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Antiphospholipid Antibody Syndrome: An Unusual Early Manifestation

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Abstract: ***Antiphospholipid antibody syndrome (APLS) is an acquired autoimmune disorder that manifests clinically as recurrent venous or arterial thrombosis and/or fetal loss. The actual frequency of APLS in the general population is unknown. The annual incidence of APLS has been estimated at approximately 5 cases per 100,000 persons, and the prevalence is approximately 40-50 cases per 100,000 persons. Genetic risk factors, infections and many drugs heighten the risk of antiphospholipid antibody-associated thrombosis. The clinical features vary significantly and can be as mild as asymptomatic APLA positivity, or as severe as catastrophic APLS. It can virtually affect any organ system. Here we present a case of a 19-year-old female with abdominal pain and normal lab investigations, revealed to be diagnosed with Budd-Chiari syndrome, with the primary being APLA positive.***

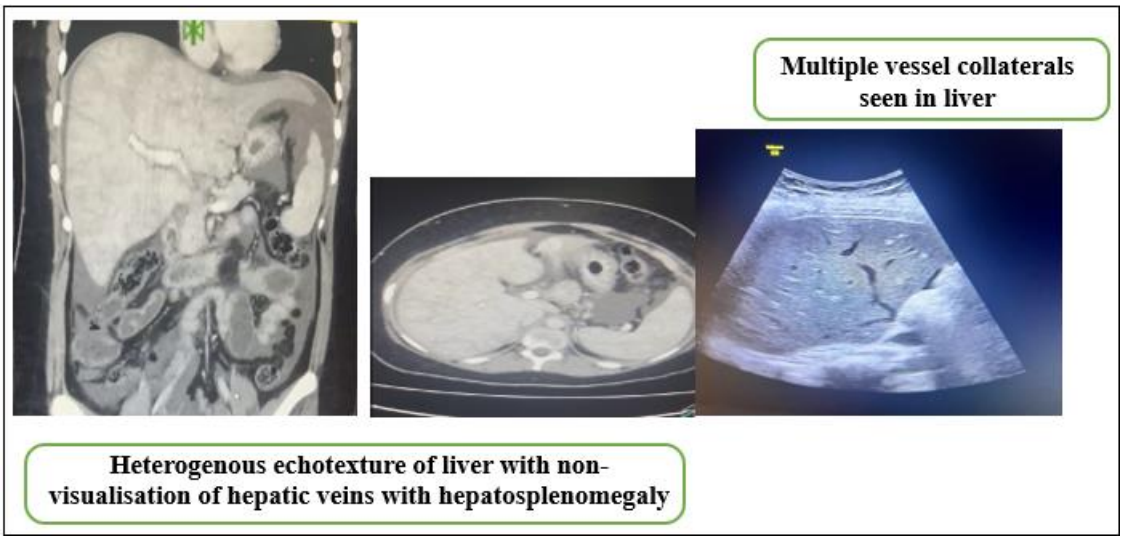
Keywords: autoimmune disorder, thrombosis, fetal loss, antiphospholipid antibodies, Budd-Chiari syndrome

1. Case Report

A 19-year-old unmarried female with no prior known comorbidities presented to casualty with abdominal pain and vomiting since 2weeks. She had similar complaints since past 1week for which she was evaluated in another hospital as IP patient, all relevant investigations were done and was diagnosed with acute gastritis and discharged with proton pump inhibitors (PPIs). At the time of presentation to our hospital, patient was conscious, oriented, vitals within normal limits. Per abdomen showed tenderness in epigastrium and right hypochondrium. Liver was palpable 4cm below the right costal margin, was tender and firm in consistency. There was moderate ascites and bowel sounds were heard normally. Outside blood report showed normal routine blood parameters which included CBC, LFT, RFT. CT abdomen done outside showed hepatomegaly with periportal edema, edematous gall bladder fossa- likely acute hepatitis and Mild ascites noted. Serology was negative. Endoscopy done showed lax LES gastritis of antrum and body. Patient got admitted and was started on PPI, pain killers and other supportive medications. Routine blood counts were repeated which included CBC, LFT, RFT, PT-INR, serology and the reports came within normal limits. USG abdomen and CECT abdomen were taken. Screening hepatic doppler was done which showed evidence of portal vein thrombosis probably budd chiari syndrome.

2. Investigations

Routine blood investigations were done. Complete blood count within normal limits with platelet count of 2.16lakhs. LFT showed total bilirubin 1.4 mg/dl, direct bilirubin-0.4mg/dl, AST- 44 IU/L, ALT- 40IU/L, ALP-72IU/L, total protein-5.8gm/dl, serum albumin-3.4 gm/dl. CRP was 20mg/dl. ESR-28mm/hr. Serum amylase and lipase within normal limits. PT-INR within normal limit with INR-1.22. Serum creatinine was 0.8mg/dl. USG abdomen was taken which showed liver mildly enlarged in size with span of 16.7cm and spleen mildly enlarged in size with span of 12.8cm. Portal vein and hepatic vein normal in course, caliber and dimension. Mild ascites was noted. ECHO was done which showed normal study. Diagnostic ascitic tapping was done and sample was sent for investigations. Ascitic fluid study showed high SAAG high protein ascites with fluid TC-150cells/cumm with neutrophils 0% and lymphocytes 100%. Fluid albumin-2.3g/dl, ADA-35.3U/L, AF protein-3.8gm/dl, LDH-118.3U/l. CECT abdomen was taken which showed hepatomegaly with heterogenous attenuation and periportal cuffing, mild pericholecystic fluid, splenomegaly, moderate ascites with left basal pleural effusion. Screening Hepatic doppler was done which showed portal vein thrombosis probably suggesting budd chiari syndrome.



3. Outcome and follow-up

Opinion of the interventional radiologist was taken. Hepatic venogram was done which showed focal narrowing near confluence with HPVG of 17mmHg. Right hepatic vein and inferior hepatic vein short segment osteal stenosis was noted. Right hepatic vein lesion was treated with venoplasty and stenting, inferior hepatic vein lesion treated by venoplasty. Post stenting venogram showed good flow through the stent with no stasis of contrast in hepatic veins. Post procedure vitals were within normal limits. Heparin infusion was started post procedure and APTT was monitored. Patient had developed left shoulder pain post procedure day-1. Cardiology opinion was taken and evaluation was found within normal limits. Patient showed clinical improvement. Workup for etiology for budd chiari syndrome was done. ANA and dsDNA were found to be negative. Anti-cardiolipin antibodies was elevated. Patient was diagnosed with APLA and was started on anti-coagulants. Patient was discharged with stable vitals and advised to continue on Tab.Warfarin 2/3mg daily and to monitor INR and maintain between 2-3. Few days after discharge, patient had sudden onset chest pain and vomiting. Went into arrest and succumbed to death at home. Probable cause of death was considered to be Myocardial infarction.

4. Discussion

The hallmark of APLS comprises the presence of persistent antiphospholipid antibodies (APLA) in the setting of arterial and venous thrombus and/or pregnancy loss. Antiphospholipid syndrome can be primary when there is no evidence of autoimmune disease, or it can be secondary to autoimmune processes like systemic lupus erythematosus (SLE) in 40% of the cases. APLS may contribute to an increased frequency of stroke or MI, especially in younger individuals.

There is another subtype with fatal manifestation termed as catastrophic APLS which is characterised by multiorgan infarctions progressing rapidly over days to weeks with mortality around 50%. Clinically, the series of events that leads to hypercoagulability and recurrent thrombosis can affect virtually any organ system, including the following: Peripheral venous system (deep venous thrombosis), Hematologic (thrombocytopenia, hemolytic anemia), Obstetric (pregnancy loss, eclampsia), Pulmonary (pulmonary embolism [PE], pulmonary hypertension), Dermatologic (livedo reticularis, purpura, infarcts/ulceration), Cardiac (Libman-Sacks valvulopathy, MI, diastolic dysfunction), Ocular (amaurosis, retinal thrombosis), Adrenal (infarction/hemorrhage), Musculoskeletal (avascular necrosis of bone), Renal (thrombotic microangiopathy) and many more.

The new classification criteria for antiphospholipid syndrome (APLS) developed by the American College of Rheumatology (ACR) and the European Alliance of Associations for Rheumatology (EULAR) in August 2023 aim to enhance the classification of APLS by considering advancements in understanding both thrombotic and nonthrombotic clinical manifestations, as well as the significance of APLS antibody profiles in thrombosis risk assessment. To classify as APLS, a patient needs a minimum of 3 points from the clinical domains and 3 points from the laboratory domains. These criteria incorporate advancements in understanding nonthrombotic clinical manifestations, traditional thrombosis risk factors in APLS antibodies patients, and risk stratification based on APLS antibody profile.

Management of APLS includes eliminate risk factors, such as oral contraceptives, smoking, hypertension, and hyperlipidemia. Prophylactic anticoagulation is needed during surgery or hospitalization. Management of any associated autoimmune disease is necessary, full anticoagulation with intravenous or subcutaneous heparin followed by warfarin therapy. Based on the most recent evidence, a reasonable target for the international normalized ratio (INR) is 2.0-3.0 for venous thrombosis and 3.0 for arterial thrombosis. Direct oral anticoagulants (i.e., direct thrombin inhibitors and factor Xa inhibitors such as rivaroxaban) have been used in patients with warfarin intolerance/allergy or poor anticoagulant control but are not found to be much effective in treatment. Surgical care should be given in the form of stenting/TIPS/ IVC filters. Regular follow up and prevention of complications is of utmost importance for good quality of life.

5. Conclusion

Any young patient presenting with features of Budd-Chiari syndrome should always be screened for APLA as well as for SLE as Budd -Chiari syndrome presenting as initial manifestation of APLS is rare and not much evidences are present. Early screening for these conditions will go a long way in preventing future complications like repeated miscarriages and other multi-system complications by timely intervention and close follow up with anticoagulants and antihepatic coma measures.

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