serum ceruloplasmin 9 mg/dl; 24-hr urinary copper 176 mcg/day.

USG: liver parenchymal disease.

*MRI Findings:*

Basal ganglia hyperintensities

*Leipzig Score:*

6.

*Treatment:*

Zinc supplements, Clonidine. (Chelators (especially D-penicillamine) may initially worsen neurological symptoms in some patients. Hence, Zinc was given because it avoids the rapid mobilization of copper.)

### *CASE 5*

*Clinical Features:*

Slurred speech and drooling (1.5 years); tremors, daytime somnolence, agitation, reduced activity (20 days); KF ring; palatal dysarthria; bilateral ptosis; mask-like facies; spastic gait and rigidity.

*Key Investigations:*

Serum ceruloplasmin 6 mg/dl; 24-hr urinary copper 199 mcg/day.

*MRI Findings:*

Basal ganglia hyperintensities.

*Leipzig Score:*

8.

*Treatment:*

Zinc supplementation. (Chelators (especially D-penicillamine) may initially worsen neurological symptoms in some patients, Hence, Zinc was given because it avoids rapid mobilization of copper.)

### *CASE 6*

*Clinical Features:*

Limb stiffening (1 year); unable to walk (1 month); dystonia; hypertonia in all limbs; family history (sibling with WD).

*Key Investigations:*

Serum ceruloplasmin 11 mg/dl; 24-hr urinary copper 167 mcg/day.

*MRI Findings:*

Basal ganglia hyperintensities

*Leipzig Score:*

6.

*Treatment:*

Zinc supplementation. (Chelators (especially D-penicillamine) may initially worsen neurological symptoms in some patients, Hence, Zinc was given because it avoids rapid mobilization of copper).

***CASE 7***

*Clinical Features:*

Slurred speech (1 month); severe thinness; clubbing; mild splenomegaly; KF ring.

*Key Investigations:*

Serum ceruloplasmin 14 mg/dl; 24-hr urinary copper 104 mcg/day.
USG: chronic liver disease, mild portal hypertension.

*MRI Findings:*

Bilateral basal ganglia hyperintensities.

*Leipzig Score:*

7.

*Treatment:*

Zinc acetate, Penicillamine, Pyridoxine.

***CASE 8***

*Clinical Features:*

Dysarthria and abdominal distension (4 months); ascites; KF ring; no hepatomegaly; LFT normal.

*Key Investigations:*

USG: degenerative liver changes, moderate ascites. Serum ceruloplasmin 8 mg/dl; 24-hr urinary copper 118 mcg/day.

*MRI Findings:*

Basal ganglia hyperintensities

*Leipzig Score:*

6.

*Treatment:*

Zinc supplementation. (Chelators (especially D-penicillamine) may initially worsen neurological symptoms in some patients. Hence, Zinc was given because it avoids rapid mobilization of copper.)

***CASE 9***

*Clinical Features:*

Giddiness (9 months); difficulty speaking, swallowing; gait abnormality (3 days); handwriting change; prior jaundice at age 10; icterus; risus sardonicus; KF ring; left-hand clawing; elbow flexion.

*Key Investigations:*

Serum ceruloplasmin 5 mg/dl; 24-hr urinary copper 214 mcg/day.
USG: chronic liver disease.

*MRI Findings:*

Hyperintensity in the basal ganglia, external capsule, ventrolateral thalami, and midbrain.

*Leipzig Score:*

7.

*Treatment:*

Penicillamine 250 mg, Zinc acetate. No renal/Fanconi features, no psychiatric manifestations, no hemolytic anemia. No patient required liver transplantation. No mortality reported (Table and Figure 1).

## Discussion

Wilson's disease is a hereditary disorder of copper metabolism resulting from ATP7B mutations, leading to impaired biliary excretion and progressive copper accumulation in the liver and brain.[1, 2] Although classically described with hepatic onset in childhood and neurological onset in young adults, increasing evidence shows that neurological presentation in adolescents is not uncommon. In our series of nine patients aged 10–16 years, the predominant mode of presentation was neurological, aligning with reports that extrapyramidal and dystonic features frequently characterize juvenile-onset disease.[3-5]

The typical age of onset reported in literature ranges from late childhood to mid-adolescence, with many cohorts showing a median age around 13 years.[6] Our findings are comparable to the large Indian series from NIMHANS, where 38% of children under 18 years presented with neurological manifestations at onset.[7, 8] Similarly, the comprehensive review by Członkowska et al. highlighted that although hepatic involvement predominates in pediatric age groups, 5–15% may initially manifest with neurological symptoms, often resembling adult phenotypes.[9] The exclusively neurological onset in several of our patients, despite their young age, reinforces this pattern and parallels earlier Indian observations emphasizing diagnostic vigilance in adolescents.[10, 11]

In contrast to studies reporting male predominance in Wilson's disease[12], five of our nine cases were female. Family history was present in three patients, consistent with the 30–50% familial clustering reported across Indian cohorts.[10, 12] Neurological features in our patients—dystonia, dysarthria, tremors, gait abnormalities, and cognitive changes—mirror previously described juvenile presentations, which primarily exhibit extrapyramidal dysfunction rather than cerebellar or psychiatric features seen more commonly in adults.[3, 12 13]

Despite predominantly neurological symptoms, most of our patients had ultrasonographic evidence of hepatic parenchymal changes. This supports international recommendations that Wilson's disease should be considered even when hepatic symptoms are subtle or absent.[5, 6] All cases showed the classic biochemical signature of low ceruloplasmin and elevated 24-hour urinary copper, reinforcing the diagnostic reliability of these markers across clinical phenotypes.

Overall, despite its small size, our series highlights key observations seen in both global and Indian literature: neurological-onset Wilson's disease is not rare in early adolescence; hepatic abnormalities may be silent yet detectable; and early recognition is essential to prevent irreversible neurological progression.

## Conclusion

Wilson's disease should be considered as one of the initial differential diagnoses even when there is only one neurological manifestation and without hepatic involvement in adolescents as well.

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## Competing Interests
The authors declare no conflict of interest.

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