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BUDD-CHIARI SYNDROME AS THE PRESENTING FEATURE OF SYSTEMIC IMMUNOGLOBULIN LIGHT CHAIN AMYLOIDOSIS

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ABSTRACT

Budd-Chiari syndrome (BCS), characterised by hepatic venous outflow obstruction, is frequently associated with hypercoagulable states. Systemic immunoglobulin light chain (AL) amyloidosis, on the other hand, commonly presents with bleeding complications. The association between the two diseases is not common, but some cases have been described in the past. We report a case of a 58-year-old male who presented with lower limb oedema, abdominal distention and severe fatigue, ultimately diagnosed with BCS secondary to systemic AL amyloidosis. This case highlights the rare association between AL amyloidosis and BCS, likely precipitated by a nephrotic syndrome-induced hypercoagulable state, and emphasises the need for clinicians to consider systemic amyloidosis when investigating the aetiology of BCS, even in the absence of typical bleeding manifestations.

KEYWORDS

Budd-Chiari syndrome, AL amyloidosis, nephrotic syndrome, liver failure

LEARNING POINTS

- AL amyloidosis is typically associated with bleeding disorders, but rarely can present with thrombotic complications as well, specifically when associated with nephrotic syndrome.
- Budd-Chiari syndrome is a life-threatening condition which should be on the differential diagnosis of subacute liver failure.
- A thorough aetiological investigation is essential in a patient with Budd-Chiari syndrome, including consideration for potential causes of nephrotic syndrome and other less typical disease associations.

INTRODUCTION

Budd-Chiari syndrome (BCS) represents an obstruction of the venous flow from the hepatic venous system, usually due to thrombotic events. The obstruction can occur all the way to the right atrium (with the supra-hepatic veins being the most common location), and most patients are found to have an underlying hypercoagulable state[1]. Immunoglobulin light chain (AL) amyloidosis is the most common type of systemic amyloidosis, and is characterised by deposition of unfolded monoclonal proteins in various organs; it is usually

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associated with coagulation problems – mainly bleeding abnormalities, but thrombotic complications may also occur rarely[2]. Here, we present a case of a patient with systemic AL amyloidosis with BCS as the main feature.

CASE DESCRIPTION

A 58-year-old male patient presented to the emergency department in July 2023 with lower limb bilateral oedemas, abdominal distention and severe fatigue. His medical history was notable for dyslipidaemia and a prosthetic knee replacement after a traumatic injury in his early adulthood. He had recently had a pelvic ultrasound as prescribed by his general practitioner, which showed bilateral inguinal adenopathies. The patient reported a steady weight loss in the previous three months and bloating, but no fever, loss of appetite or other constitutional symptoms. On physical examination, the patient was borderline hypotensive with normal pulse, was eupnoeic and neurologically intact. He had abdominal enlargement with a positive wave sign, with no jaundice of the skin or sclera, and on auscultation his breathing sounds were significantly diminished in both lung bases.

His laboratory results showed increased haemoglobin (19.3 g/dl; normal 13–17 g/dl) and normal platelet and leukocyte counts, slightly decreased sodium (128 mmol/l; normal 135–145 mmol/l) and severely decreased albumin (1.8 g/dl; normal > 3.5 g/dl), with normal renal function. Hepatic parameters were also abnormal, with increased aspartate aminotransferase and alanine aminotransferase (143 U/l and 97 U/l, respectively; both normal at < 40 U/l) and significantly increased alkaline phosphatase (942 U/l; normal < 125 U/l).

Given the analytical and clinical findings, the patient underwent a computed tomography of the abdomen which showed diffuse heterogeneity of the liver parenchyma with no contrast enhancement of the supra-hepatic veins, which altogether were highly suggestive of BCS *(Fig. 1)*. It also showed moderate ascites, a normal sized spleen and some inguinal adenopathies. A diagnostic paracentesis was performed, and the serum-ascites albumin gradient was compatible with a portal hypertension aetiology for the ascites.

The patient was initiated on systemic anticoagulation with low-molecular-weight heparin and was admitted to the internal medicine ward for further investigation. On the next day, a 24-hour urine collection was consistent with nephrotic proteinuria (> 10g/day) and the patient had elevated serum triglycerides, which altogether was highly suggestive of a nephrotic syndrome.

Because of the persistent low blood pressures, a transthoracic echocardiogram was also performed, which showed left ventricular hypertrophy with a diminished longitudinal strain and an 'apical sparing' pattern, frequently seen in cardiac amyloidosis. At that time, the serum light chains



Figure 1. Contrast enhanced computed tomography of the abdomen showing diffuse heterogeneity of the liver parenchyma (nutmeg liver) and no contrast enhancement of supra-hepatic veins, suggestive of Budd-Chiari syndrome.

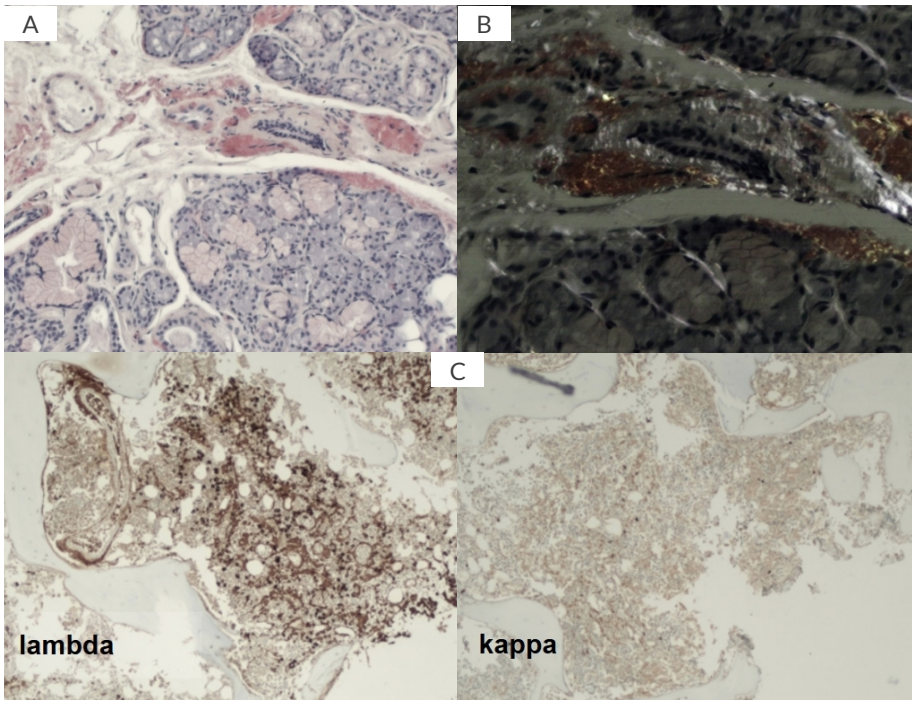


Figure 2. Histopathology images from salivary glands (A and B) and bone marrow (C). A) Salivary gland stained with Congo Red, showing acini and ducts with fibrosis stained in red, suggestive of amyloid deposits; B) Green birefringence confirms the presence of amyloid deposits; C) Bone marrow immunohistochemistry shows an elevated number of plasma cells (stained brown) with evident lambda light chain restriction.

demonstrated a very low kappa/lambda ratio, and serum and urinary immunofixation were both positive for a lambda light chain monoclonal gammopathy – for that reason, both salivary glands and bone marrow were biopsied. Histology of the salivary glands displayed perivascular deposition of an amorphous substance staining positive for Congo red, suggestive of amyloid fibrils. Bone marrow histology showed infiltration of plasmocyte neoplastic cells (corresponding to 20% of the total cellularity) and amyloid deposits *(Fig. 2)*. These findings were all consistent with a light chain multiple myeloma with multisystemic AL amyloidosis, presenting with BCS.

The patient's liver function kept worsening after the start of low-molecular-weight heparin, and peripheral oedemas and ascites did not improve with parenteral diuretics. A transjugular intrahepatic portosystemic shunt was considered but the risks were considered to outweigh the benefits as the patient kept worsening, with acute liver failure developing at day 12 of admission. Because of the subsequent fulminant multiorgan failure, the patient was deemed not eligible for systemic treatment of the AL amyloidosis and died on day 17.

DISCUSSION

Most patients diagnosed with BCS have an underlying condition predisposing to hypercoagulation, such as myeloproliferative neoplasms. On the other hand, amyloidosis is usually associated with bleeding complications – the presented case is an example where the nephrotic syndrome, caused by renal involvement of AL amyloidosis, induced a hypercoagulable state leading to the thrombosis of the supra-hepatic veins, with a fulminant evolution to liver failure and multiorgan dysfunction. There have also been reports of BCS occurring in systemic amyloidosis without concurrent nephrotic syndrome, but its pathophysiology remains unclear[3]. Because of its high rates of complications and high morbidity, a thorough aetiologic investigation should be pursued to identify the underlying cause of thrombosis in Budd-Chiari syndrome patients, allowing the appropriate treatment. Since all the signs and symptoms are highly unspecific, the clinician should be aware of this association to allow an early diagnosis and treatment of the condition.

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