

Autoimmune Hepatitis Disguised As Hemochromatosis

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Introduction

- Autoimmune hepatitis (AIH) is a diagnosis of exclusion and generally involves the presence of autoantibodies and elevated serum globulins. Absence of autoantibodies in some cases makes the diagnosis more challenging. We present a case of seronegative AIH with findings suggestive of hemochromatosis (HC) that proved misleading.

Case Presentation

- A 65-year-old female with history of SLE and hypothyroidism presented with RUQ abdominal pain and jaundice. No history of alcohol or herbal supplement use. Exam was unremarkable except for jaundice. Labs were significant for elevated liver enzymes, iron, ferritin and transferrin saturation (**Table 1**), but negative for acute viral hepatitis, ANA, ASMA, AMA, A1AT, ceruloplasmin, elevated IgG, and mutations in HFE genes. Liver biopsy was consistent with severe, active AIH (**Fig. 1**).
- The patient was diagnosed with seronegative AIH that responded appropriately to steroids with clinical and laboratory recovery (**Table 1**).

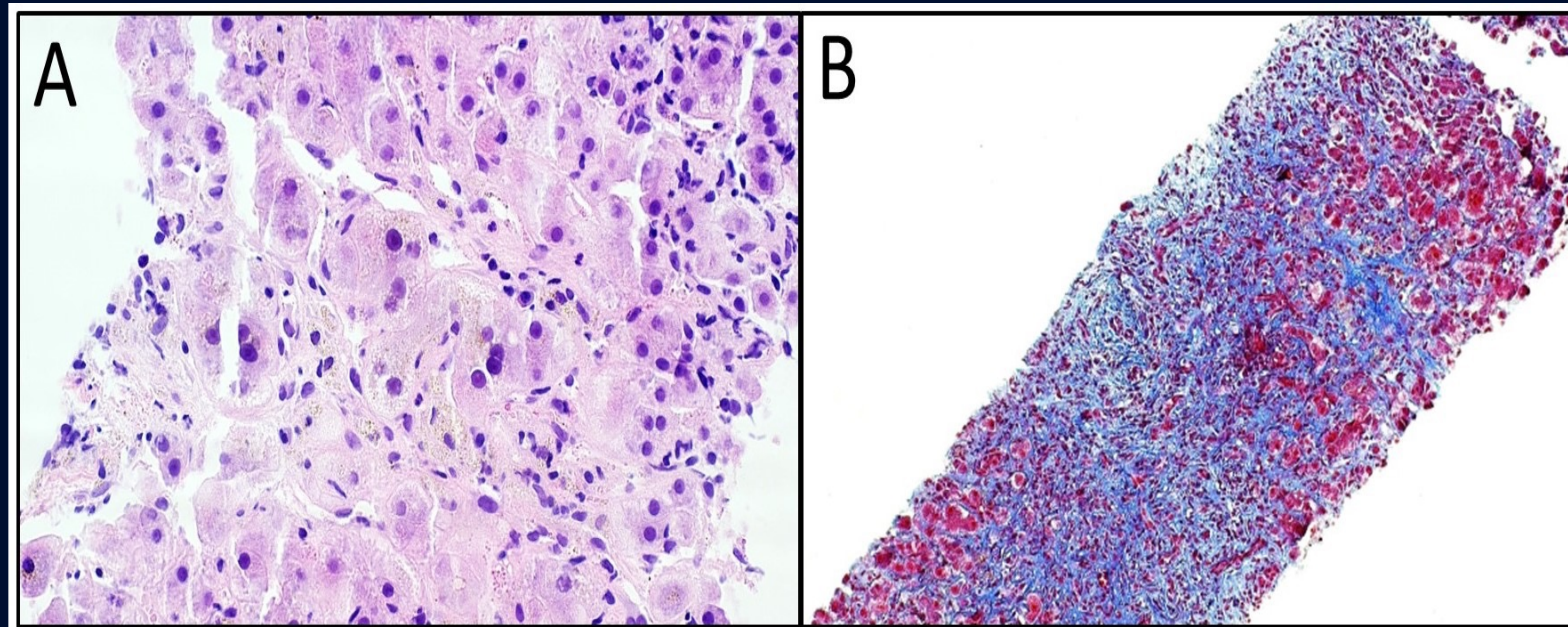


Fig 1A: Severe interface hepatitis with marked inflammation (acute and chronic).

Fig 1B: Trichrome stain showing extensive chicken-wire fibrosis consistent with cirrhosis.

LABS	INITIAL PRESENTATION	1.5 MONTHS ON TREATMENT
AST (U/L)	696	39
ALT (U/L)	654	41
ALP (U/L)	258	109
Total Bilirubin (mg/dL)	8.2	1.0
Iron (mcg/dL)	219	122
Transferrin Saturation (%)	88	36
Ferritin (mcg/dL)	3,117	339

Table 1: Trend of select liver function tests and iron studies from initial presentation to 1.5 months on immunosuppressant therapy.

Discussion

- Our patient is a female with history of autoimmune diseases who presented with acute liver injury that was initially concerning for HC given the absence of autoantibodies and presence of elevated serum iron, ferritin and transferrin saturation. However, negative HFE mutations and **liver biopsy** ruled out HC and confirmed AIH.
- She scored 4 points on Simplified Diagnostic Criteria for AIH (SDC), indicating possible AIH and scored **18 points** on Revised Diagnostic Criteria (RDC), indicating **definite AIH**.
- Seronegative AIH** is seen in ~10% of AIH cases and is associated with a **more aggressive course** and can rarely be associated with elevated transferrin saturation. In these cases, **supplemental use of RDC and liver biopsy** can be helpful in avoiding delay in diagnosing this potentially aggressive disease.

References

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