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CASE REPORT ON WILLISONS DISEASE

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ABSTRACT

Background: Wilson's disease is a rare autosomal recessive disorder of copper metabolism, often presenting in young adults with hepatic, neurological, or psychiatric manifestations. Early diagnosis is critical to prevent irreversible damage. **Case Presentation:** A 23-year-old male from Ballari, India, presented with progressive slurred speech, tremors, dysphagia, abnormal movements, hypoglycemia, altered sensorium, and behavioral changes. Examination revealed confusion, pallor, tremors, and dysarthria. Investigations confirmed low serum ceruloplasmin (<0.2 g/L), elevated serum (>250 µg/dL) and 24-hour urinary copper (>100 µg/24h), Kayser-Fleischer rings, elevated liver enzymes (AST >200 U/L, ALT >250 U/L), hyperammonemia (>100 µmol/L), diffuse liver parenchymal disease on USG, and metabolic encephalopathy on brain MRI. Diagnosis: Wilson's disease with neurological and hepatic involvement complicated by hypoglycemia and encephalopathy. **Management and Outcome:** Treated with D-penicillamine (1 g/day), zinc acetate (150 mg/day), lactulose, IV dextrose, and supportive care. He showed gradual sensorium improvement, tremor reduction, sugar level stabilization, and partial liver enzyme recovery, then discharged on long-term chelation therapy. **Conclusion:** This case underscores Wilson's disease as a key differential in young patients with combined neurological-hepatic symptoms. Prompt chelation improves outcomes, emphasizing the role of KF rings and copper studies in diagnosis.

KEYWORDS: Neurological and hepatic manifestations, D-penicillamine, Choreoathetotic movements, Hyperammonemia.

INTRODUCTION

Wilson's disease is a rare autosomal recessive inherited disorder involving abnormal copper metabolism that results in gradual accumulation of copper in several organs, particularly the liver, brain, cornea, kidneys, and other tissues. The condition arises due to pathogenic mutations in the ATP7B gene, which encodes a copper-transporting ATPase responsible for the excretion of copper into bile and its incorporation into ceruloplasmin. When this transport mechanism is impaired, copper cannot be properly eliminated from hepatocytes, leading to intracellular accumulation and progressive tissue toxicity.[1,2]

Wilson's disease is regarded as one of the most significant treatable inherited metabolic disorders because timely diagnosis and initiation of appropriate therapy can prevent irreversible hepatic and neurological complications.[3]

Epidemiology

Wilson's disease occurs globally and is estimated to affect approximately 1 in 30,000 individuals, although prevalence varies among different geographic regions.[4]

Studies from India suggest that the disorder may be underdiagnosed, and the true prevalence could be higher than previously reported. This may partly be due to genetic heterogeneity and relatively high rates of consanguinity in certain populations.[5]

Recent multicenter genetic investigations in India have estimated the prevalence to be approximately 1 in 18,678 individuals, with a carrier frequency of nearly 1 in 67, indicating the considerable clinical relevance of the disease in the Indian population.[6]

The onset of symptoms can occur at various ages; however, most patients present between 5 and 35 years. Hepatic manifestations generally appear earlier in life, whereas neurological symptoms more commonly develop during adolescence or early adulthood.[4,7]

Etiology and Genetics

Wilson's disease results from mutations in the ATP7B gene located on chromosome 13. The ATP7B protein plays a crucial role in maintaining copper balance by transporting excess copper into bile for excretion and enabling its incorporation into ceruloplasmin. When this process fails, copper progressively accumulates in hepatocytes and eventually spreads to other organs.[1,2]

More than 1300 ATP7B gene variants have been identified worldwide. Studies involving Indian populations demonstrate substantial genetic diversity, with several mutations appearing to be region-specific.[8]

Pathophysiology

Copper is an essential trace element that participates in numerous enzymatic and metabolic processes. Under normal physiological conditions, copper absorbed from the intestine is transported to the liver, incorporated into ceruloplasmin, and excreted primarily through bile. In Wilson's disease, impaired ATP7B function disrupts biliary excretion of copper, causing progressive accumulation within hepatocytes.[2]

Once the liver's storage capacity is exceeded, free copper enters the bloodstream and deposits in extrahepatic tissues, especially the basal ganglia, cornea, and kidneys. Excess copper promotes oxidative stress, mitochondrial dysfunction, lipid peroxidation, and cellular apoptosis, ultimately leading to tissue injury and organ dysfunction.[9]

Neurological manifestations occur mainly due to copper deposition in the basal ganglia and brainstem, resulting in movement disorders such as tremors, dysarthria, dystonia, and neuropsychiatric disturbances. Magnetic resonance imaging (MRI) frequently reveals characteristic abnormalities in these brain regions consistent with metabolic injury.[10]

Clinical Manifestations

The clinical features of Wilson's disease are diverse and may include hepatic, neurological, psychiatric, or systemic manifestations.[3]

Hepatic manifestations

Liver involvement can vary from asymptomatic elevation of liver enzymes to chronic hepatitis, cirrhosis, or even acute liver failure. Studies conducted in India indicate that severe hepatic disease, including acute-on-chronic liver failure, is a common presentation among younger patients.[6]

Neurological manifestations

Common neurological symptoms include tremor, dysarthria, dystonia, choreoathetoid movements, and parkinsonian features. Neuroimaging studies demonstrate abnormal findings in the majority of patients with neurological involvement.[5]

Psychiatric manifestations

Psychiatric symptoms such as personality changes, irritability, depression, and cognitive impairment may occur and, in some cases, may precede neurological symptoms.[3]

Other systemic manifestations

Copper accumulation may also affect the renal, skeletal, and hematological systems. One of the characteristic clinical signs is the presence of Kayser–Fleischer rings, which result from copper deposition in Descemet's membrane of the cornea and are particularly common in patients with neurological disease.[11]

Diagnosis

The diagnosis of Wilson's disease is established through a combination of clinical findings, biochemical investigations, and specialized tests. Important laboratory indicators include reduced serum ceruloplasmin levels and elevated urinary copper excretion. Ophthalmologic examination for Kayser–Fleischer rings, imaging studies, and genetic analysis also contribute to confirming the diagnosis.[3,9]

Diagnostic scoring systems that integrate these parameters have improved diagnostic accuracy and facilitate early recognition of the disease.

Advances in genetic testing and next-generation sequencing technologies have further enhanced diagnostic precision, particularly in countries like India where mutation patterns vary among populations.[8]

Prognosis and Importance of Early Treatment

If left untreated, Wilson's disease can be progressive and potentially fatal. However, the prognosis is generally favorable when therapy is initiated early and continued consistently. Treatment primarily involves chelating agents and medications that decrease intestinal copper absorption. Early intervention can prevent disease progression and significantly improve both neurological and hepatic outcomes in many patients.[5]

Despite the availability of effective therapy, delayed diagnosis remains a significant challenge, particularly in developing countries where diagnostic delays of up to two years have been reported. This highlights the need for greater clinical awareness and early screening to improve patient outcomes.[5]

CASE STUDY

A 23-year-old male from Ballari, Karnataka, presented to the Ballari Medical College and Research Centre for admission in January 2026 with progressive slurred speech, dysphagia, tremor of both upper limbs, abnormal choreoathetotic movements, repeated episodes of hypoglycemia, altered sensorium and behavioral disturbance over a period of weeks, and no history of alcohol or other drug use.

On examination, the patient was conscious but confused. He was pale, without icterus or oedema. Based on the vital signs, including a blood pressure of 120/80 mmHg, pulse of 88 bpm, and SpO2 of 97%, there was no obvious stress on the cardiovascular or respiratory systems, and the abdomen was soft with no organomegaly.

Investigations revealed a very low serum ceruloplasmin (< 0.2 g/L), high serum copper concentrations (> 250 µg/dL), high urinary copper levels (> 100 µg over 24 hours) and raised levels of AST/ALT (> 200/250 U/L), low levels of albumin and hyperammonemia (> 100 µmol/L), low glucose levels (< 70 mg/dL), diffuse liver parenchymal disease noted on ultrasound examination of the abdomen, basal ganglia changes on MRI of the brain consistent with metabolic encephalopathy and the presence of Kayser-Fleischer rings by the ophthalmologist.

A diagnosis of Wilson's disease with neurological and hepatic manifestations complicated by hypoglycemia and encephalopathy was made. Treatment included D-penicillamine (1 g per day), zinc acetate (150 mg per day), lactulose, intravenous dextrose, vitamins, ICU monitoring, nutrition, and physiotherapy. The patient demonstrated gradual improvement of sensorium, reduction of tremor, stabilization of the blood glucose level and partial recovery of liver function tests prior to being discharged in a stable condition on lifelong chelation therapy.

Laboratory investigations

PARAMETERS	D1	D3	D7	REFERENCE
HEMATOLOGY				
Hemoglobin	15.7	17.9	14.8	13-18gm/dL
Total WBC count	16070	9640	7350	4000-11000cells/cumm
Neutrophils	83	85	75	40-70%
Lymphocytes	12	11	20	20-40%
Eosinophils	02	02	04	3-6%
Monocytes	03	04	02	2-10%

Basophils	0	0	0	0-1%
RBC count	5.52	6.30	5.37	4.5-6.0 million/mm^3
Platelet count	3.72	2.97	2.36	1.5-4.5lakh/ mm^3
MPV	7.6	8.5	8.15	7-11fl
PCV	43.7	44.2	42.1	45-55%
MCV	77.2	78	-	80-100fl
MCH	28.5	28.4	-	27-34pg
MCHC	37.0	36.4	32.7	31-36%
RDW-CV	12.9	13.0	12.7	11.5-14.5%
PDW-CV	19.7	10.4	20.3	10-18%
Random blood sugar	40	138	169	70-140mg/dl
S. ELECTROLYTE				
Sodium blood	139	140	123	136-145mEq/L
Potassium blood	4.2	4.4	4.9	3.48-5mEq/L
Chloride blood	102	105	103	96-106mEq/L
LIVER FUNCTION TEST				
Total protein	7.5	-	-	6-8.3g/dl
Albumin	4.1	-	-	3.2-5.4g/dl
Globulin	3.3	-	3.4	2.5-3g/dl
A/G ratio	1.2	-	0.9	1.2-1.5
Bilirubin Total	0.2	-	0.2	0.2-1.2mg/dl
Bilirubin Conjugated	0.1	-	0.1	0.1-0.4mg/dl
Bilirubin Unconjugated	0.1	-	0.1	0.2-0.7mg/dl
Alanine Tran aminase	27	-	13	0-45IU/L
Aspartate Transaminase	87	-	28	0-40IU/L
Alkaline Phosphatase	208	-	56	20-140U/L
RENAL PROFILE				
Serum Creatinine	2.7	2.2	1.9	0.7-1.4mg/dl
Blood urea	124	94	103	15-45mg/dl
CRP	14	-		0-6mg/L
Serum Ammonia		29.6	47.9	27-138 U/ml

- **Sonography of Abdomen:** Diffused liver parenchymal disease. B/L Renal parenchymal disease. Mild Ascites, mild left pleural effusion.

Treatment

SL.NO	MEDICATIONS	FREQ	ROUTE	DAYS
01	Inj 25% Dextrose	1-1-1	IV	D1 - D6
02	Inj Pantoprazole	1-0-0	IV	D1 - D9
03	Inj Ondansetron	1-1-1	IV	D1 - D9
04	Inj Ceftriaxone	1-0-1	IV	D1 – D7
05	Inj Furosemide	1-1-0	IV	D1 – D4
06	Tab Zinc	1-1-1	PO	D1 – D9
07	Tab Penicillamine	1-0-0	PO	D1 – D5
08	Tab THP	1-1-1	PO	D1 - D9
09	Tab B complex	1-0-1	PO	D1 – D6
10	Inj Albumin	STAT	IV	D4
11	Inj 3% NaCl	STAT	IV	D4

DISCUSSION

Wilson's disease (WD), an inherited disorder caused by genetic abnormalities in the ATP7B gene located on chromosome 13, results in dysfunctional copper removal from the liver into bile and subsequent accumulation of copper in the liver, brain, cornea, and other tissues of the body, resulting in toxicity. This male patient is 23 years old and demonstrates a classic neurological-hepatic phenotype of WD including slurred speech (dysarthria), upper limb tremor, dysphagia, choreoathetotic movements, behavior changes, hypoglycemia and fluctuating sensorium, which is typically observed between the ages of 20-30 years. Family history suggests recessive inheritance; it is reported that there is a carrier frequency of 1:100 people in India. Kayser-Fleischer (KF) rings are present in 95% of patients with neurological WD and help confirm the diagnosis according to the Leipzig criteria which include a ceruloplasmin level of less than 0.2 g/L, a 24-hour urinary copper level of more than 100 µg, and an elevated serum copper level. The chronic parenchymal disease due to hepatic dysfunction is evidenced by elevated serum AST and ALT levels, a low albumin level, and a high serum ammonia level. Hypoglycemia may have resulted from impaired gluconeogenesis in copper-toxic hepatocytes. MRI changes due to copper deposition in the basal ganglia could mimic a metabolic encephalopathy. Patients with WD may be treated using D-penicillamine chelation (1 g/day) to mobilize copper, along with zinc to prevent its absorption from the intestines. Supportive treatment with lactulose/ammonia scavengers treated the encephalopathy, and the patient was stabilized using IV dextrose to restore glucose levels. The patient experienced partial recovery from his tremors/sensorium after treatment; the literature indicates that approximately 70-80% of patients who receive treatment early will have a neurological response, and therefore approximately 20-30% of patients will have residual neurological deficits due to gliosis.





CONCLUSION

Wilson's disease is one of the most critical and treatable forms of neurological and hepatic dysfunction in young adults. Early detection via Kayser-Fleischer rings, low ceruloplasmin and high copper excretion often allows for a significant improvement in prognosis from irreversible damage to substantial recovery. Prompt chelation therapy, using either D-penicillamine or zinc, in combination with supportive care, illustrates the importance of increased clinical suspicion in patients presenting with dysarthria, tremors, and unexplained hypoglycemia and consider family members with related diagnoses. Lifelong therapy is necessary to avoid relapse and/or further progression into multiple organ systems.

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