International Journal of Biomedical And Advance Research

ISSN: 2229-3809 (Online)
Journal DOI:10.7439/ijbar
CODEN:IJBABN

Case Report

Essential Thrombocythemia

Adnan Bashir Bhatti*[1], Farhan Ali[2] and Siddique Akbar Satti[3]

[1]*Department of Medicine, Capital Development Authority Hospital, Islamabad, Pakistan*
[2]*Senior Registrar, Department of Medicine , Capital Development Authority Hospital, Islamabad, Pakistan.*
[3]*Consultant Physician & Head of Department of Medicine, Capital Development Authority Hospital, Islamabad, Pakistan.*

***Correspondence Info:**
Adnan Bashir Bhatti, M.D
Department of Medicine,
Capital Development Authority Hospital, Islamabad, Pakistan
Email: dr.adnanbashir@gmail.com

Abstract

Essential thrombocythemia is an uncommon disease. It involves an overproduction of megakaryocytes in the marrow. These platelets may not function normally and can cause a blockage in blood vessels and other complications. We report two cases of essential thrombocythemia with different clinical presentations. One with off and on headache and other with stroke, facial palsy and gangrene of hand.

Keywords: Thrombocythemia, megakaryocytes, headache, stroke, facial palsy, gangrene, myeloproliferative disorder, polycythemia vera, lymphadenopathy.

1. Introduction

The classic myeloproliferative disorder includes polycythemia vera, primary myelofibrosis and essential thrombocythemia.[1] Essential thrombocythemia is neither a cytogenetically nor a morphological defined disease entity, but rather is a diagnosis of exclusion. Therefore, the diagnosis of essential thrombocythemia requires that both reactive thrombocytosis and other chronic myeloid disorders have been omitted.[2] Most patients with essential thrombocythemia enjoy a normal life expectancy without associated disease-related complications[3, 4]. In morphological and cytogenetically defined essential thrombocythemia the delayed development of either acute myeloid leukemia (AML) or post essential thrombocythemia myelofibrosis is unusual [5].

2. Case Report

2.1 Case One

A 35 years old female presented to Capital Development Authority (CDA) Hospital, Islamabad, Pakistan, with complaint of recurrent headache and blackouts. She was known case of hypertension for the last six months and she was taking different medications from a local general practitioner for headache but her complaint was not improving. Initially she was treated for migraine; now for the last few days she complained of numbness in hands and feet. Physical examination her weight was 55 kg and height 142cm; there was no pallor, edema, clubbing, leukonychia, jaundice, lymphadenopathy. Her pulse rate was 80/min, blood pressure 110/80 mmHg, respiratory rate 14/min, temperature was normal and there was no bony tenderness anywhere. On systemic examination pain and touch sensation were decreased in feet but reflexes and power were normal. Dorsalis pedis pulsation were feeble but other peripheral pulses were intact. In gastrointestinal system spleen was enlarged 6cm below the subcostal margin, nontender, firm in consistency. Cardiovascular

system, respiratory system, ear, throat, dental and fundus examination were normal.

Investigation revealed blood glucose fasting, random, liver function test, renal function test, fasting lipid profile, urine examination, uric acid level, vitamin B12 level, ESR, CRP, bleeding time, clotting time, prothombin time, serum iron level, total iron binding capacity, serum electrolytes, serum protein C and S level, X- ray chest and ECG were normal. Doppler's study of the lower limbs was normal. Ultrasound abdomen and pelvis showed spleen enlargement. Blood complete picture showed hemoglobin 12.5gm/dl , hematocrit 48%, platelet count 1,517,000, total leukocyte count $6x10^{9}$, polymorphs 62%, lymphocytes 23%, monocytes 3%, eosinophil 1%. Peripheral blood smear revealed abundance of megakaryocytes.

Bone marrow revealed giant megakaryocytes and hyperplasia, trephine biopsy revealed hyper cellular marrow, no fibrosis and increased platelets. A diagnosis of essential thrombocytopenia was made and the patient was treated with hydroxyurea 15mg/kg body weight and low dose Aspirin. The patient was monitored and her platelet count decreased after treatment.

2.2 Second Case

A 43 years old normotensive and normoglycemic female presented with pain and blackish discoloration of right hand and weakness of right half of the body with a left sided deviation of face.

She has a past history of off and on headache and pain in the eyes which were relieved temporarily by taking analgesics. She belongs to the poor socioeconomic group. She did not visit to any physician for treatment of these ailments.

On physical examination her weight was 60 kg and height 140cm, there was no pallor, edema, clubbing, leukonychia, jaundance, and lymphadenopathy. Her pulse rate was 76/min, Blood pressure 130/70 mmHg, respiratory rate 14/min, temperature was normal. On central nervous examination she was conscious and oriented, power 3/5 in right upper and lower limbs; muscle tone and reflexes were exaggerated on right side of body while right plantar was upgoing. Sensory system was intact; right sided facial palsy while remaining nerves were intact. Cerebellar system was also intact. On local examination of hand gangrene was present. Right radial and ulnar pulsations were feeble but other peripheral pulses were intact. In gastrointestinal system tip of the spleen was palpable. Cardiovascular system, respiratory system, ear, throat, dental and fundus examination was normal.

Blood complete picture showed hemoglobin 11.5gm/dl , hematocrit 48%, platelet count 2,411,000, total leukocyte count $11x10^{9}$, polymorphs 65%, lymphocytes 21%, monocytes 2%, eosinophil 3%. Peripheral blood smears revealed abundance of megakaryocytes. Bone marrow and trephine biopsy showed features of thrombocythemia.

Magnetic resonance angiography (MRA) of right hand showed distal radial and ulnar arteries were visualized up till the wrist behind the wrist radial artery were completely obliterated and not visualized in hand while ulnar artery demonstrated beaded appearance beyond the wrist.

The artery to the radial aspect of thumb common palmer digital artery, deep palmer arch, deep branches of the ulnar artery, palmer metacarpal artery, princeps pollicis artery and proper palmer artery were marked attenuated with poor visualization concerning almost complete occlusion.

CT scan brain showed infarct in the left cerebral hemisphere in supply of middle cerebral artery. Ultrasound abdomen and pelvis showed enlarged spleen. ECG & echocardiography were normal other investigations regarding causes of stroke were normal. All investigations regarding secondary thrombocythemia were also normal.

So the diagnosis of right hand gangrene and right sided hemi paresis with right facial palsy secondary to primary thrombocythemia were made. Patient was admitted in the intensive care unit and hydroxyurea were started according to body weight. Unfortunately her right hand amputation was done. Supportive treatment and physiotherapy was started for stroke. She was monitored for platelet count. It came in normal range after two weeks of therapy. She was discharged in stable condition on asprin, clopidegrol and hydroxyurea.

Fig 1. Gangrene of Right Hand (Second Case)

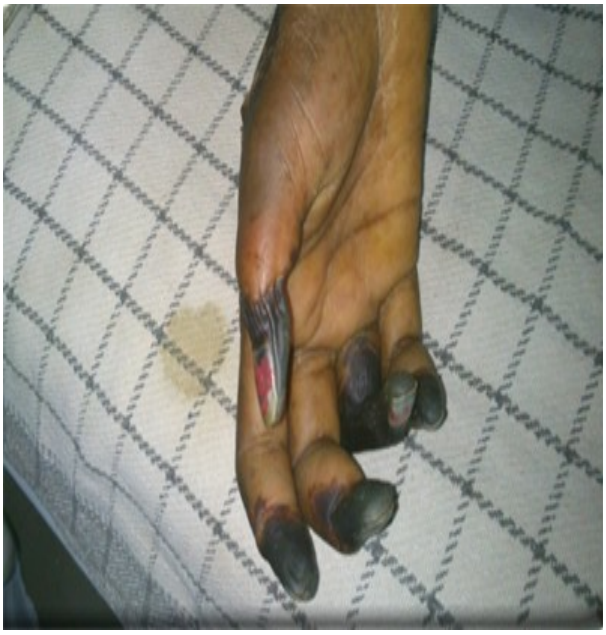


Fig 2. Gangrene of Right Hand (Second Case)

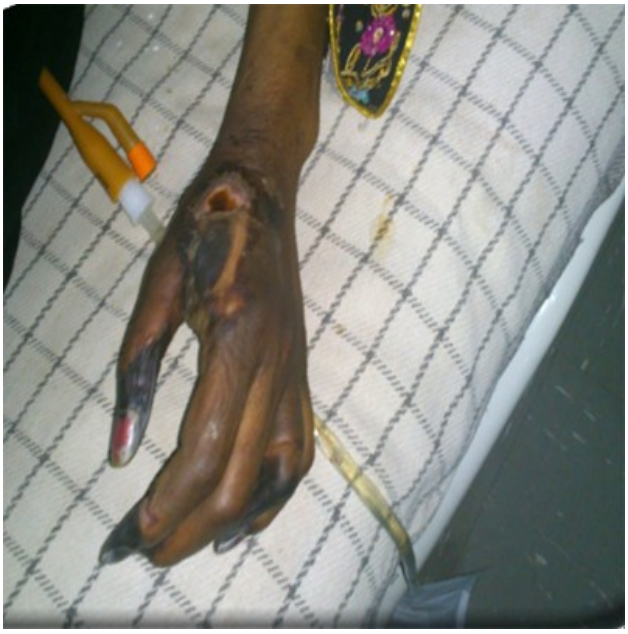
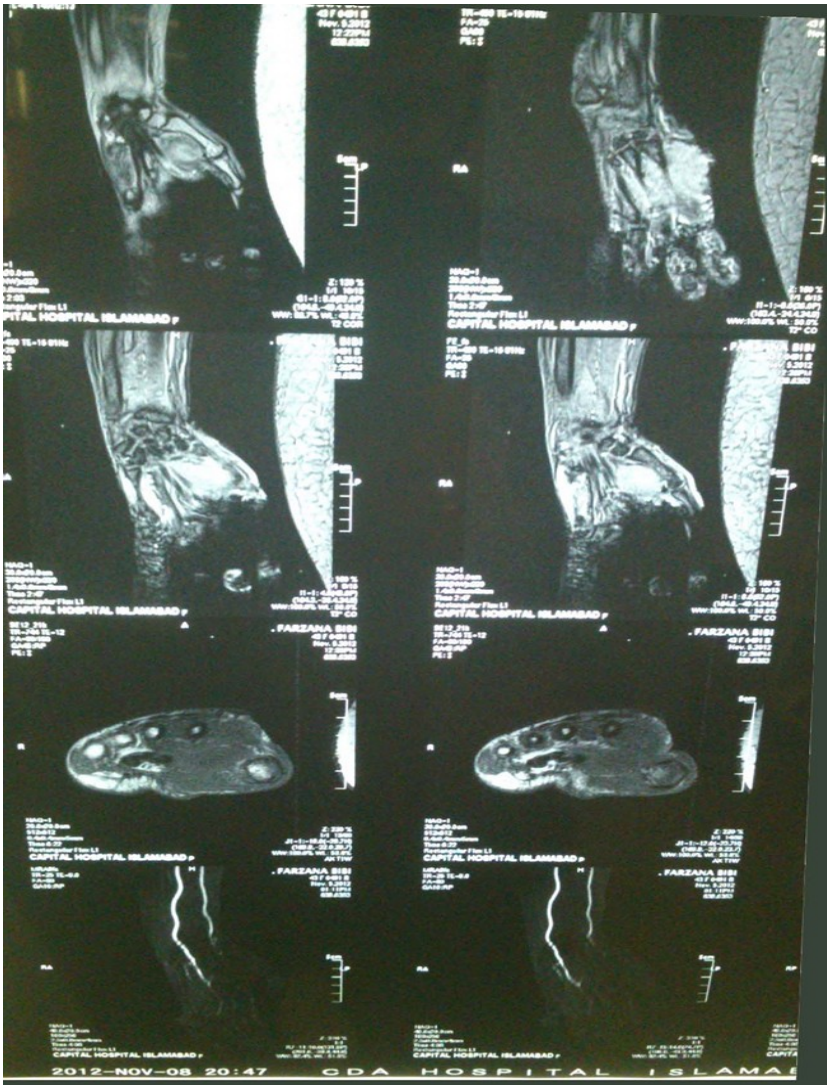


Fig 3. Right sided hemi paresis and facial palsy (Second Case)

(Pictures after consent of patient)

Fig 4. Magnetic resonance angiography (MRA) of right hand (Second Case)



3. Discussion

Thrombocytosis is the presence of high platelet counts in the blood more than six hundrad thousand per ml[6]. Abnormalities in the number and function of platelets may contribute to thromboembolic complications in patients with essential thrombocythemia.[7] Thrombocytosis can be a reactive (secondary thrombocytosis) or a clonal bone marrow (myeloproliferative) process. Reactive thrombocytosis, which is more common, is caused by increased levels of thrombopoietin (TPO), other cytokines, or catecholamine that may be produced in inflammatory, infectious, stressful or neoplastic conditions. Clonal thrombocytosis is commonly seen in chronic myeloproliferative, [8] and myelodysplastic syndrome with 5q-syndrome. [9]

As essential thrombocythemia is a very rare disease. In our first case patient was treated from different general practitioners as a routine case of headache due to migraine and hypertension. When she presented to Capital Development Authority Hospital, a tertiary care center, her diagnosis of essential thrombocythemia was made. This case is reported because it is not a common disease and to demonstrate that routine examination of blood complete picture is very important. Routine investigation like blood complete picture is often missed. We incidentally diagnosed the cause of headache as thrombocythemia.

Our second case presented with stroke, facial palsy and gangrene of the hand. It is one of the complication of thrombocythemia. Thrombocytosis is typically discovered as an incidental laboratory abnormality when the complete blood count is obtained for some unrelated reason. [8]

We made diagnosis on the basis of exclusion criteria in which we excluded all causes of reactive thrombocythemia by appropriate investigations. We followed the revised diagnostic criteria for essential thrombocytosis which were proposed in 2005.[10] Due to the lack of a specific molecular marker, the diagnosis of essential thrombocythemia can only be settled after a step-by-step elimination of the other clinical situations associated with a protracted elevation of the platelet number.[11] Although we were unable to perform cytogenic studies but our cases satisfy the revised diagnostic criteria of essential thrombocythemia. Numbness of feet is due to vasospasm in our first case. We treated our patients with aspirin and hydroxyurea, but second case was also treated in the departments of surgical, neurological and rehabilitation for gangrene and stroke respectively. Follow up of blood complete picture shows that it reduced the platelet count and patient showed much improvement. Hydroxyurea is effective in preventing thrombosis in high-risk patients with essential thrombocythemia. [7] The new agent Anagrelide has recently been introduced for the treatment of essential thrombocytosis. However, recent studies showed that Anegrilide is not significantly more effective than traditionally used hydroxyurea. [6]

This case report shows that thrombocythemia can present with mild (headache) to sever (stroke and gangrene) sign and symptoms. Headache should not be considered as a casual symptom. Proper work up is very important to prevent under-diagnosis and complications of this rare disease.

References

1. Tefferi, A. The Philadelphia chromosome negative chronic myeloproliferative disorders: a practical overview. *Mayo Clin Proc* 1998; 73:1177.
2. Harrison, CN. Current trends in essential thrombocythaemia. *Br J Haematol* 2002; 117:796.
3. Passamonti, F, Rumi, E, Pungolino, E, et al. Life expectancy and prognostic factors for survival in patients with polycythemia vera and essential thrombocythemia. *Am J Med* 2004; 117:755.
4. Rozman, C, Giralt, M, Feliu, E, et al. Life expectancy of patients with chronic nonleukemic myeloproliferative disorders. *Cancer* 1991; 67:2658.
5. Fenaux, P, Simon, M, Caulier, MT, et al. Clinical course of essential Thrombocythemia in 147 cases. *Cancer* 1990; 66:549.
6. Harrison CN, Campbell PJ, Buck G, Wheatley K, et al.Hydroxyurea compared with anagrelide in high- risk essential thrombocythemia. *N EJ M* 2005; 353:33-45.
7. Sergio Cortelazzo, Guido Finazzi, Marco Ruggeri, OscarVestri, et al. Hydroxyurea for Patients with Essential Thrombocythemia and a High Risk of Thrombosis. *NEJM* 1995; 332:1132-1137.
8. Schafer AI. Thrombocytosis. *N Engl J Med* 2004; 350:1211-9.
9. List AF, Avery A, Doll DC, Sandberg. Myelodysplastic syndromes. In: Greer JP, Foerster J, Lukens JN, Rodgers GM, Paraskevas F, Glader B, editors. Wintrobe's clinical hematology. 11 th ed. Philadelphia, PA: Lippincott Williams and Wilkins; 2004. p. 2207-34.
10. Wintrobe's clinical hematology. 11 th Ed. Philadelphia, PA: Lippincott Williams and Wilkins; 2004. p. 2207-34.
11. Campell PJ, Green AR (2005). "Management of polycythemia vera and essential thrombocythemia". Hematology Am Soc Hematol Educ Program 2005: 201–8. doi:10.1182/asheducation-2005.1.201. PMID 16304381.
12. Murphy S, Peterson P, Illand H, Lazlo J. Experience of the Polycythemia Vera Study Group with essential thrombocythemia: a final report ondiagnostic criteria, survival, and leukemic transition by treatment. *Semin Hematol.* 1997; 34: 29–39.