RESEARCH PAPER | Medical Science | Volume : 5 | Issue : 11 | November 2015 | ISSN - 2249-555X

Acute Abdomen – "An Unusual Medical Cause"

KEYWORDS

Sharol Ashma Menezes
Senior Resident, Department of Medicine, Father Muller Medical College, Kankanady, Mangalore Karnataka. PIN-575002

K Sundara Bhat
Professor of Medicine, FMMC, Mangalore, Karnataka. PIN-575002

ABSTRACT *Paroxysmal nocturnal haemoglobinuria is a rare acquired clonal disorder of hematopoietic stem cells characterized by increased susceptibility of erythrocytes to complement-mediated haemolysis. PNH is characterized by intravascular haemolysis, thromboembolic episodes and bone marrow failure1. Thromboembolism is a life-threatening complication which occurs in approximately one-third of PNH patients1,2,3. Sites affected are often at atypical localization. They most commonly involve abdominal veins such as hepatic, splenic, mesenteric veins, portal and inferior vena cava, and cerebral veins1,3,4.*

We report the rare case of an adult male, who presented with abdominal pain and was found to have paroxysmal nocturnal haemoglobinuria.

Introduction-
Paroxysmal nocturnal haemoglobinuria is a rare, potentially life threatening disease. The triad of hemolytic anemia, pancytopenia, and thrombosis makes paroxysmal nocturnal hemoglobinuria a truly unique clinical syndrome[1,2,3,4].

This disease has been referred to as the great impersonator because of the variety of symptoms observed during the initial manifestation and course of paroxysmal nocturnal hemoglobinuria.

We report the rare case of an adult male, who presented with abdominal pain and was found to have paroxysmal nocturnal haemoglobinuria.

Clinical details
A 50 yr old male patient presents with history of pain abdomen, passage of black coloured stool and loss of appetite since 2 weeks. He had history of chronic liver disease with portal hypertension and portal gastropathy with no history of alcohol consumption. He had pallor and icterus on general examination. He did not have pedal edema. Per abdomen examination revealed diffusely tender abdomen with tender palpable liver and a nontender palpable spleen. There was no shifting dullness.

Investigations
Hemoglobin-9.7 g /dl
Total Leucocyte Count -3200 cell/mm3
Differential Count -N69% L28% E1% M2%
ESR 9 mm
Platelets: 24,000 cells/mm3
Reticulocyte% - 5%
Prothrombin Time - 10.6/16.1 s (control/test)
INR- 1.63
APTT- 23/26 s (control/test)
HIV/HBSAG-NEGATIVE
Electrolytes were normal
(Na-132mEq/L, K-4.2mEq/L, Cl-95mEq/L)
Urea-22 mg/dl, Creatinine - 0.8 mg/dl
Fasting Blood Glucose - 95 mg/dl
S.albumin - 3.5 g/dl, S.Globulin-2.5 g/dl, A/ G:1.4
S.bilirubin:3.9mg/dl, DB:1.2mg/dl, IB:2.7mg/dl
SGOT-72 U/L, SGPT-90 U/L, ALP-156 U/L
S.Amylase-39 U/L S.Lipase-24U/L
LDH-815 U/L
Urine for blood-negative

USG abdomen: Chronic parenchymal liver disease with portal hypertension, gall bladder calculi, mild ascites, diffusely oedematous small bowel loops ? Secondary to ischemia

UGI endoscopy report: UGI bleed from portal gastropathy

CT scan of abdomen and pelvis: Chronic parenchymal liver disease with portal hypertension. Venous thrombosis at superior mesenteric vein and splenic vein confluence with significant luminal narrowing. Jejunal and ileal wall edema. Cholecystolithiasis.

Peripheral Smear

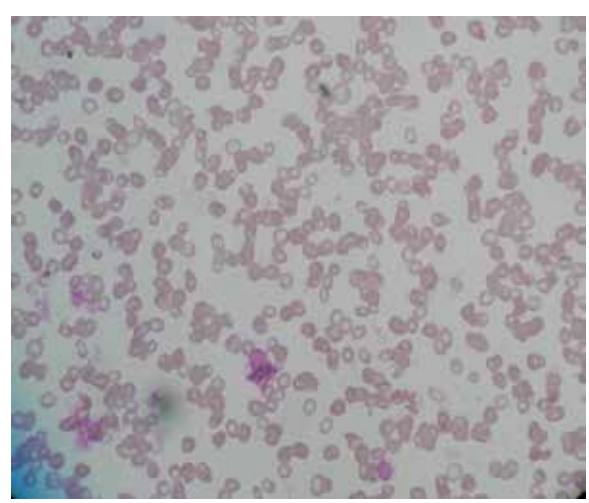

Figure 1: Peripheral smear examination showing pancytopenia

RBCs are predominantly normocytic normochromic. Moderate degree of anisocytosis is present with presence of scattered macrocytic and microcytic hypochromic cells and occasional polychromatophils

Volume : 5 | Issue : 11 | November 2015 | ISSN - 2249-555X

WBCs were reduced in number.

Differential count shows: Neutrophils-61%, Lymphocytes-35%, Monocytes-03%, Eosinophils- 01%

Platelets showed marked reduction in number.

No hemoparasite/ abnormal cells seen on smears studied.

Impression: Pancytopenia

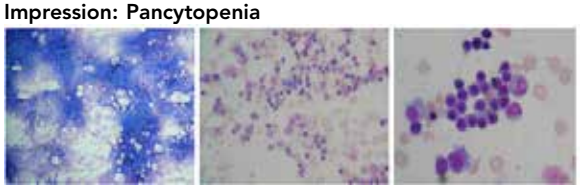

Figure 2: Showing the bone marrow aspiration in scanner view (left), low power view (middle) and oil emersion view (right)

Material- Adequate

Cellularity – Mildly hypercellular marrow with good cell trails

M:E ratio – 0.14:1

Erythropoesis : increased and shows normoblastic maturation

Myelopoesis- normal and show orderly maturation sequence.

Megakaryocytes- normal in number and morphology.

Lymphoplasmacytes- within normal limits.

No Atypical/ Granuloma / Parasite seen in smear studied.

Impression : Mildly hypercellular marrow with normoblastic erythroid hyperplasia.

Bone marrow biopsy:

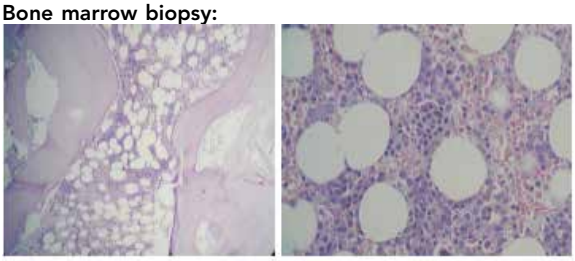

Figure 3: Showing bone marrow biopsy under low (left) and high (right) power fields

Section studied shows **mildly hypercellular marrow spaces** enclosed in mature bony trabaculae. The **erythroid series show hyperplasia with normoblastic maturation**. The other haematopoietic elements (myeloid and megakaryocyte) are within normal limits. No granuloma seen.

Impression: Mildly hypercellular marrow with erythroid hyperplasia

In view of the clinical picture showing pancytopenia, raised LDH, serum bilirubin and occurrence of thrombosis at unusual sites, a diagnosis of PNH was considered. Flow cytometry studies were asked for.

Flowcytometry report:

Flow cytometry showed evidence of PNH clone based on GPI-linked antibodies on granulocytes and monocytes.

Discussion

Paroxysmal nocturnal hemoglobinuria (PNH) is a descriptive term for the clinical manifestation of red blood cell (RBC) breakdown with release of hemoglobin into the urine that is manifested most prominently by dark-colored urine in the morning.

Paroxysmal nocturnal hemoglobinuria (PNH) has been reclassified from purely an acquired hemolytic anemia due to a hematopoietic stem cell mutation defect

PNH presents with pallor suggesting anemia, bleeding manifestations suggesting thrombocytopenia, hepatomegaly and ascitis in the presence of budd-chiari syndrome and spleenomegaly in the presence of splenic vein thrombosis.

Management:

Anticoagulation

Eculizumab, a humanized monoclonal antibody blocks activation of terminal complement C5 components is currently used in the treatment of PNH patients. Treatment with eculizumab reduces transfusion requirements, decreases the chances of anemia and improves quality of life[5,6].

Allogenic bone marrow transplantation

Considering the age our patient he was started on anticoagulation. He improved symptomatically. His review computed tomogram showed no new thromboses.

Conclusion

The presence of pancytopenic blood picture , haemoglobinuria and large vein thrombosis should prompt us to meticulously evaluate for PNH. All patients with hemoglobinuria, unexplained hemolysis, abdominal or cerebral vein thrombosis, thrombocytopenia or macrocytosis or signs of hemolysis should undergo PNH testing.

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