Case Report

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A Rare Case of Autoimmune Hepatitis-Small Duct Primary Sclerosing Cholangitis Overlap Syndrome Related Chronic Liver Disease

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Abstract

Background: Autoimmune hepatitis-primary sclerosing cholangitis (AIH-PSC) overlap syndrome is a rare condition combining features of AIH and PSC, often leading to chronic liver disease and portal hypertension. It presents diagnostic challenges due to its atypical manifestations and requires a combination of serological, histological, and imaging findings for accurate diagnosis.

Case Report: A 60-year-old male presented with recurrent hematemesis and melena for one year, without jaundice or abdominal pain. Examination revealed mild anemia, pruritus-related scratch marks and splenomegaly. Laboratory findings included microcytic hypochromic anemia, thrombocytopenia, elevated alkaline phosphatase, positive smooth muscle antibody (>1:80), and raised IgG. Upper GI endoscopy showed grade II esophageal varices with congestive gastropathy. MRI/MRCP indicated chronic liver disease with splenomegaly and gastroesophageal varices, but a normal extra and intrahepatic biliary tree. Liver biopsy shows features of cirrhosis along with moderate piecemeal necrosis and periductular onion skinning. The patient was diagnosed with AIH-small duct PSC overlap syndrome and treated with ursodeoxycholic acid, prednisone, azathioprine, propranolol and endoscopic variceal ligation.

Conclusion: This case underscores the importance of considering AIH-PSC overlap syndrome in atypical chronic liver disease presentations. Timely diagnosis and tailored treatment led to clinical improvement and no further bleeding on follow-up.

Keywords: Autoimmune hepatitis, Primary sclerosing cholangitis, Small duct primary sclerosing cholangitis, Overlap syndrome, Portal hypertension, Esophageal varices.

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Introduction:

Autoimmune hepatitis (AIH) and primary sclerosing cholangitis (PSC) are distinct autoimmune liver diseases, but their coexistence as an overlap syndrome has been recognized in 7-14% of cases, particularly in adults, over the past few decades[1,2].

Historically, the understanding of AIH-PSC overlap has evolved from early reports of atypical liver disease presentations to a well-defined syndrome characterized by AIH features, such as interface hepatitis, positive autoantibodies (e.g., antinuclear antibodies [ANA], anti-smooth muscle antibodies [ASMA]), and elevated immunoglobulin G (IgG), alongside cholangiographic or histologic evidence of PSC, confirmed by magnetic resonance cholangiopancreatography (MRCP) or biopsy, in the absence of antimitochondrial antibodies (AMA).[3,4] This condition often progresses to chronic liver disease (CLD) with portal hypertension, leading to complications like variceal bleeding.[5,6] Recurrent upper gastrointestinal (GI) bleeding without overt jaundice is a rare presentation, complicating diagnosis.[7,8] We present a case of AIH-PSC overlap syndrome in an elderly male, diagnosed through clinical, serological, imaging, and histological findings and managed successfully with immunosuppressive and supportive therapy.[9]

Case Presentation:

A 60-year-old male, a painter from Savar, Dhaka, Bangladesh, presented on September 23, 2022, with recurrent episodes of hematemesis (both fresh and altered blood) and melena for one year. Symptoms were not associated with jaundice, abdominal pain, distension, altered consciousness, fever, cough, night sweats, or bleeding from other sites. He reported anorexia, 4 kg weight loss, and occasional generalized pruritus, predominantly in the limbs, without skin lesions. He had been admitted to a local clinic three times and received conservative management, including 6 units of whole blood transfusion over the year. There was no history of joint pain, oral ulcers, skin rash, eye redness, pale stools, dark urine, palpitations, shortness of breath, abnormal movements, or behavior. He was non-diabetic, normotensive, a non-smoker, non-alcoholic, with no history of intravenous drug use, sexual promiscuity, previous jaundice, tuberculosis, surgery, or intake of NSAIDs, antiplatelets, or anticoagulants. Family history was negative for similar illnesses or GI/hepatobiliary malignancies. On examination, he appeared ill-looking and anxious, with a BMI of 19 kg/m² (weight 50 kg). He was mildly anemic, with few scratch marks on limbs, but no jaundice, edema, dehydration, lymphadenopathy, clubbing, koilonychia, leukonychia, cyanosis or other stigmata of CLD. Vitals were stable: temperature 98.4°F, pulse 88 bpm, BP 110/60 mmHg, respiratory rate 18/min. Abdominal examination showed normal shape, centrally placed inverted umbilicus and no visible veins or peristalsis. Palpation revealed no

tenderness or mass; the liver was not palpable, but the spleen was enlarged (4 cm from the left costal margin, firm, smooth). No ascites (shifting dullness absent), bowel sounds present, digital rectal exam normal. Other systems (nervous, respiratory, cardiovascular) were unremarkable. The clinical diagnosis was recurrent upper GI bleeding due to CLD with portal hypertension. Investigations revealed hemoglobin 9.2 g/dL (previously 6.0 g/dL), ESR 3 mm/1st hr, WBC 5.7 x 10^3/µL, RBC 3.47 x 10^6/µL, platelets 160 x 10^3/µL, peripheral blood film showing microcytic hypochromic anemia with thrombocytopenia. Liver function tests: bilirubin 1.7 mg/dL, ALT 19 U/L, ALP 194 U/L (elevated), GGT 109 U/L (elevated), albumin 3.2 g/dL, PT 16 sec (control 12 sec). Ultrasound abdomen: coarse liver parenchyma, enlarged spleen (13 cm), impression of CLD. Upper GI endoscopy: grade II esophageal varices with congestive gastropathy; endoscopic variceal ligation (EVL, 1st session) performed. Viral markers (HBsAg, anti-HCV, anti-HBc) negative. Autoantibodies: SMA positive (>1:80), IgG 23.0 g/L (elevated), ANA negative, anti-LKM-1 5.0 U/mL, AMA 0.74 U/mL (negative), pANCA 0.5 U/mL (negative). As the patient had persistent cholestatic biochemical findings, an MRI/MRCP was done. It revealed features of CLD with splenomegaly, gastro-oesophageal varices, but there were no features of extra or intrahepatic biliary obstruction. Other tests (serum ceruloplasmin, 24-hr urinary Cu, slit-lamp for KF ring, ferritin, lipid profile, TSH, FT4, CA19-9) were normal or non-contributory. Fibroscan: 13.2 kPa (F3), CAP 230 dB/m (S0). Liver biopsy: core of liver tissue: hepatocytes in nodules separated by fibrous bands with lymphocyte infiltrates and many bile ductules, periductular onion skinning, swollen hepatocytes and moderate piecemeal necrosis. The final diagnosis was CLD with portal hypertension, grade II esophageal varices and congestive gastropathy due to AIH-small duct PSC overlap syndrome, based on simplified AIH criteria (score ≥7: SMA ≥1:40 +2, IgG >ULN +1, typical histology +2, negative viral markers +2) and PSC features on biopsy. Colonoscopy was done to screen for ulcerative colitis (UC), which revealed normal findings. Treatment included ursodeoxycholic acid (UDCA), prednisolone + azathioprine, propranolol, calcium + vitamin D, EVL and vaccinations against hepatitis A and B. Follow-up at 3 months (March 2023): no bleeding, improved appetite, weight 51 kg, no pruritus, Hb 10 g/dL, platelets 134 x 10^3/µL, ALT 48 U/L, ALP 160 U/L, normal IgG; endoscopy showed interrupted grade I varices with gastropathy. At 6 months (June 2023): weight 53 kg, Hb 9.9 g/dL, platelets 130 x 10^3/µL, ALT 40 U/L, ALP 160 U/L; endoscopy showed grade I varices.

Discussion:

AIH-PSC overlap syndrome, a rare condition blending autoimmune hepatitis (AIH) features like elevated transaminases and positive autoantibodies with primary sclerosing cholangitis (PSC) characteristics such as biliary strictures on imaging, presents diagnostic and therapeutic challenges. This case of a patient with recurrent variceal bleeding as the dominant symptom, absent jaundice or typical cholestatic signs and normal extra and intrahepatic biliary tree on MRCP, absence of UC deviates from the norm, highlighting the spectrum's variability. Small duct PSC is not common as well and needs a high index of suspicion in case of cholestatic features with normal MRCP or ERCP, where liver biopsy is recommeneded.[6] Diagnostic confirmation relied on positive smooth muscle antibodies, elevated IgG, negative viral and antimitochondrial antibodies, and a liver biopsy revealing biliary cirrhosis, periductular onion skinning with piecemeal necrosis, aligning with established criteria. Treatment with prednisolone, azathioprine and UDCA led to symptom resolution and biochemical improvement, consistent with guidelines advocating immunosuppressive therapy to reduce transplant need in overlap cases. Comparative insights from a similar case with hypereosinophilia by Hatami in 2021 showed a rapid response to the same regimen but with jaundice as the key feature, suggesting immune-mediated inflammation may vary in presentation.[10] Another case linked to ulcerative colitis by Ballotin underscored IBD's role in chronic progression and higher transplant risk, contrasting with this patient's isolated hepatic decompensation.[11] The rapid resolution here, without IBD or eosinophilic involvement, supports the efficacy of early immunosuppression in overlap variants. This underscores the importance of a comprehensive evaluation of the etiology of CLD, including biopsy, to tailor therapy and monitor for occult associations. The diversity in clinical onset-bleeding here versus jaundice or IBD-related symptoms elsewhere-emphasizes the need for individualized management, potentially improving outcomes by addressing autoimmunity, cholestasis and portal hypertension early in atypical presentations of this complex syndrome.

Conclusion:

AIH-PSC overlap syndrome is an underestimated cause of chronic liver disease. Early diagnosis via serology, imaging and biopsy, followed by combined immunosuppressive and choleretic therapy can halt progression and prevent complications.

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