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Case Report
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THE UNVEILED MYSTERY

Mohammed Shanil P.[1]* and Farsana Jasmin K.[2]

[1]DM Cardiology Resident, Government Medical College, Kannur.
[2]MD Radiotherapy Resident, Government Medical College, Kozhikode.

***Corresponding Author: Mohammed Shanil P.**
DM Cardiology Resident, Government Medical College, Kannur.

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ABSTRACT
Paroxysmal nocturnal hemoglobinuria (PNH) is a rare bone marrow failure disorder that presents with triad of peripheral blood cytopenias, hemolytic anemia and thrombosis. The absence of two glycosylphosphatidylinositol (GPI)-anchored proteins, CD55 and CD59, leads to uncontrolled complement activation that accounts for hemolysis and other PNH manifestations. GPI anchor protein deficiency is due to acquired somatic mutations in phosphatidylinositol glycan class A (*PIGA*) gene. Terminal complement inhibition with eculizumab and allogeneic bone marrow transplantation (BMT) are the only widely effective therapies for patients with classical PNH. Compared to eculizumab and ravulizumab that block the fifth component of complement (C5), pegcetacoplan blocks the third component of complement (C3).

KEYWORDS: Paroxysmal nocturnal hemoglobinuria, glycosylphosphatidylinositol, eculizumab, ravulizumab, pegcetacoplan.

INTRODUCTION
Paroxysmal nocturnal hemoglobinuria (PNH) is a clonal hematopoietic stem cell disorder that manifests with hemolytic anemia, bone marrow failure, and thrombosis. In 1882, PNH was first described by Dr Paul Strübing and Enneking, in 1925, introduced the term paroxysmal nocturnal hemoglobinuria. In 1937, Thomas Ham reported that PNH erythrocytes were hemolyzed when incubated with normal, acidified serum. This seminal discovery resulted in the first diagnostic test for PNH, the acidified serum (Ham) test. Abdominal pain, esophageal spasm, dysphagia, and erectile dysfunction are common symptoms associated with classical PNH and are a direct consequence of intravascular hemolysis. The diagnosis is usually challenging mystery since it is masked by a variety of symptoms.

CASE
A 23 year-old lady was admitted to our hospital with exertional dyspnea and fatiguability for 2 months followed by recurrent headache for 1 week. During the course in the hospital, she had seizures and severe abdominal pain with high coloured urine. General examination revealed severe pallor and Vitals within normal limits.

OBSERVATION
Routine blood investigations showed pancytopenia (Hb-4.5, Total count--2400, platelet count-30k), high ESR (80mm/hr) and unconjugated hyperbilirubinemia. Additional investigations showed raised LDH and microcytic hypochromic anemia with poikilocytosis in peripheral smear. She was given immediate 2 pints of blood transfusion .MRI brain showed filling defect involving straight sinus extending to right transverse sinus with post contrast meningeal enhancement with diagnosis of Cerebral venous sinus thrombosis. Flow cytometry showed deficiency of CD55 and CD 59. Thus definite diagnosis of PAROXYSMAL NOCTURNAL HEMOGLOBINURIA confirmed. During the course in the hospital, she was counselled and treated with Warfarin by monitoring INR along with iron tablets thus showing clinical improvement on follow-up.

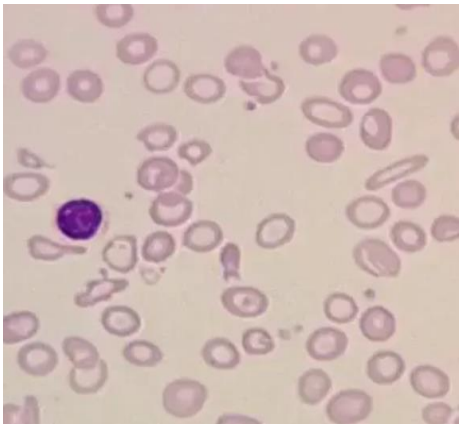

Figure 1: Microcytic Hypochromic Anemia.

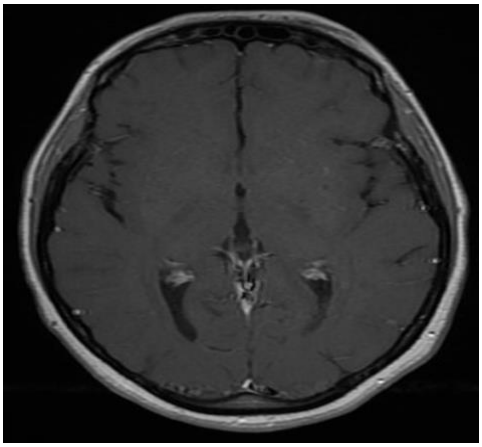

Figure 2: Mri-T1w1-Straight Sinus Thrombosis.

CLINICAL SIGNIFICANCE

Physicians should have a high index of suspicion for Paroxysmal nocturnal hemoglobinuria while evaluating patient with features of anemia and venous thrombosis. Terminal complement inhibition with eculizumab and allogeneic bone marrow transplantation (BMT) are the only widely effective therapies for patients with classical PNH.

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