

## Case Report

# Wilson's Disease and Autoimmune Hepatitis Coexistence: A Cause of Diagnostic Delay

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## Abstract

This case report describes a 27-year-old housewife who presented with a progressive and insidious onset of abdominal distension, loss of appetite, and fatigue which started 3 months before her presentation. The physical assessment showed moderate ascites and small liver size, and no other peripheral evidence of chronic liver disease was observed. A diagnosis of liver cirrhosis was confirmed by investigations and imaging, where ascites responded well to therapy with diuretics. Investigations for the cause of cirrhosis was established through the diagnosis of autoimmune hepatitis, and she was started on steroids and azathioprine with partial improvement in liver biochemistry.

She presented 8 months later with neuropsychiatric symptoms in the form of slurred speech and difficulty walking. Accordingly, Wilson's disease was suspected to be the cause and further investigations confirmed this. A chelating agent, D penicillamine, was added to her immunosuppressive treatment. Nine months later she showed slow improvement in her neurological symptoms and was referred for assessment for liver transplantation.

Wilson's disease, although rare, should be suspected in patients with decompensated liver disease. The dominance of clinical and epidemiological features of autoimmune hepatitis as a common causative pathology for chronic liver disease in young and middle-aged ladies may hide the presence of other serious different pathologies such as Wilson's disease.

**Keywords:** autoimmune, hepatitis, Wilsons, cirrhosis

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Received 19 October 2022

Accepted 15 July 2023

Published 27 September 2023

Production and Hosting by  
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Editor-in-Chief:

Prof. Nazik Elmalaika Obaid  
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## 1. Introduction

Autoimmune hepatitis (AIH) and Wilson's disease (WD) are recognized as the worldwide causes of acute and chronic liver diseases with possible devastating outcomes if left untreated. Early recognition and intervention for any or both are paramount to improve patient outcomes and survival. The rare coexistence of autoimmune hepatitis and Wilson's disease might cause a delay in detecting each other if a diagnosis of one

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disease was established earlier[1]. In contrast, they may present simultaneously or as Wilson's disease with superimposed autoimmune hepatitis [2, 3].

Uncontrolled liver damage due to the presence of hidden Wilson's disease in a background of autoimmune hepatitis, and vice versa, has poor long-term results including death. This case report reflects the importance of early recognition of Wilson's disease in a patient diagnosed with autoimmune hepatitis.

## 2. Case presentation:

A 27-year-old housewife came to our hospital suffering from abdominal distension, loss of appetite, and fatigue. She is a mother of 3 kids and previously had a free medical background. Her abdominal distension was gradual over a period of 3 months and was not associated with pain or vomiting and with no change in her bowel habit. There was moderate and unintentional loss of weight that was preceded by a period of appetite loss. The patient also noticed mild lower limbs swelling but had no jaundice, changes in mentality, bleeding tendency or easy bruising. A systemic review of other systems revealed no abnormalities. Her younger sister died 10 years ago with a similar presentation of abdominal distension, but this was associated with jaundice. She was treated traditionally, unfortunately she died before reaching a diagnosis. Our patient had no previous history of surgery, blood transfusion or hospital admission, preexisting administration of drugs or herbal medicines or alcohol consumption.

On initial examination, she was found to be pale, not jaundiced with normal vital signs. She had no peripheral evidence of chronic liver disease or encephalopathy. Abdominal examination revealed grade 2 ascites, normal liver span, and impalpable spleen, and she had bilateral pitting lower limbs edema.

A diagnosis of decompensated liver disease due to liver cirrhosis was established by high-quality ultrasonography scan. Further investigations for the cause of liver cirrhosis revealed low hemoglobin at 8g/dl and low platelet count at 93,000. She has normal renal function and mild elevation in the liver enzyme AST at 48IU/L (Ref.range 0-35 IU/L). She had a negative screening for hepatitis B and C viral infections, high titers of anti-smooth muscle antibodies at 1/160 (Ref.value 1/20) and elevated IgG level at 2185mg/dl (Ref.value < 1600) suggesting autoimmune hepatitis (Table 3). She underwent upper gastrointestinal endoscopy which reported grade 1 esophageal varices.

Based on the blood tests, she was diagnosed with autoimmune liver cirrhosis with portal hypertension. Accordingly, she was given Immunosuppressive therapy with prednisolone 30mg/day together with azathioprine 50mg bid, diuretic therapy in the form of

spironolactone 100mg once a day, and non-selective beta blocker propranolol 40mg bid. After one month her ascites were not detected clinically and peripheral edema had subsided. Her liver enzymes settled within the reference ranges (ALT ASTdropped to 22 IU/L from 48 IU/L), and accordingly her steroids dose was tapered to 15 mg/day in addition to a single daily dose of 50 mg azathioprine.

Unfortunately, 8 months later the patient was brought back to the hospital suffering from unsteadiness which made her unable to walk without support, difficulties with her sleep, slurring of her speech, auditory hallucination, and loss of her social character tending with a preference to stay alone and silent. On the basis of her well-established chronic liver disease and new-onset neurological manifestations, a laboratory workup for Wilson's disease was performed which revealed low caeruloplasmin at 6.1mg/dl ( Ref.range 16-50 mg/dl), high 24-hour urinary copper excretion at 217 micg (Ref.range 70-130) and positive Kayser-Fleisher rings in slit lamp ophthalmological examination (Table 4).

She was started on chelating therapy with D penicillamine 500 mg twice a day in addition to her previous immunosuppressive medications.

Two months after starting the combined therapy, the patient started to show reported clinical improvement with the ability to walk without support, as well as improvement in communication, socialization, and sleep patterns.

### 3. Discussion

Autoimmune hepatitis is a common cause of liver injury that can affect all age groups of all ethnicities with more predominance among young and middle-aged women [4]. It can present as a lonesome pathology, but in many circumstances, it can occur in combination with other liver diseases leading to more accelerated hepatocytes damage. Overlap syndrome is a well-described clinical syndrome in which autoimmune hepatitis and primary biliary cirrhosis or autoimmune hepatitis and primary sclerosing cholangitis may occur simultaneously. In this situation, the patient presents with both hepatitic and cholestatic pictures in the liver function test. [5] Another example is the simultaneous presentation of autoimmune hepatitis and non-alcoholic fatty liver disease [6]. Prolonged use of steroid therapy to control hepatocytes damage in autoimmune hepatitis can result in peripheral insulin resistance, obesity, and fatty liver [1]. So, there are several situations where the coexistence of two hepatic diseases in the same patient at the same time have been reported in the literature. Poor handling in the literature of the coexistence of autoimmune hepatitis and Wilson's disease as possible concomitant

causative pathologies of chronic liver disease makes many clinicians underestimate this dual presentation. In addition, similar manifestations of both diseases may cause some dilemmas in establishing two separate diagnoses in the same patient [7]. Diagnosis delay of Wilson's disease which occurs in a background of autoimmune hepatitis has a bad outcome and might end with liver transplantation as a sole treatment option [3]. Some patients with autoimmune hepatitis may show sluggish response to immunosuppressive therapy with minimal or no improvement in liver biochemistry [8]. Such a situation should encourage physicians to search for alternative pathologies, like Wilson's disease, as a less common alternative cause of liver injury. On the other side, and as in our case, immunosuppressive medications for autoimmune hepatitis can give good results in overall clinical improvement and biochemical tests. This improvement caused some delay in requesting tests to diagnose this masqueraded Wilson's disease, until it proclaimed itself with advanced liver disease and finally neuropsychiatric manifestation due to central nervous system involvement. Using the revised scoring system of the International Autoimmune Hepatitis Group, our case met the criteria for a probable diagnosis of autoimmune hepatitis during the pre-treatment period with a total score of 14. In reported cases, Wilson's disease can often mimic autoimmune hepatitis in presentation and histology. Wilson's disease presenting as autoimmune hepatitis may occur with greatly elevated autoantibodies like ASMA and ANA. High levels of IgG are also reported. [9] As in our case this could be the situation from the start, but taking the possibility of autoimmune hepatitis as a commoner cause of chronic liver disease in this young lady, together with the presence of positive titers of autoantibodies, may have influenced our clinical judgment.

#### 4. Conclusion

However, early diagnosis and treatment can improve the outcome of Wilson's disease patients and reduce disability and early death [10]. Clinicians should always put in mind that Wilson's disease is prone to long-term misdiagnosis or unclear diagnosis [11], especially if it presents timidly with other liver diseases. The presence of a clinical picture resembling autoimmune hepatitis, especially with mild elevation in autoantibodies and without histological confirmation of the diagnosis, does not exclude Wilson's disease as a causative pathology of that picture. The most important determinant of the prognosis of Wilson's disease that presents alone or simultaneously with other pathology is the early recognition and diagnosis.

## Acknowledgments

## Ethical Approval

The case was approved from the Ethical Committee at Ibn Sina Research Unit, Ibn Sina Hospital, and the patient signed informed written consent for publishing her case.

## Conflict of Interests

All authors certify that there is no conflict of interest with any financial organization regarding the material discussed in this case report manuscript.

## Availability of Data and Material

## Funding

None.

## Author Contributions

MGM; concept, design, literature search, manuscript preparation, manuscript editing, manuscript draft writing, and proofreading. SBM; copy, editing, revision, and guarantor. EMA; drafting and revision. All authors carried out the entire case report.

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