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CASE REPORT

DUAL THROMBOPHILIC DISORDER PRESENTING AS RECURRENT MYOCARDIAL INFARCTION IN A YOUNG MALE: APLA WITH FACTOR V LEIDEN MUTATION

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ABSTRACT

Background: Antiphospholipid syndrome (APS) and Factor V Leiden mutation are independent thrombophilic disorders predisposing to arterial and venous thrombosis. Their coexistence significantly amplifies thrombotic risk. Myocardial infarction (MI) is an uncommon presentation of APS, and recurrent MI in association with dual thrombophilia is exceedingly rare. **Case Presentation:** A 41-year-old male presented with acute-onset retrosternal chest pain radiating to the left arm, accompanied by vomiting. He had a prior history of ST-elevation myocardial infarction (STEMI) one month earlier. Laboratory investigations revealed elevated cardiac biomarkers and severe renal and hepatic dysfunction. Thrombophilia profile confirmed dual positivity for antiphospholipid antibodies (IgG and IgM) with positive lupus anticoagulant, along with Factor V Leiden mutation detected by PCR. Echocardiography demonstrated severe left ventricular systolic dysfunction (LVEF 15–20%) with moderate mitral and tricuspid regurgitation. The patient was managed with dual antiplatelet therapy, anticoagulation, statins, and supportive care, followed by transition to long-term warfarin therapy targeting INR 3. **Discussion:** The coexistence of APS and Factor V Leiden mutation represents a synergistic prothrombotic state leading to recurrent arterial events even in young individuals without conventional cardiovascular risk factors. The case emphasizes the need for comprehensive thrombophilia screening in young patients presenting with unexplained arterial thrombosis. **Conclusion:** Dual thrombophilic disorders such as APS with Factor V Leiden mutation markedly increase thrombotic risk and may manifest as recurrent myocardial infarction in young adults. Early recognition and lifelong anticoagulation are crucial to prevent recurrence and improve outcomes. **Keywords**: Antiphospholipid syndrome, Factor V Leiden mutation, recurrent myocardial infarction, dual thrombophilia, young adult, warfarin.

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INTRODUCTION

Antiphospholipid syndrome (APS) is an autoantibody-mediated acquired thrombophilia characterized by recurrent arterial or venous thrombosis, microvascular manifestations, and/or pregnancy morbidity.[1]Clinical manifestations range from asymptomatic aPL positivity (no history of vascular or pregnancy events) to catastrophic APS (multiple thromboses occurring over days). [2]Factor V Leiden is a point mutation of factor V, resulting in an elimination of the cleavage site in factor V and factor Va. This genetic defect leads to an increased risk of thrombosis, especially in homozygous or pseudo-homozygous factor V Leiden mutations.[3] Both of these increases the risk of thrombosis in arterial and venous system. MI as a presentation of APLA syndrome is seen in about 10% patients but there is no such data regarding the recurrent MI and concurrent APLA with Factor V Leiden mutation.

CASE HISTORY

A 41-year-old male, resident of Chittorgarh, presented to Maharana Bhupal Government Hospital, RNT Medical College, Udaipur on September 9, 2025, with acute onset retrosternal chest pain radiating to the left arm and shoulder, accompanied by vomiting. The patient had a significant history of STEMI one month prior to this current presentation, making this a recurrent acute coronary event in a relatively young individual.1 month back, he was advised coronary angiography

but patient refused. He had a smoking history of 1 pack daily since 2015 but had reportedly quit about 5 years back. There was no history of diabetes mellitus, hypertension, chronic kidney disease, tuberculosis, alcohol addiction, or family history of premature coronary artery disease.

Examination: On physical examination, the patient was conscious, alert, and oriented with vital signs revealing hypertension (BP 160/90 mmHg), pulse rate of 80-94/min, respiratory rate of 16-18/min, and oxygen saturation of 96-98% on room air. Cardiovascular examination showed apex beat at the 5th intercostal space in the left midclavicular line, with no abnormality initially detected. Respiratory system examination revealed bilateral equal air entry with no added sounds, abdominal examination was unremarkable with no organomegaly, and neurological examination showed normal higher functions with no focal deficits.

Lab Investigations & Imaging: Laboratory investigations revealed significantly elevated cardiac biomarkers with Troponin I of 0.36 ng/ml (normal <0.04 ng/ml) on July 26, 2025, and Troponin T of 0.092 ng/ml (normal 0.0-0.014 ng/ml) on September 10, 2025, confirming myocardial injury. Complete blood count showed hemoglobin of 15.4-16.0 g/dl, mild leukocytosis with total leukocyte count of 12.0 x 10^³muL with neutrophil predominance of 78.8%, and normal platelet count of 229 x 10^3muL. Renal function tests were severely deranged with serum creatinine of 5.6-5.7 mg/dl (normal 0.7-1.2 mg/dl), blood urea of 132-144 mg/dl (normal 16.6-48.5 mg/dl), and uric acid of 13.1-13.3 mg/dl (normal 3.4-7.0 mg/dl). Liver function tests showed significant elevation with SGOT (AST) of 318-384 U/L (normal 10-50 U/L), SGPT (ALT) of 346-659 U/L (normal 10-50 U/L), total bilirubin of 0.325-0.849 mg/dl, and serum albumin of 2.71-3.58 g/dl.ECG showed t wave inversion in lead 1;aVL;V1-V6 with ST depression in V4-V6.

The comprehensive thrombophilia workup revealed striking findings suggestive of dual thrombophilia. Antiphospholipid syndrome profile showed positive anti-phospholipid IgM of 19.50 U/mL (normal <12.00) and anti-phospholipid IgG of 21.03 U/mL (normal <12.00), raised anti-cardiolipin IgG of 47.63 GPl/ml (normal <12.00) and anti-cardiolipin IgM of 16.00 MPL/ml (normal <12.00), and positive lupus anticoagulant with prolonged APTT of 58.25 sec (normal 22.2-30.7 sec) and elevated dRVVT Screen Ratio of 2.18 (normal 0.85-1.20). Hereditary thrombophilia workup revealed Factor V Leiden mutation detected by PCR, while Protein C functional activity was 91.30% (normal 83-168%), Protein S free was 82.46% (normal 74.1-146.1%), and homocysteine was 11.6 Î¼mol/L (normal 3.7-15.0 Î¼mol/L), all within normal limits. Echocardiography revealed severely compromised cardiac function with left ventricular ejection fraction (LVEF) of 15-20%, left ventricular internal diameter in diastole of 5.72 cm indicating dilatation, fractional shortening of 9.27% which was severely reduced, dilated left atrium measuring 4.3 cm, moderate mitral regurgitation, and moderate to severe tricuspid regurgitation with pulmonary artery systolic pressure of 45-50 mmHg. No wall motion abnormalities were specifically documented, and there was no evidence of clot, pericardial effusion, or vegetations. Abdominal ultrasound showed altered liver echo texture suggestive of hepatic dysfunction, bilateral echogenic kidneys indicating Grade I renal parenchymal disease, a right renal cortical cyst measuring 25x14 mm, but maintained cortico-medullary differentiation. Renal Doppler study showed normal renal arterial flow at origin and hilum with no evidence of renal artery stenosis. Additional investigations showed blood sugar levels of 91.6-113.0 mg/dl within normal limits, lipid profile revealing low total cholesterol of 69.1-78.5 mg/dl, low HDL of 19.6-25.9 mg/dl, and low LDL of 31.0-39.0 mg/dl. Infectious disease workup was negative for dengue serology, malaria, HIV, HBsAg, and HCV. Coagulation studies showed normal prothrombin time of 14.2 seconds with INR of 1.05.

Management and Outcome: The patient was managed with dual antiplatelet therapy consisting of aspirin 75 mg and clopidogrel 75 mg daily, high-intensity statin therapy with atorvastatin 80 mg daily, beta-blocker metoprolol 25 mg twice daily, ACE inhibitor ramipril 2.5 mg once daily, anticoagulation with enoxaparin 0.4 mL subcutaneous twice daily, diuretic furosemide 40 mg for heart failure management, and supportive care including intravenous fluids, proton pump inhibitor, and symptomatic treatment. Cardiology consultation confirmed the diagnosis of coronary artery disease with acute coronary syndrome (NSTEMI) complicated by acute kidney injury and cardiorenal syndrome, while nephrology consultation was obtained for management of acute kidney injury and chronic kidney disease. Patient felt marked reduction in chest pain and exertional dyspnea after treatment. There was reduction in the pedal edema. Patient could not be taken for coronary angiography study or CT Coronary Angiography due to deranged Renal function. Patient was started on oral vitamin K antagonist- Warfarin 5mg OD and uptitrated till the target INR of 3 was achieved.

Final Diagnosis: The final clinical diagnoses included primary diagnoses of coronary artery disease with acute coronary syndrome (NSTEMI), recurrent myocardial infarction (second event within one month), severe left ventricular systolic dysfunction (LVEF 15-20%), and congestive heart failure. Secondary diagnoses encompassed primary antiphospholipid syndrome with positive lupus anticoagulant and positive anti-phospholipid antibodies (IgG and IgM) and positive anti cardiolipin IgM and IgG, Factor V Leiden mutation, acute kidney injury with cardiorenal syndrome type 1, hepatic dysfunction likely secondary to cardiac hepatopathy, and newly diagnosed hypertension.

DISCUSSION

This case represents a rare presentation of recurrent myocardial infarction in a 41-year-old patient with dual thrombophilia, highlighting the critical importance of comprehensive thrombophilia evaluation in young adults presenting with arterial thrombosis. The coexistence of primary antiphospholipid syndrome and Factor V Leiden mutation creates a synergistic prothrombotic state that significantly exceeds the risk of either condition alone. Antiphospholipid antibodies exert their pathogenic effects by binding to phospholipid-binding proteins such as β2-glycoprotein I, leading to endothelial activation, complement cascade activation, and thrombus formation through non-atherosclerotic mechanisms [4]. Factor V Leiden mutation results in activated protein C resistance, preventing normal anticoagulant pathway function and maintaining procoagulant activity [5]. The combination of these mechanisms creates a particularly high-risk hypercoagulable state, with combined thrombophilias conferring 20-50 fold increased thrombotic risk compared to isolated defects[6].

Myocardial infarction occurs in less than 3% of APS patients initially, making arterial presentations uncommon compared to venous thrombosis [7]. The pathogenesis involves coronary thrombosis rather than premature atherosclerosis, explaining occurrence in young patients without traditional cardiovascular risk factors [8]. The "two-hit" hypothesis suggests that Factor V Leiden mutation may serve as the additional genetic risk factor amplifying antiphospholipid antibody thrombotic potential [9]. Management requires lifelong anticoagulation with vitamin K antagonists, as direct oral anticoagulants show increased arterial thrombosis risk in APS patients [10]. The severe cardiac dysfunction (LVEF 15-20%) and development of cardiorenal syndrome emphasize the serious prognostic implications of recurrent arterial events in young patients with combined thrombophilia. This case underscores the need for systematic thrombophilia screening in young adults with unexplained arterial thrombosis and highlights the importance of recognizing combined prothrombotic conditions that may require aggressive, specialized management which includes lifelong anticoagulation to prevent catastrophic recurrent events.

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