

A rare case of Wilson's Disease coinciding with Autoimmune Hepatitis: A Case Report

Colton F. Jones MD¹; Karecia Byfield MD²; Raymond Smith MD²; Jose Manuel Martinez Manzano MD²
Department of Internal Medicine, Albert Einstein Medical Center¹

Introduction

- Wilson's disease (WD) is rare autosomal recessive disorder that leads to impaired biliary copper excretion and the accumulation of copper in multiple organs, including the brain, cornea, and liver.
- It manifests with a wide variability of hepatic and neuropsychiatric signs and symptoms.
- Early diagnosis is of paramount importance because chelation therapy can protect against fibrosis and improve patient prognosis.

Case Presentation

- A 49-year-old female with a medical history of alcohol use disorder presented to the hospital with altered mental status.
- Of note, she was admitted to the Hospital three years ago with elevated liver enzymes and was diagnosed with autoimmune hepatitis (AIH) in the setting of elevated liver enzymes, positive anti-smooth muscle antibody, and consistent biopsy findings.
- She responded favorably to corticosteroid therapy and was discharged but was lost to follow-up.
- On this presentation, her vital signs were within normal limits. She had scleral icterus, a fluid thrill, and asterixis.
- Laboratory studies showed pancytopenia with a white blood cell count of 3.7 per mm³, hemoglobin of 8.5 g/dl, and platelet count of 84,000 per mm³.

Case Presentation (continuation)

- Liver enzymes showed elevated aspartate aminotransferase (152 IU/L; normal 5-34 IU/L), alanine aminotransferase (60 U/L; normal 0-55 IU/L), elevated total bilirubin of 10.1 mg/dl (normal 0.2-1.2mg/dl) with direct bilirubin of 6.5 mg/dl, and normal alkaline phosphatase.
- Coagulation studies were suggestive of coagulopathy with Coombs negative hemolytic anemia.
- Autoimmune studies were positive for anti-smooth muscle antibody again. A comprehensive toxicology workup was unremarkable.
- Computed tomography (CT) of the head was unrevealing. An abdominal CT showed hepatic steatosis, ascites, and multiple portosystemic shunts.
- Paracentesis revealed spontaneous bacterial peritonitis. She was admitted to the Hospital and was treated with ceftriaxone, lactulose and rifaximin around the clock. Her mental status did not improve. Further investigations showed ceruloplasmin of 14mg/L (normal 20-60mg/dl), serum copper of 41mcg/dl (normal 70-175mcg/dl).
- Ophthalmology evaluation showed Kayser Fleisher rings on slit lamp examination, and a repeated liver biopsy showed histological findings consistent with WD.
- She was treated with Zinc sulfate 220mg every 8 hourly and had clinical improvement within a week. She was discharged home in stable condition.

Discussion

- We present a case of WD with superimposed AIH that responded to copper chelation therapy.
- In review of the literature, a limited number of reports have described overlapping features of WD and AIH.
- The role of autoantibodies in WD is still not fully understood; it may represent an early pathological mechanism, or an incidental association between the two entities.

Conclusion

- This case illustrates the importance of avoiding premature closure and to consider WD as a potential cause of acute liver failure, even in patients with a known history of AIH.