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WILSON DISEASE PRESENTING AS ACUTE FEBRILE ILLNESS WITH SEIZURES AND ACUTE LIVER DYSFUNCTION: A CASE REPORT

Hepatology

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ABSTRACT

Background: Wilson disease is a rare autosomal recessive disorder of copper metabolism caused by mutations in the ATP7B gene, leading to impaired copper excretion and accumulation in organs such as the liver and brain. **Case Presentation:** We report a 19-year-old male presenting with acute febrile illness, seizures, and altered sensorium. Initial evaluation suggested an infectious etiology. However, further investigations revealed markedly elevated liver enzymes, hyperammonemia, low serum ceruloplasmin levels, and elevated urinary copper excretion. Slit-lamp examination demonstrated Kayser–Fleischer rings. MRI brain showed symmetrical hyperintensities in the basal ganglia. **Conclusion:** Wilson disease can present atypically as acute febrile illness with neurological manifestations. Early recognition and treatment are essential to prevent irreversible organ damage.

KEYWORDS

Wilson disease, Kayser–Fleischer ring, basal ganglia, acute liver failure, case report

1. INTRODUCTION

Wilson disease (WD) is an inherited disorder of copper metabolism caused by mutations in the ATP7B gene, resulting in decreased biliary copper excretion and accumulation in hepatocytes and extrahepatic tissues. The global prevalence is approximately 1 in 30,000 individuals.

Clinical manifestations are variable and include hepatic, neurological, and psychiatric features. Hepatic involvement ranges from asymptomatic elevation of liver enzymes to acute liver failure. Neurological manifestations include tremors, dystonia, dysarthria, and behavioral disturbances.

Kayser–Fleischer rings, caused by copper deposition in the cornea, are a characteristic finding, especially in patients with neurological involvement.

This case highlights an atypical presentation of Wilson disease as an acute febrile illness with seizures.

2. CASE PRESENTATION

A 19-year-old male presented with fever for four days, associated with headache and generalized abdominal pain. He subsequently developed multiple episodes of generalized tonic–clonic seizures with postictal confusion.

There was no history of chronic illness, drug intake, or prior neurological symptoms.

On examination, the patient was febrile, tachycardic, had altered sensorium, and icterus was present. Neurological examination suggested metabolic encephalopathy without focal deficits.

Laboratory investigations revealed AST and ALT >3000 IU/L, elevated serum ammonia, negative viral markers, microcytic hypochromic anemia, low serum ceruloplasmin levels, and elevated 24-hour urinary copper excretion.

Slit-lamp examination revealed Kayser–Fleischer rings.

Based on clinical, biochemical, and radiological findings, a diagnosis of Wilson disease presenting as acute liver dysfunction with neurological involvement was made.

The patient was managed with supportive care, antiepileptics, and ammonia-lowering measures. The patient showed clinical improvement during hospitalization.

MRI brain showed symmetrical hyperintensities in bilateral basal ganglia consistent with metabolic encephalopathy.

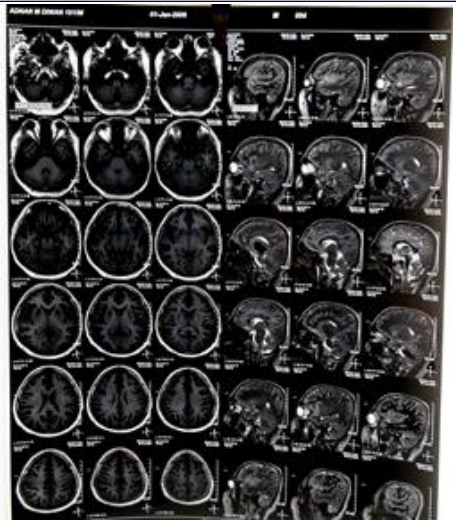

Figure 1: MRI brain showing bilateral basal ganglia hyperintensities.

3. DISCUSSION

Wilson disease results from impaired copper transport leading to toxic accumulation in the liver and brain.

Acute liver failure is a rare but severe presentation and may mimic viral hepatitis or sepsis, making early diagnosis challenging.

Diagnosis is based on low serum ceruloplasmin, elevated urinary copper excretion, and the presence of Kayser–Fleischer rings.

MRI findings typically include symmetrical hyperintensities in the basal ganglia, thalamus, and brainstem.

The Leipzig scoring system is useful in equivocal cases.

Treatment includes copper chelation therapy (penicillamine, trientine), zinc therapy, dietary copper restriction, and liver transplantation in severe cases.

This case is notable for its atypical presentation as acute febrile illness with seizures, emphasizing the need for high clinical suspicion.

4. CONCLUSION

Wilson disease should be considered in young patients presenting with unexplained hepatic dysfunction and neurological symptoms. Early

diagnosis and prompt treatment significantly improve prognosis and prevent irreversible complications.

ETHICAL APPROVAL
Not required for case report as per institutional guidelines.

CONSENT
Written informed consent was obtained from the patient for publication.

CONFLICT OF INTEREST
None declared.

FUNDING
None.

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