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A Case Report of Anti-Phospholipid Syndrome in a Male with Recurrent Deep Vein Thrombosis

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Abstract: Anti-phospholipid syndrome (APS) is an autoimmune-mediated thrombophilia characterized by recurrent thrombotic events, particularly affecting young adults. It is linked to the presence of anti-phospholipid antibodies that interfere with normal coagulation pathways, leading to abnormal blood clot formation. This case report discusses a rare occurrence of APS in a middle-aged male who presented with recurrent deep vein thrombosis (DVT). Comprehensive diagnostic evaluation, management strategies, and follow-up protocols are elaborated to provide insights into this complex clinical entity.

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I. INTRODUCTION

Anti-phospholipid syndrome (APS) is an autoimmune disorder marked by the presence of anti-phospholipid antibodies, which predispose individuals to both arterial and venous thrombotic events. It was first recognized as a distinct clinical entity due to its association with recurrent miscarriages, stroke, and thrombosis. APS is considered one of the most common causes of acquired thrombophilia, particularly affecting young adults.

➢ *Epidemiology*

APS predominantly affects females of childbearing age, but when occurring in males, it presents with a more severe thrombotic phenotype. Approximately one-third of APS patients experience thrombotic complications. In men, the most common presentation is venous thrombosis, often in the form of deep vein thrombosis (DVT) or pulmonary embolism. The mean age of presentation is around 30.1 years.

➢ *Pathophysiology*

The pathogenesis of APS involves the production of anti-phospholipid antibodies, including lupus anticoagulant, anti-cardiolipin antibodies, and anti-beta-2 glycoprotein I antibodies. These antibodies interfere with the coagulation cascade, endothelial function, and platelet activation, leading to a hypercoagulable state. They also promote inflammation, which further exacerbates thrombus formation.

II. CASE REPORT

A 36-year-old male presented to the emergency department with bilateral swelling of the lower limbs accompanied by pain, particularly while walking, persisting for two days. He reported a history of similar episodes over the past year, for which he received symptomatic relief without a definitive diagnosis.

➢ *Medical History*

- No known history of diabetes, hypertension, or cardiovascular disease.
- No history of recent trauma, surgery, or prolonged immobilization.
- No family history of thrombophilia or autoimmune disorders.

➢ *Physical Examination*

- Vital signs: Blood pressure - 110/70 mm Hg, Pulse - 88 beats per minute, Respiratory rate - 18 breaths per minute, Temperature - afebrile.
- Bilateral lower limb swelling was noted, with more pronounced edema on the right side.
- No pallor, cyanosis, clubbing, or lymphadenopathy.
- Negative Homan's sign.

➢ *Differential Diagnosis*

- Deep vein thrombosis (DVT)
- Chronic venous insufficiency
- Lymphedema
- Cellulitis
- Autoimmune thrombophilia (e.g., APS)

➢ *Investigations*

A comprehensive diagnostic workup was conducted to evaluate the cause of recurrent DVT. The findings were as follows:

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➢ *Laboratory Tests*

- Complete Blood Count: Normal white blood cell count and hemoglobin levels, Platelet Count: 1.43 lakh/µL (within normal range).
- Coagulation Profile:
- Prothrombin Time (PT): 15 seconds (normal range).
- Activated Partial Thromboplastin Time (APTT): 25 seconds.
- International Normalized Ratio (INR): 1.0.
- D-Dimer: Elevated, indicating active thrombus formation.
- Lupus Anti-Coagulant:
- Screen (LVT) - 59.4 seconds (elevated).
- Confirm - 40.1 seconds (elevated).
- Anti-Phospholipid Antibodies:
- Anti-cardiolipin antibodies: Positive.
- Beta-2 glycoprotein I antibodies: Positive.
- Thrombophilia Profile: Negative for Factor V Leiden mutation, Prothrombin gene mutation, and Protein C/S deficiency.

➢ *Imaging Studies*

- Venous Doppler Ultrasound:
- Right lower limb: Thrombus in the distal saphenous femoral vein.
- Left lower limb: Chronic thrombus in the common femoral and superficial femoral veins.
- No evidence of pulmonary embolism on chest CT scan.

III. DIAGNOSIS

➢ *The patient was diagnosed with anti-phospholipid syndrome (APS) based on:*

- Recurrent deep vein thrombosis (DVT).
- Positive lupus anticoagulant and anti-cardiolipin antibodies.
- Absence of other underlying prothrombotic conditions.

➢ *Management*

The primary goal of management in APS is to prevent recurrent thrombotic events through long-term anticoagulation therapy. The patient was initiated on the following treatment regimen:

- Intravenous Heparin (5000 IU TID) - To achieve rapid anticoagulation.
- Warfarin (Tab ACITROM 2 mg OD) - Oral anticoagulant to maintain an INR of 2-3.
- Ecosprin-AV (75/10 mg OD) - Antiplatelet agent for additional protection against arterial thrombosis.
- HOMOCHECK OD - For monitoring and maintaining INR within therapeutic range.

➢ *Titration and Monitoring*

- INR was monitored bi-weekly until therapeutic range was achieved.
- Once stabilized, monthly INR checks were conducted.
- No adverse drug reactions or bleeding complications were observed.

IV. DISCUSSION

Anti-phospholipid syndrome (APS) is a prothrombotic autoimmune disorder that predisposes individuals to both venous and arterial thromboembolism. The presence of anti-phospholipid antibodies interferes with the coagulation cascade by activating endothelial cells and platelets, leading to a hypercoagulable state.

➢ *Challenges in Diagnosis*

Diagnosing APS can be challenging due to the variability of clinical manifestations. In this patient, the absence of traditional risk factors and the recurrent nature of DVT raised suspicion of an underlying autoimmune cause, confirmed by laboratory findings of lupus anticoagulant and anti-cardiolipin antibodies.

➢ *Long-Term Management*

Lifelong anticoagulation is recommended for patients with APS and recurrent thrombotic events. Warfarin remains the mainstay of treatment, but newer oral anticoagulants are being evaluated. Regular monitoring of INR is crucial to maintaining therapeutic levels and preventing complications.

➢ *Prognosis and Follow-Up*

- Lifelong follow-up with a hematologist is recommended.
- Regular INR monitoring and dose adjustment are essential.
- The patient was educated on recognizing symptoms of recurrent thrombosis and bleeding.

V. CONCLUSION

This case illustrates a rare presentation of anti-phospholipid syndrome (APS) in a middle-aged male with recurrent DVT. It emphasizes the importance of considering APS in patients with unexplained thrombotic events and highlights the need for comprehensive diagnostic evaluation and lifelong anticoagulation management. Early diagnosis and appropriate anticoagulation can significantly improve outcomes and prevent life-threatening complications.

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