



Case Study

INTEGRATIVE MANAGEMENT OF WILSON'S DISEASE WITH AYURVEDIC INTERVENTIONS: A PAEDIATRIC CASE REPORT WITH BIOCHEMICAL OUTCOMES

Purohit Kosha B^{1*}, Chauhan Vishal D², Patel Rutu³, Vhora Maleka⁴

^{*1}PG Scholar, ³Professor and HOD, ⁴Professor, PG Dept. of Kaumarbhritya, J.S. Ayurveda Mahavidhyalaya, Nadiad, Gujarat, India.

²3rd year PG Scholar, PG Department of Kriyasharira, J.S. Ayurveda Mahavidhyalaya, Nadiad, Gujarat, India.

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ABSTRACT

Wilson's disease (WD) is a hereditary disorder characterized by defective copper metabolism due to ATP7B gene dysfunction, resulting in progressive accumulation of copper in the liver, brain and other tissues. **Methodology:** In July 2024, an 11-year-old boy presented with known case of Wilson's disease for two years with generalized pallor, generalized weakness, impaired cognition function such as lack of communication, reduced appetite and fatigue. **Clinical findings:** Laboratory findings revealed markedly reduced serum ceruloplasmin 2.62mg/dL and elevated urinary copper excretion 345.58µg/24 hrs and a slit lamp assay for the KF ring was negative. **Treatment:** The patient was managed with Ayurvedic formulations *Gandhaka Rasayana* 2gm twice daily, *Sharapunkha Churna* 1.5gm twice daily after food with water, *Bhumyamalaki Churna* 1.5gm twice daily, after food water and *Brahma Rasayana* 5gm twice daily along with *Pathya Ahara* and *Vihara*. **Results:** A significant reduction in urinary copper levels was observed along with improvement in appetite, physical strength and cognitive performance. **Conclusion:** The observed clinical and biochemical improvement suggests that such an integrative approach may be beneficial in the comprehensive management of Wilson's disease.

INTRODUCTION

Wilson's disease (hepatolenticular degeneration) is an autosomal recessive disorder that can be associated with liver disease, degenerative changes in the brain and Kayser-Fleischer (K-F) rings in the cornea. The incidence is approximately 1/30,000 births worldwide. The abnormal gene for Wilson's disease is found on chromosome 13 encodes ATP7B, a copper transporting P-type adenosine triphosphatase (ATPase) which is mainly expressed in hepatocytes, it is critical for biliary copper excretion and for copper incorporation into ceruloplasmin. Absence or malfunction of ATP7B results in decreased biliary copper excretion and diffuse accumulation of copper in the cytosol of hepatocytes.

WD includes hepatomegaly (with or without splenomegaly), subacute or chronic hepatitis and acute hepatic failure, cryptogenic cirrhosis, portal hypertension, ascites, oedema, variceal bleeding and other hepatic dysfunction. Neurologic symptoms can be preceded by as long as 10 years.^[1,2]

From an Ayurvedic perspective, this condition can be interpreted under the concept of *Sahaja Vyadhi* (congenital disorder), which originates due to defects in *Beeja* (sperm/ovum), *Beejabhaga*, or *Beejabhagavayava* during *Garbha Kala* (fetal development). The ATP7B gene mutation may be considered analogous to *Beejabhaga Dushti*,^[3] resulting in defective metabolic processes from birth. The clinical manifestations of Wilson's disease, can be correlated with *Vata-Pitta Dushti*. *Pitta* plays a crucial role in metabolic processes, including transformation and biochemical activities, while *Vata* governs neurological functions and movement.

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Patient Information

An 11-year-old boy, diagnosed with Wilson's disease in May 2022 with complaints of vomiting and lethargy, followed by the development of jaundice. Laboratory investigations revealed elevated liver enzymes, with SGPT (ALT) of 110 IU/L (11/05/2022) and SGOT (AST) of 87 IU/L, along with raised serum bilirubin levels, suggestive of hepatocellular injury. Screening for infectious aetiologies, including HAV IgM, HEV IgM, HBsAg, and HIV, was found to be non-reactive. Ultrasonography of the abdomen demonstrated mild splenomegaly and the liver showed significant heterogeneous echotexture with nodularity and surface irregularity, suggestive of chronic liver parenchymal disease (figure 3). Subsequently, 24-hour urinary copper excretion was found to be markedly elevated 345.58µg/24hr (table no. 1). Based on the clinical presentation, biochemical abnormalities and radiological findings the diagnosis of Wilson's disease was confirmed.

Birth history: The patient was born by full-term normal delivery with a birth weight of 2.9kg, cried immediately after birth, and had no perinatal complications.

Family history: Non-contributory as per parental report, and developmental milestones were appropriate for age with no history of delay.

Personal history: The patient follows a vegetarian diet, with reduced appetite, sound sleep, regular bowel habits (once daily) and normal micturition frequency (6-7 times per day).

Clinical findings: Vital parameters were within normal limits. On systemic examination, per abdominal examination showed a soft and non-tender abdomen, with no palpable hepatomegaly or splenomegaly. Central nervous system examination revealed that the patient was conscious and well-oriented to time and place; however, mild impairment in cognitive function was noted.

Diagnostic assessment

The Leipzig scoring system^[4] standardizes the diagnosis and management of Wilson disease. A score ≥ four confirms diagnosis; this patient scored six, indicating definite Wilson's disease.

Laboratory investigations mentioned in table no. 1 also confirms the diagnosis.

Table 1: Lab Investigations

Year	Date	Urinary Copper	
		Urinary copper (ICPMS) µg/L	Urinary copper (24 hr)
2022	17/05/2022	150.25 µg/L (↑)	345.58 µg/24hr (↑)
	05/08/2022	430.94 µg/L (↑)	818.79 µg/24hr (↑)

Timeline: Timeline of the study described in table 2.

Table 2: Timeline of the Study

Time	Disease condition	Treatment
May 2022	Diagnosed as Wilson's Disease.	Started allopathic medications Cap. Cilamin (D-penicillamine-250 mg), one tablet in the morning - one tablet in the evening. Tab. Inderal (20 mg) - 1tab BDS Tab. Benadone - one tablet on alternate days.
2023	No satisfactory clinical improvement	Ayurvedic medications from other sources.
14/06/2024 (Before Treatment)	Increased Urinary copper 861.74 µg/24hr (figure no.1)	Cap. Cilamin (D-penicillamine-250 mg), two tablets in the morning - one tablet in the evening. Tab. Inderal (20 mg) - one tablet TDS Tab. Benadone - one tablet on alternate days.
July 2024 (Initial Visit)	Patient presented with generalized pallor, weakness and impaired cognition function such as lack of communication, impaired appetite.	Patient attended the hospital on an OPD basis. Integrative treatment was initiated. Ayurvedic medications: <i>Gandhaka Rasayana</i> 2gm BD, <i>Sharapunkha Churna</i> 1.5gm BD, <i>Bhumyamalaki Churna</i> 1.5gm BD, and <i>Brahma Rasayana</i> 5gm BD, along with ongoing allopathic therapy.

July 2024 - October 2024	Monthly follow-up showed gradual improvement in appetite, energy level, and communication.	Continued the same Ayurvedic and allopathic treatment with monthly clinical monitoring.
October 2024 - February 2025 (Follow-up)	Progressive improvement observed. Weight increased to 28.8 kg, height 130cm. Better school performance with marked improvement in communication and cognition.	Continued Ayurvedic medications (<i>Gandhaka Rasayana</i> , <i>Sharapunkha Churna</i> , <i>Bhumyamalaki Churna</i> and <i>Brahma Rasayana</i>) along with allopathic treatment. Regular monthly follow-up.
27/02/2025 (Follow-up)	Twenty-four-hour urinary copper reduced to 378.4µg/24hr (24/02/2025). Clinical condition improved.	Continued integrative treatment with dose reduction of allopathic medications as detailed in Table 6, along with continued Ayurvedic therapy.
March 2025 - February 2026	Continued monthly follow-up demonstrated continued clinical improvement, good appetite, improved cognition, and normal daily activities.	Continued reduced-dose allopathic medications with regular monitoring and supportive ayurvedic treatment.
23/02/2026 (Follow-up)	Significant improvement noted. Twenty-four-hour urinary copper reduced further to 117.0µg/24h (Figure 2). Weight increased to 31.6kg and height to 137cm.	Continued Ayurvedic treatment with maintenance dosage. Monthly follow-up maintained.
10/04/2026 Last follow up	Patient remained clinically stable. Weight 32.5 kg and height 144 cm.	Patient was advised to continue oral Ayurvedic medications at home: <i>Gandhaka Rasayana</i> 2 g twice daily after food with water; <i>Sharapunkha Churna</i> 1.5 g twice daily after food with water; <i>Bhumyamalaki Churna</i> 1.5 g twice daily after food with water; <i>Brahma Rasayana</i> 5gm twice daily before food with milk. Regular follow-up was advised.

Table 3: Ayurvedic therapeutic intervention

S.No.	Medicine	Dose	Frequency	Time of Administration	Anupana	Duration
1.	<i>Gandhaka Rasayana</i>	2 gm	Twice daily	After food	Water	July 2024 – April 2026
2.	<i>Sharapunkha Churna</i>	1.5gm	Twice daily	After food	Water	July 2024 – April 2026
3.	<i>Bhumyamalaki Churna</i>	1.5gm	Twice daily	After food	Water	July 2024 – April 2026
4.	<i>Brahma Rasayana</i>	5gm	Twice daily	Before food	Milk	July 2024 – April 2026

The treatment duration extended from July 2024 to April 2026. The patient was followed up monthly throughout the treatment period, with clinical assessment and biochemical monitoring, including serial 24-hour urinary copper estimation.

Pathya Ahara Vihara (Diet & Lifestyle)

Avoid copper-rich foods (nuts, chocolate) and include milk, ghee and light digestible diet and yoga and pranayama.

Follow-up and outcomes

Clinically, the patient demonstrated a gradual reduction in 24-hour urinary copper excretion (Table 4), along with improvement in serum copper and haemoglobin levels (Table 5). Based on the sustained clinical and biochemical improvement, the doses of allopathic medications were gradually tapered during follow-up (Table 6). Throughout the treatment period, the patient also showed progressive gains in weight and height, improved appetite, enhanced physical strength, and better cognitive function, evidenced by improved communication, enhanced memory, and better school performance. The patient tolerated the integrative treatment well, with no adverse events reported during the follow-up period. At the last follow-up, the patient remained clinically stable

Purohit Kosha B et al. Integrative Management of Wilson's Disease with Ayurvedic Interventions: A Paediatric Case Report and was advised to continue the prescribed oral Ayurvedic medications at home with regular follow-up. The patient continues to demonstrate steady clinical and biochemical improvement on ongoing follow-up.

Table 4: Before and after treatment Urinary Copper level

Year	Date	Urinary Copper	
		Urinary Copper (ICPMS) (< 50 µg/L)	Urinary Copper (<40µg/24 hours)
Before treatment 2024	14/06/2024	410.35 µg/L (↑)	861.74 µg/24hr (↑)
After treatment 2025	24/02/2025	103.67 µg/L (↑)	378.4 µg/24hr (↑)
	29/09/2025	205.98 µg/L (↑)	360.46 µg/24hr (↑)
2026	23/02/2026	3.0 µg/L	117.0 µg/24hr

Table 5: Serum copper and haemoglobin level

Date	Serum copper	Haemoglobin
01/07/2024	< 0.001 mg/dL	10.1gms/dL
27/01/2025	0.006 mg/dL	11.2gms/dL
20/03/2026	0.33 mg/dL	12.6gms/dL

Table 6: Dose reduction of medicine

Follow up date	Tab. Cilamin (250mg)	Tab. Inderal (20mg)
24/06/2024	Two tablets at morning - one tablet at evening	One tablet three times a day
27/02/2025	One tablet at morning - one tablet at evening	One tablet twice a day
26/09/2025	One tablet once in a day	One tablet once in a day
20/03/2026	One tablet once in a day	Discontinued

USG whole abdomen

12/05/2022: Cirrhosis of liver with mild splenomegaly findings of coarse hepatic echotexture with nodular surface changes were suggestive of cirrhotic involvement with mild splenomegaly.

26/09/2025: Changes of chronic liver parenchymal disease with mild splenomegaly. The liver (size-11.8cm) maintained a coarse echotexture but showed only mild surface irregularity, with no evidence of worsening nodularity, focal lesions, ascites, or features of portal hypertension. The spleen remained mildly enlarged with minimal change in size, indicating persistent but non-progressive splenomegaly.

DISCUSSION

In Ayurvedic classics there is no exact correlation is available for this disease entity that exactly matches the feature of Wilson's disease, but it can be correlated with the *Sahaja Vyadhi* (heredity) or *Janmajata Vyadhi* (congenital). It can be correlated with *Vata-Pitta Dushti* leading to metabolic derangement (*Dhatwagni Mandhya*). Due to excessive copper accumulation, main goal of treatment is reduction of copper in liver so we have given *Gandhaka Rasayana* which has anti-copper property,

The term sulphur derived From the Sanskrit word "*Shulvari*" (*Shulva + Ari*) meaning enemy of *Shulva* or copper metal (anti-copper)^[5]. It is known for its *Rasayana*, *Vishaghna* and detoxifying properties that may help regulate metal metabolism and oxidative stress. D-penicillamine contains a reactive thiol (sulfhydryl, -SH) group that gives it the ability to bind with heavy metals, including copper. It forms a stable, soluble complex with copper ions throughout the body, including in tissues where copper has accumulated. This new water-soluble copper-penicillamine complex cannot be reabsorbed by the renal tubules and is readily excreted from the body via the urine.^[6] *Sharpunkha* (Tephrosia purpurea) is known as *Plihasatru*,^[7] it has anti-inflammatory, diuretic, anthelmintic and hepatoprotective properties. It promotes bile secretion improves digestion and reduces portal hypertension. *Bhumyamalaki* is hepatoprotective, analgesic, anti-inflammatory and anti-bacterial properties. The protein isolate of *Phyllanthus niruri* indicates hepatoprotective effect against acetaminophen-induced toxicity.^[8] *Brahma Rasayana* provides *Dirghayu*^[9], improved strength, appetite, potent antioxidant, rejuvenating,

neuroprotective, nervine tonic, nootropic (cognitive enhancer), memory booster, cardioprotective, haematinic (increases haemoglobin levels).^[10] Together, they may help in restoring systemic balance and preventing disease progression.

Patient Perspective

The patient's family reported visible improvement in physical strength, better concentration, and overall satisfaction with treatment. They expressed a desire to continue Ayurvedic therapy under supervision.

CONCLUSION

This case demonstrates that Ayurvedic formulations can play a safe and effective therapeutic approach in Wilson's disease, particularly in reducing the adverse effects associated with long-term conventional therapy. The patient showed marked clinical and biochemical improvement, evidenced by a significant reduction in urinary copper excretion along with enhancement in general health, appetite, strength and cognitive function. Notably, the patient remained clinically stable throughout the follow-up period without any adverse effects.

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*Address for correspondence

Dr. Purohit Kosha B

PG Scholar,
PG department of Kaumarbhritya,
J.S. Ayurveda Mahavidhyalaya,
Nadiad, Gujarat.

Email:

princy.purohit8888@gmail.com

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