Case Report: Wilson's Disease Presenting as Chronic Liver Disease in a Non-Alcoholic Adult Male

Authors

Dr. Asifa Tabassum Bilgi, MD[1], Dr. Muddasir Mohammed Fariduddin, MD[1], Dr. Ahmed Mohammed Fariduddin, MBBS[2]

Affiliations

[1] Department of Internal Medicine, Ayaan Institute of Medical Sciences, Hyderabad, Telangana, India

[2] Faith Hospital, Hyderabad, Telangana, India

Corresponding Author

Dr. Asifa Tabassum Bilgi

Department of Internal Medicine, Ayaan Institute of Medical Sciences, Hyderabad, Telangana, India

Email: ambilgi@gmail.com

Abstract

Objective: This study aims to highlight the importance of early recognition and diagnosis of Wilson's disease (WD) in young adults presenting with chronic liver disease (CLD) of unknown etiology.

Design, Materials, and Methods: A single case study of a 30-year-old non-alcoholic male presenting with CLD and negative viral hepatitis markers. A comprehensive evaluation including serological tests, 24-hour urinary copper levels, and ceruloplasmin levels was conducted to confirm the diagnosis of WD.

Results: The patient exhibited hepatic manifestations without neurological symptoms. Despite a negative Kayser-Fleischer (KF) ring, elevated urinary copper excretion and reduced ceruloplasmin levels led to a confirmed diagnosis of WD. Early initiation of copper chelation therapy with penicillamine and zinc supplementation resulted in significant clinical and biochemical improvement over three months.

Conclusion: Wilson's disease should be considered in young patients with unexplained CLD, even in the absence of neurological symptoms and KF rings. Prompt diagnosis and treatment can prevent disease progression and improve long-term outcomes.

Keywords: Wilson's disease, chronic liver disease, copper metabolism, chelation therapy, ATP7B gene

Introduction

Wilson's disease is an uncommon genetic disorder inherited in an autosomal recessive pattern due to mutations in the ATP7B gene. This genetic alteration impairs the liver's ability to transport and excrete copper effectively, leading to excessive copper accumulation, particularly in the liver and brain. As a result, affected individuals may develop hepatic, neurological, and psychiatric symptoms.

The condition typically emerges between the ages of 5 and 35, with a highly variable clinical spectrum ranging from asymptomatic liver dysfunction to acute liver failure and severe neurological impairment. Although

Wilson's disease is manageable with appropriate treatment, delays in diagnosis can result in irreversible organ damage and potentially fatal complications.

Due to its diverse presentation, Wilson's disease should be suspected in young adults with unexplained chronic liver disease (CLD), especially when viral, autoimmune, or metabolic causes have been excluded. This report emphasizes the significance of early detection, accurate diagnosis, and timely intervention in preventing disease progression and improving patient outcomes.

Case Presentation

A 30-year-old male, with no history of alcohol consumption, presented with progressive jaundice, abdominal distension, and lower limb swelling over the past 4 months. He had no history of neurological symptoms such as tremors, behavioral changes, or dysarthria. His family history was unremarkable for liver disease or metabolic disorders. On examination, he exhibited icterus, pedal edema, and ascites, while neurological assessment was normal. Routine liver function tests showed elevated transaminases and bilirubin, with a low serum albumin level. Viral hepatitis markers (HBV, HCV) were negative. Further investigations, including a 24-hour urinary copper test, revealed elevated copper excretion, and serum ceruloplasmin was found to be reduced. These findings confirmed the diagnosis of Wilson's disease, leading to the initiation of copper chelation therapy and supportive care.

Diagnosis: Wilson's Disease with Hepatic Manifestations (Neurologically Asymptomatic)

Management

Copper Chelation Therapy:

- Penicillamine (250–500 mg/day, gradually increased)
- Trientine (alternative for penicillamine intolerance)

Zinc supplementation: To reduce intestinal copper absorption

Liver support measures: Diuretics for ascites, nutritional counseling

Regular Monitoring: Liver function tests, urinary copper levels, ceruloplasmin

Outcome & Follow-Up

- Significant improvement in liver parameters after three months of treatment
- Resolution of icterus, ascites, and pedal edema
- Normalization of liver function tests
- Continued long-term therapy to prevent disease progression

Discussion

Wilson's disease should always be considered in young individuals presenting with chronic liver disease (CLD) of unknown origin, particularly when common causes such as viral hepatitis, autoimmune disorders, and alcohol-related liver disease have been excluded. Given its diverse clinical presentation, diagnosing Wilson's disease can be challenging, especially in cases where neurological symptoms are absent, and Kayser-Fleischer (KF) rings are not detectable on slit-lamp examination. However, the absence of these classical signs does not rule out the condition.

In such cases, biochemical markers play a crucial role in diagnosis. A significantly increased 24-hour urinary copper excretion remains a key indicator of Wilson's disease, helping to confirm copper accumulation in the body. Other supportive findings include low serum ceruloplasmin levels and elevated hepatic copper content on liver biopsy.

Early identification and timely initiation of copper-chelating agents, such as penicillamine or trientine, are essential in preventing disease progression. Treatment not only helps in reducing copper overload but also minimizes the risk of complications, such as cirrhosis and hepatic failure. Additionally, zinc supplementation can be used as an adjunct therapy to inhibit intestinal copper absorption. Regular monitoring of liver function tests and copper levels is necessary to assess treatment response and ensure long-term disease control.

Since Wilson's disease is a lifelong condition, adherence to therapy and regular follow-ups are crucial in preventing irreversible organ damage. With early diagnosis and appropriate management, patients can achieve significant clinical improvement and lead a normal life.

Conclusion

This case highlights the importance of considering Wilson's disease in non-alcoholic young adults presenting with CLD, even in the absence of neurological symptoms or KF rings. Early diagnosis and timely treatment with copper chelators significantly enhance patient outcomes and prevent irreversible complications.

Acknowledgments

We extend our gratitude to the Department of Internal Medicine, Yenepoya Medical College , for their support in managing this case.

Conflict of Interest

None declared.

References

1. Roberts EA, Schilsky ML. Diagnosis and treatment of Wilson disease: an update. Hepatology. 2008;47(6):2089-2111.
2. European Association for the Study of the Liver (EASL). Clinical Practice Guidelines: Wilson's disease. J Hepatol. 2012;56(3):671-685.
3. Ala A, Walker AP, Ashkan K, Dooley JS, Schilsky ML. Wilson's disease. Lancet. 2007;369(9559):397-408.