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Unveiling a Rare Presentation: Axillary Vein Thrombosis as an Initial Sign of Essential Thrombocythemia

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Abstract: ***This article presents a unique case study highlighting an unusual manifestation of Essential Thrombocythemia ET where axillary vein thrombosis serves as the presenting feature. Although thrombotic complications are commonly associated with ET, axillary vein thrombosis remains a seldom reported occurrence within ET diagnoses. The case involves a 70 - year - old male with a history of multiple medical conditions who exhibited symptoms of upper limb pain and swelling. Through comprehensive diagnostic procedures, including genetic studies, bone marrow aspiration, and imaging, ET was confirmed as the underlying cause. The article delves into the complexity of essential thrombocythemia, discussing its molecular pathogenesis, diagnostic challenges, and management strategies. The presented case underscores the importance of recognizing atypical presentations and utilizing a multidisciplinary approach for accurate diagnosis and effective disease management.***

Keywords: Axillary vein thrombosis, Essential thrombocythemia, Case report, Molecular pathogenesis, Diagnostic challenges

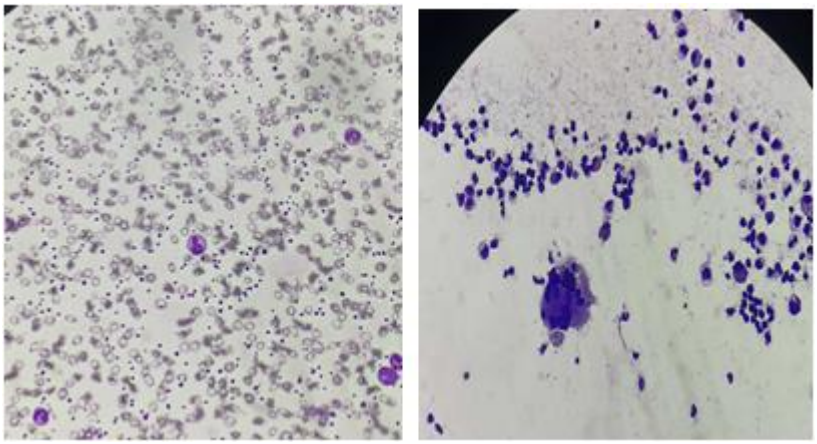
1. Introduction

Essential thrombocythemia (ET), also known as primary thrombocythemia, is a rare chronic myeloproliferative neoplasm characterized by persistent elevation of platelet counts in the peripheral blood. The exact incidence and prevalence of ET remain uncertain, but it is estimated to affect approximately 1 to 2 per 100, 000 individuals per year, with a slight female predominance. The underlying molecular pathogenesis of ET involves mutations in genes such as Janus kinase 2 (JAK2), calreticulin (CALR), and myeloproliferative leukaemia (MPL). However, a significant proportion of ET cases are labelled as "triple - negative," lacking mutations in these driver genes.

2. Case Report

A 70yearold malesenior citizen who is a known case of Hypertrophic Obstructive Cardiomyopathy (HOCM) (mild Left Ventricular Outflow obstruction), Hypertensive, Non Diabetic and CVA - AIS came to hospital with left hand pain and swelling giving a prior history of physiotherapy of left shoulder for what was diagnosed as periarthritis left shoulder. On examination, upper limb swelling was associated with erythema, tender with shiny skin with palpable peripheral arterial pulses.

His basic laboratory routines revealed Haemoglobin of 9.6 g/dl with thrombocytosis 12.3 lakhs/cummand differential count of 19400 cells/cumm. Peripheral smear gave impression of Erythropenia showing Normocytic Normochromic and Microcytic Hypochromic picture with Lymphopenia and Thrombocytosis. C Reactive Protein (CRP) was negative and Iron studies revealed Serum Ferritin on lower side of normal range giving impression of Mild Anaemia. Renal parameters, Liver function tests and Serum Electrolytes were in physiological levels. International Normalised Ratio (INR) was 1.1 and D - Dimers was 800 nanogram/millilitre.



Pheripheral Smearbone Marrow Aspirate

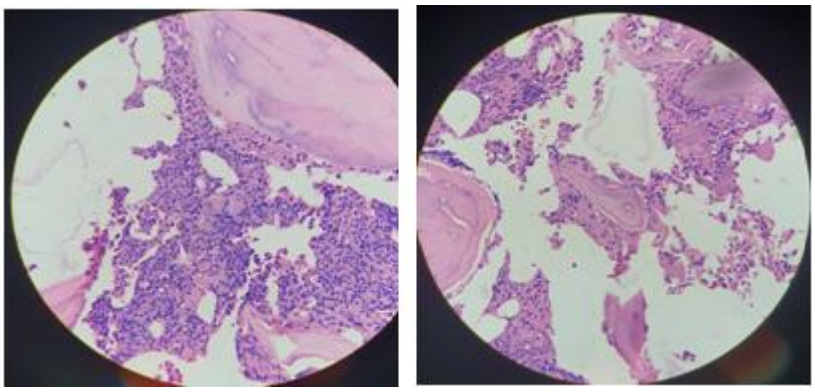


Figure A Figure B

Figure A & B: BONE MARROW BIOPSY

On imaging, Venous doppler revealed axillary vein thrombosis extending beyond cephalic vein and shown intramuscular oedema. And UltraSono - Graph (USG) of abdomen shown moderate splenomegaly of 16.4cms and mild hepatomegaly of 14cms.

And he was started on hydroxyurea and IV Unfractioned Heparin (UFH); 48 hrs later his platelets got dropped to 8.2 lakhs/ cumm. As patient is having thrombocytosis and splenomegaly, he underwent Bone marrow aspiration and biopsy which gavean impression of increased cellularity with scattered Megakaryocytes suggestive of myeloproliferative neoplasm and possibility of essential thrombocythemia maybe considered. For which Genetic studieswere done which revealed JAK2 as positive and MPL, CALR as negative. Bone marrow biopsy and genetic studies gave inference of essential thrombocythemia.

Outcome

Patient got better symptomatically and discharged with Novel oral anticoagulants/ apixaban, hydroxyureaand his thrombocytosis came down to 5 lakhs/cumm.

3. Discussion

Essential thrombocythemia (ET) presenting as axillary vein thrombosis is a rare manifestation. But here patient presented clinically due to prior history of manipulation by physiotherapist which got diagnosed as ET after excluding every possible aetiology of thrombosisand secondary causes of thrombocytosis with findings of bone marrow aspiration and biopsy which revealed no fibrotic changes in bone marrow that rules out Primary Myelofibrosis and Essential thrombocythemia got confirmed with identification of genetic mutations. The underlying molecular pathogenesis of ET involves mutations in genes leading to aberrant activation of JAK - STAT signalling and dysregulated haematopoiesiscausing clonal activation of hematopoietic cells causing thrombosis. Low - risk patients are often monitored closely without immediate therapy, whereas high - risk patients or those with symptomatic disease require treatment. The mainstay of therapy includes aspirin for thromboprophylaxis, cytoreductive agents (e. g., hydroxyurea, interferon - alpha, anagrelide) to reduce platelet counts, and antiplatelet agents for high - risk patients. Novel targeted therapies, such as JAK inhibitors and other emerging agents, are being explored in clinical trials and show promising results in the management of ET.

4. Conclusion

In conclusion, essential thrombocythemia is a complex disorder characterized by persistent thrombocytosis and carries a risk of thromboembolic and haemorrhagic complications. Understanding its underlying pathophysiology, diagnostic criteria, and treatment options is crucial for effective disease management and improved patient outcomes.

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