Original Research Paper

Volume - 10 | Issue - 12 | December - 2020 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Internal Medicine

A LATENT FORM OF ESSENTIAL THROMBOCYTHEMIA WITH PORTAL VEIN THROMBOSIS-RARE PRESENTATION

Vishnu shankar* MD(Internal Medicine) Final Year Post Graduate *Corresponding Author

N. Senthil MD(Internal Medicine), Professor

ABSTRACT Essential thrombocythemia is frequently associated with abdominal thrombotic complications .We report a case of 65 years old male presented with complaints of abdominal distension for 1 month and abdominal pain ,nausea,vomiting, for past 5 days.On clinical examination patient found to have ascites and splenomegaly . CT abdomen showed chronic portal vein thrombosis with multiple collateral formation ,subsequently patient underwent endoscopy which showed Grade II large esophageal varices .Patient had increased platelet since admission inspite of hypersplenism,hence bone marrow biopsy was done which showed hypercellular reactive marrow showing trilineage hematopoiesis with increase in megakaryocytes.JAK 2 mutation turned to be positive ,hence diagnosed as Essential thrombocythemia.Treated with T.Hydroxyurea

KEYWORDS : Essential Thrombocythemia, Portal Vein Thrombosis, JAK2 Mutation

INTRODUCTION

Essential thrombocythemia(ET) is one of the chronic myeloproliferative neoplasm characterized by clonal proliferation of myeloid cells. Essential thrombocythemia is also called Essential thrombocytosis characterized by excessive clonal platelet production with tendency for thrombosis and haemorrhage. Epidemiologic studies shows an annual incidence rate for Essential Thrombocythemia of 1 to 2.5 new cases/1,00,000 population per year.Life expectancy appears to be near normal in ET We report a case of latent form of Essential thrombocythemia with poratl vein thrombosis. In this the diagnosis of essential thrombocythemia was initially missed because the typical disease phenotype was masked by hypersplenism. The correct diagnosis was only reached when patient had persistent thrombocytosis.

CASE REPORT

A 65 year old male came with complaints of abdominal distension for 1 month followed abdominal pain ,nausea ,vomiting for past 5 days. No known comorbidities were present. He was initially evaluated in local hospital,took a CT abdomen which shows chronic portal vein thrombosis, splenomegaly, referred here for further management.

On admission patient vitals were stable ,pallor present .clinical examination of abdomen showed multiple pigmented spots over abdonibal wall,moderate splenomegaly (5cm below costal margin)with fluid thrill present.Other system examination were normal.

Routine blood investigation haemoglobin of 8.1g/dl,Hematocrit of 28% and platelet count of 4.79 lakh.Liver function and Renal function were with in normal limits.

Ultrasound abdomen with portal vein doppler shows Non visualization of portal vein with formation of multiple collaterals confirming the cavernous transformation of portal vein, splenomegaly (19.2cm), moderate ascites.

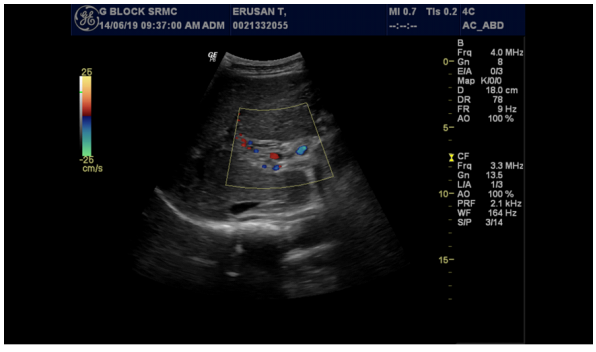

Figure1,ultrasound image of chronic portal vein thrombosisi

Medical gastro opinion obtained for endoscopy which showed Large Grade II esophageal varices,fundal varices for which EVL was done.

Peripheral smear done shows microcytic hypochromic anaemia with anisopoikilocytosis showing pencil shaped and tear drop cells.Treated with Injection Iron sucrose 200mg IV OD for 3 days. ,repeat haemoglobin after 1 week improved to 8.9g/dl with platelet count of 5.7 lakhs.

Alpha-fetoprotein(AFP) and Carcinoembryonic antigen(CEA) were send to rule out malignancy and turned out to be negative.

Since patient had persistent thrombocytosis inspite of hypersplenism, hence bone marrow biopsy was done to rule out myeloproliferative disorder.It shows hypercellular reactive marrow showing trilineage hematopoiesis with diffuse increase in number of megakaryocytes.

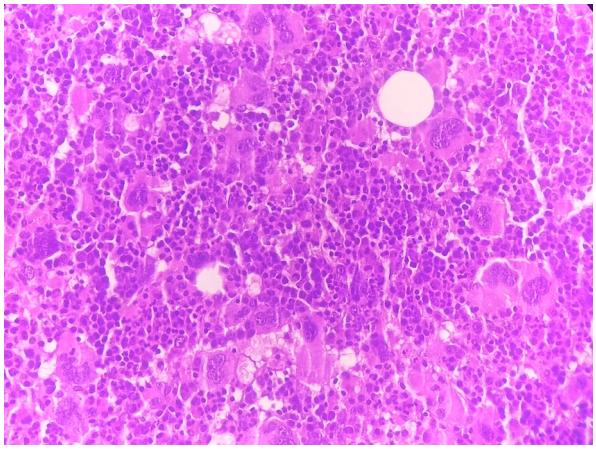

Figure2,Bone marrow biopsy

sample sent for JAK 2 mutation analysis was positive. Diagnosis of Essential thrombocythemia was established .Started on T. Hydroxyurea 500mg OD under close hematological monitoring. Started on T.Aspirin 75mg OD for chronic portal vein thrombosis.

DISCUSSION

Upto one half of patients with Essential thrombocythemia are discovered incidentally when thrombocytosis noted on complete blood count. Others present with disease related symptoms(eg, headache, dizziness,visual changes)or complications (eg, thrombosis, bleeding first trimester fetal loss). Approximately 90% percent of cases have somatically acquired driver mutation in JAK2, CALR or MPL.

Essential thrombocythemia is frequently associated thrombotic complications in the large abdominal vessels.ET carries the best prognosis among the MPD ,but abdominal vein thrombosis identified as a risk factor for poor survival.

Treatment options include aspirin(or an equivalent therapy)and cytoreductive therapy to control the platelet count such as

hydroxyurea, anagrelide and interferon,however there is still lot of controversy regarding the role of anticoagulation in patients with chronic portal vein thrombosis.

In our case,the diagnosis of Portal vein thrombosis was made initially,MPD was not considered because the platelet was only slightly elevated,so reactive thrombocytosis due iron deficiency anaemia was considered.Since platelet count kept on increasing after treating anaemia ,suggested that peripheral blood counts are masked by hypersplenism.Discovery 0f Mutation in V617F of the Janus Kinase2 gene provides a new molecular tool.

CONCLUSION

For patients with portal vein thrombosis ,if no common causes ,if no common causes such as cirrhosis and neoplasm are found, investigations into an uncommon cause of MPD is indicated using bone marrow biopsy and JAK2 mutation.It is important to realize that features of Essential thrombocythemia in peripheral blood could be masked by hypersplenism .

REFERENCES

1. Jensen MK, de Nully Brown P, Nielsen OJ, Hasselbalch HC. Incidence, clinical features and outcome of essential thrombocythaemia in a well defined geographical area. Eur J Haematol 2000; 65: 132-139
2. Ogren M, Bergqvist D, Björck M, Acosta S, Eriksson H, Sternby NH. Portal vein thrombosis: prevalence, patient characteristics and lifetime risk: a population study based on 23,796 consecutive autopsies. World J Gastroenterol 2006; 12: 2115-2119
3. Denninger MH, Chaït Y, Casadevall N, Hillaire S, Guillin MC, Bezeaud A, Erlinger S, Briere J, Valla D. Cause of portal or hepatic venous thrombosis in adults: the role of multiple concurrent factors. Hepatology 2000; 31: 587-591
4. Valla DC, Condat B. Portal vein thrombosis in adults: pathophysiology, pathogenesis and management. J Hepatol 2000; 32: 865-871
5. Gangat N, Wolanskyj AP, Tefferi A. Abdominal vein thrombosis in essential thrombocythemia: prevalence, clinical correlates, and prognostic implications. Eur J Haematol 2006; 77: 327-333
6. Landolfi R, Di Gennaro L, Falanga A. Thrombosis in myeloproliferative disorders: pathogenetic facts and speculation. Leukemia 2008; 22: 2020-2028
7. Harrison CN. Essential thrombocythaemia: challenges and evidence-based management. Br J Haematol 2005; 130: 153-165
8. Harmanci O, Bayraktar Y. Portal hypertension due to portal venous thrombosis: etiology, clinical outcomes. World J Gastroenterol 2007; 13: 2535-2540