Case Report

Unusual presentation of antiphospholipid antibody syndrome in a young male: Diagnostic and therapeutic challenge

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ABSTRACT

The acquired autoimmune thrombophilia known as antiphospholipid syndrome (APS) or antiphospholipid antibody syndrome is defined by arterial or venous thrombosis in the presence of persistent antiphospholipid antibodies. APS in young guys is uncommon and frequently underdiagnosed because of unusual presentation and multisystem involvement, whereas it is more frequently observed in females, particularly accompanying maternal morbidity, acute abdominal distension, jaundice, and black urine. These were the symptoms observed in a 35-year-old male patient who had no past comorbidities. Massive ascites and hepatocellular dysfunction were found upon examination. Investigations revealed conjugated hyperbilirubinemia, significantly increased liver enzymes, positive lupus anticoagulant, and negative anticardiolipin and anti-β2 glycoprotein I antibodies. Imaging showed severe portal vein thrombosis that extended into the splenic and superior mesenteric veins with mesenteric ischemia, as well as Budd-Chiari syndrome with hepatic vein non-opacification. The patient had hepatic vein stenting after receiving therapeutic anticoagulation with low molecular weight heparin right away. He was switched to acenocoumarol for long-term oral anticoagulation.

Key words: Anticoagulation, Antiphospholipid syndrome, Budd-Chiari syndrome, Case report, Portal vein thrombosis, Thrombophilia, Young male

A persistent presence of antiphospholipid antibodies (lupus anticoagulant, anticardiolipin antibodies, and anti-β2 glycoprotein I antibodies) linked to arterial and/or venous thrombosis and pregnancy morbidity characterizes antiphospholipid syndrome (APS), also known as antiphospholipid antibody (APLA) syndrome [1]. Due to its link with autoimmune illnesses and maternal difficulties, APS is traditionally documented more frequently in young to middle-aged females; nevertheless, its incidence in males is becoming increasingly acknowledged but is still underreported. In men, the lack of pregnancy-related symptoms frequently postpones clinical suspicion, and the diagnosis is usually not made until thrombotic problems arise. This increases the chance of catastrophic or recurrent thrombosis and delays diagnosis [2]. The pathophysiology of APLA syndrome is complex and involves antibody-mediated activation of endothelial cells, monocytes, and platelets, leading to a hypercoagulable state. These antibodies also interfere with natural anticoagulant pathways and promote complement activation, resulting in vascular inflammation and thrombosis. The clinical spectrum is highly heterogeneous, ranging from deep vein thrombosis and pulmonary embolism to arterial thrombosis, such as stroke, myocardial infarction, and peripheral arterial occlusion. In rare cases, microvascular thrombosis may lead to multi-organ dysfunction, commonly seen in catastrophic APLA syndrome, which carries a high mortality rate [3].

Diagnosing young male patients with unusual presentations is quite difficult. Young boys may appear with isolated or atypical thrombotic episodes without obvious risk factors, in contrast to typical situations when a clear autoimmune background or obstetric history may arouse suspicion. These could include myocardial infarction at a young age without conventional cardiovascular risk factors, renal vein thrombosis, cerebral venous sinus thrombosis, or portal vein thrombosis (PVT) [4]. The Sydney classification criteria, which call for at least one clinical event (vascular thrombosis or pregnancy morbidity) and continuous laboratory positivity of antiphospholipid antibodies on two or more occasions spaced at least 12 weeks apart, have updated the diagnostic criteria for APLA syndrome [1].

However, temporary antibody positivity during infections, inconsistency in laboratory testing, and the lack of initial clinical suspicion in atypical populations like young boys continue to make early diagnosis

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difficult in clinical practice. Recurrent thrombotic events are more likely as a result of the delayed beginning of suitable long-term anticoagulation medication. From a therapeutic perspective, APLA syndrome management primarily involves long-term anticoagulation, most commonly with Vitamin K antagonists, such as warfarin. The intensity and duration of anticoagulation depend on the type and severity of thrombotic events. In arterial thrombosis or recurrent venous thrombosis, high-intensity anticoagulation is often required. However, management becomes particularly challenging in young patients with atypical presentations, as balancing bleeding risk with prevention of recurrence is complex [5].

This case is unusual because the reported clinical presentation, diagnosis, and management are rarely encountered in routine practice. The condition occurred in an uncommon setting and demonstrated distinctive clinical features that posed diagnostic and therapeutic challenges. Furthermore, successful management and favorable outcomes provide valuable insights for clinicians and contribute to the limited existing literature on this rare presentation. Reporting this case may help increase awareness, facilitate early recognition, and guide appropriate management in similar situations.

CASE PRESENTATION

An 8-day history of growing abdominal distension accompanied by scleral icterus, dark-colored urine, and a single episode of high-grade fever was brought to the emergency room by a 35-year-old male patient with no major prior medical history. The patient denied using alcohol, abusing drugs, taking hepatotoxic substances, traveling recently, or experiencing similar incidents in the past. Neither a familial history of thrombophilia nor a history of prior thrombotic episodes was present. The acute and quickly developing clinical appearance raised the possibility of a serious vascular or hepatobiliary pathology.

Upon physical examination, the patient had a temperature of 98.4°F, a blood pressure of 124/78 mmHg, a pulse rate of 88/min, a respiratory rate of 18/min, and was conscious, alert, and hemodynamically stable. He had severe tight ascites and was icteric, making it difficult to palpate his spleen and liver. There were no signs of lymphadenopathy, peripheral edema, or chronic liver disease stigmata. The results of the neurological, pulmonary, and cardiovascular exams were ordinary. The patient denied having signs of neurological impairments, gastrointestinal hemorrhage, heart illness, or previous thrombotic events.

Laboratory tests showed significantly increased transaminases and extensive liver damage, which gradually improved while the patient was in the hospital. Alanine aminotransferase was 5,678 U/L, and aspartate aminotransferase was 3,452 U/L at admission; on day 10, these values had gradually dropped to 452 U/L and 126 U/L, respectively (Table 1). With a direct fraction of 3.2 mg/dL, total bilirubin was increased to 4.8 mg/dL. With a prothrombin time of 18.2 s, International normalized ratio (INR) of 1.6, and activated partial thromboplastin time of 38.4 s, the coagulation profile displayed minor disruption. Hemoglobin was 11.2 g/dL, total leukocytes were 8,900/mm^3, and platelets were 145,000/mm^3, according to a complete blood count. Anticardiolipin and anti-β2 glycoprotein I antibodies were negative, but the thrombophilia workup showed a positive lupus anticoagulant. A hypercoagulable state was indicated by slightly lower levels of protein C (68%), protein S (62%), and antithrombin III (78%) (Table 2).

Chronic liver parenchymal disease with extensive ascites, hepatomegaly, and PVT was discovered by abdominal ultrasonography. The abdomen's contrast-enhanced computed tomography also showed non-opacification of the hepatic veins consistent with Budd-Chiari syndrome, partial thrombosis of the portal vein spreading into the splenic and superior mesenteric veins, and mild hepatomegaly with parenchymal abnormalities. In addition, modest bilateral pleural effusion and gut wall thickening suggestive of mesenteric ischemia were seen. In line with post-sinusoidal portal hypertension, diagnostic paracentesis revealed a significant serum-ascites albumin gradient with higher protein content.

Budd-Chiari syndrome with concomitant PVT related to APS was diagnosed based on clinical symptoms, laboratory data, and imaging features. Clinical and radiological correlation led to the exclusion of cardiac causes, such as right heart failure and constrictive pericarditis (Fig. 1).

The patient received immediate therapeutic anticoagulation with low molecular weight heparin in addition to supportive care and therapeutic paracentesis to relieve tight ascites. Due to significant hepatic venous outflow obstruction, interventional radiology was able to successfully perform hepatic vein stenting (Figs. 2 and 3).

During hospitalization, the patient showed remarkable clinical improvement with significant reduction in ascites, resolution of jaundice, and more than 90% reduction in liver enzyme levels. He remained stable

Table 1: Liver function tests showed severe hepatocellular injury with gradual improvement

Day	Alanine aminotransferase (U/L)	Aspartate aminotransferase (U/L)	Alkaline phosphatase (U/L)	Gamma-Glutamyl transferase (U/L)
Day 1	5,678	3,452	234	187
Day 3	3,245	2,156	198	156
Day 5	1,789	1,234	178	142
Day 7	945	567	165	134
Day 10 (discharge)	452	126	145	98

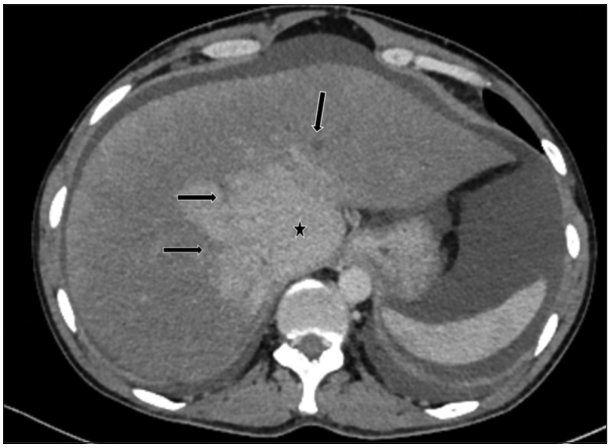

Figure 1: Contrast-enhanced computed tomography abdomen (axial view) showing non-opacification of hepatic veins and hepatomegaly with parenchymal changes consistent with Budd-Chiari syndrome. There is non-opacification of all three hepatic veins (black arrows), indicating hepatic venous outflow obstruction. The caudate lobe and central liver show early enhancement, with delayed enhancement of the peripheral liver, creating a characteristic "flip-flop" appearance (star). The above features are suggestive of Budd-Chiari syndrome

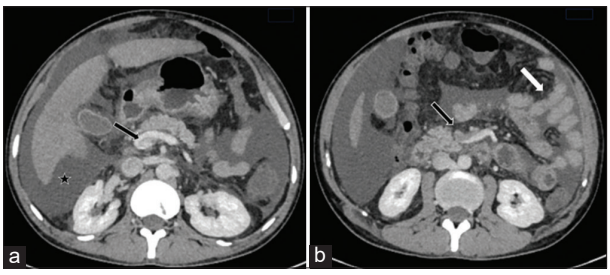

Figure 2: Computed tomography imaging demonstrating portal vein thrombosis with extension to the splenic vein and superior mesenteric vein, along with ascites formation. (a) Demonstrates non-opacification at the confluence of the portal and splenic veins (black arrow), along with gross ascites (star); (b) Shows non-opacification of the superior mesenteric vein (black arrow) and edematous jejunal loops (white arrow), suggestive of mesenteric venous thrombosis with associated bowel wall edema

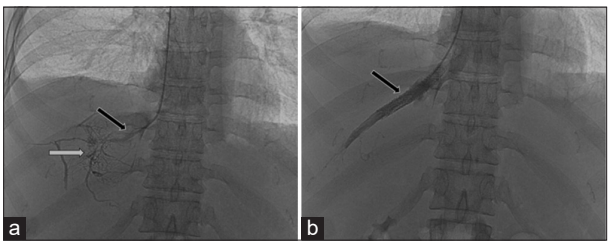

Figure 3: Coronal reconstruction showing the extent of thrombosis involving both portal and hepatic venous systems, with associated mesenteric changes and hepatic congestion. Right hepatic vein (RHV) angioplasty and stenting performed through transjugular approach. (a) Pre-intervention venogram shows non-opacification and narrowing of the RHV (black arrow), with prominent collateral venous channels (white arrow). (b) Post-angioplasty and stenting venogram demonstrates restored patency and normal opacification of the RHV (black arrow), indicating successful revascularization

without any bleeding complications and was discharged after 10 days in improved condition. At discharge, he was advised lifelong anticoagulation therapy with regular INR monitoring and multidisciplinary follow-up with hepatology, hematology, and interventional radiology teams for surveillance of recurrent thrombosis and portal hypertension-related complications.

Table 2: Biochemical, hematological, and thrombophilia profile

Investigation	Result	Reference/ Interpretation
Total bilirubin	4.8 mg/dL	Elevated
Direct bilirubin	3.2 mg/dL	Elevated (conjugated hyperbilirubinemia)
Indirect bilirubin	1.6 mg/dL	Mildly elevated
PT	18.2 s	Prolonged (control 12.5 s)
INR	1.6	Increased
APTT	38.4 s	Mildly prolonged (control 32 s)
Hemoglobin	11.2 g/dL	Mild anemia
Total leukocyte count	8,900/mm^3	Within normal limits
Platelet count	145,000/mm^3	Borderline low-normal

PT: Prothrombin time, INR: International normalized ratio, APTT: Activated partial thromboplastin time

To prevent catastrophic hepatic outcomes, early detection, thorough thrombophilia evaluation, and prompt multidisciplinary management are crucial. This case highlights an unusual and severe presentation of APS in a young male that manifests as simultaneous Budd-Chiari syndrome and PVT.

The patient received rapid therapeutic anticoagulation with low molecular weight heparin in addition to supportive care, which included dietary assistance and fever management. To relieve the symptoms of tight ascites, therapeutic paracentesis was used. Hepatic vein stenting was successfully performed due to a severe obstruction of the hepatic venous outflow.

To stop the thrombus from spreading further, the patient was first treated by starting therapeutic-dose low molecular weight heparin right away. Therapeutic paracentesis was used to relieve the symptoms of large ascites. Hepatic vein stenting was effectively carried out under interventional radiology guidance to restore venous drainage in light of the substantial hepatic venous outflow obstruction caused by Budd-Chiari syndrome. The patient was switched to long-term oral anticoagulation with acenocoumarol after stability.

Throughout their hospital stay, the patient's ascites gradually decreased, their jaundice resolved, and their liver enzyme levels improved by more than 90%. There were no bleeding issues noted. After 10 days, he was released in stable condition and was prescribed lifelong anticoagulation along with routine INR monitoring and multidisciplinary follow-up with interventional radiology, hepatology, and hematology to avoid and monitor recurrent thrombotic episodes.

DISCUSSION

This case illustrates a rare and severe manifestation of APS in a young male. It appears as concurrent PVT

and Budd-Chiari syndrome (hepatic vein thrombosis), resulting in abrupt hepatic congestion and characteristics of portal hypertension. Venous and arterial thrombosis in the context of persistent antiphospholipid antibodies, most frequently lupus anticoagulant, anticardiolipin, or anti-β2 glycoprotein I antibodies, is the hallmark of APS, a systemic autoimmune thrombophilic illness [5].

Acute liver dysfunction with huge ascites, jaundice, and substantial transaminase elevation was the patient's most common clinical presentation. At first, it appeared to be either acute hepatitis or decompensated chronic liver disease. However, imaging tests showed severe PVT that extended into the splenic and superior mesenteric veins in addition to hepatic vein outflow blockage, confirming a combination of Budd-Chiari syndrome and PVT. A significant hypercoagulable state is indicated by this rare dual venous involvement. When other antiphospholipid antibodies are ruled out and a positive lupus anticoagulant is present, it may indicate a seronegative or single-positive APS variation, which is nonetheless linked to a high risk of thrombosis [6].

Endothelial cell activation, elevated tissue factor expression, platelet activation, and complement-mediated inflammatory damage are some of the mechanisms involved in the pathophysiology of APS-related thrombosis. Both venous and arterial circulations are impacted by these mechanisms, which encourage a prothrombotic condition. Furthermore, the patient's decreased levels of natural anticoagulants, such as protein C, protein S, and antithrombin III, increased the thrombotic tendency and contributed to widespread involvement of the splanchnic venous system [7].

A considerable percentage of non-cirrhotic hepatic vein thrombosis cases in young adults are caused by the uncommon but well-known Budd-Chiari syndrome due to APS. As seen in this patient, coexisting PVT exacerbates portal hypertension and raises the risk of intestinal ischemia [8].

Due to the unusual demographic profile (young male), lack of traditional autoimmune disease characteristics, and non-specific initial presentation that resembled acute hepatocellular injury, the diagnosis in this instance was especially difficult. This frequently results in a delayed suspicion of thrombophilia. In acute life-threatening thrombotic events, early treatment is frequently started based on strong clinical suspicion; however, the revised Sydney criteria require both clinical evidence of thrombosis and persistent APLA positivity on two occasions at least 12 weeks apart for the diagnosis of APS [1].

Budd-Chiari syndrome, along with APS, necessitates a comprehensive approach. The mainstay of treatment to stop further thrombus expansion and recurrence is still anticoagulation. Interventional therapies, such as angioplasty or stenting are recommended in cases of hepatic venous outflow obstruction; in resistant cases, a transjugular intrahepatic portosystemic shunt may be necessary. This patient's hepatic vein insufficiency requires early intervention. Since direct oral anticoagulants have demonstrated poorer results in high-risk APS patients, especially those with arterial or recurrent venous thrombosis, long-term anticoagulation with Vitamin K antagonists continues to be the standard of therapy [7].

The successful outcome in this instance emphasizes how crucial it is to identify and treat APS-associated splanchnic vein thrombosis as soon as possible. Timely anticoagulation and radiology intervention led to significant biochemical and clinical improvement despite the severe presentation [8].

However, there is still a significant chance of recurrence, which calls for continuous anticoagulation and careful monitoring.

CONCLUSION

This case highlights a rare and life-threatening presentation of APS in a young male manifesting as simultaneous Budd-Chiari syndrome and extensive PVT. The atypical demographic profile and non-specific initial clinical features contributed to diagnostic difficulty and potential delay in identification. Prompt radiological evaluation and comprehensive thrombophilia workup were crucial in establishing the diagnosis.

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