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Case Report

Paroxysmal nocturnal haemoglobinuria presented as acute kidney injury

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ABSTRACT

Paroxysmal nocturnal hemoglobinuria (PNH) is rare disease, caused by acquired somatic mutations in PIG-A gene. Renal involvement in PNH varies from reversible acute kidney injury (AKI) to chronic irreversible damage. We are presenting a case of PNH in young female who presented as AKI. Final diagnosis was made by flowcytometry which showed evidence of PNH clone based upon analysis of a variety of GPI linked antibodies (FLAER, CD59) on monocytes, granulocytes and RBCs. PNH presenting as recurrent acute renal failure is extremely rare. We reported this case to highlight a rare, but potentially reversible cause of acute renal failure. These types of cases need high index of suspicion. Early diagnosis and treatment will help in preventing repeated episodes of AKI and thus chronic kidney disease.

Keywords: Acute kidney injury, Flow cytometry, Paroxysmal nocturnal hemoglobinuria

INTRODUCTION

Paroxysmal nocturnal hemoglobinuria (PNH) is a rare acquired disease, caused by non-malignant clonal expansion of one or more hematopoietic stem cells that acquired somatic mutations in PIG-A gene linked to chromosome X. This mutation results in lower erythrocyte expression of CD55 and CD59 surface proteins and consequently increased susceptibility to the complement system. Renal involvement in PNH is not usually apparent and varies from reversible acute kidney injury (AKI) to chronic irreversible damage. Not many reports of renal involvement in PNH are available in literature.[1,2] We report a case of PNH who presented as AKI.

CASE REPORT

An 18-year-old girl presented with history of fever which was acute in the onset high grade associated with diarrhoea, nausea and vomiting for four days. She also noticed decreased urine output in last three days. It was associated with decrease appetite. There was no history of abdominal pain, skin rashes, reddish urine, joint pains, sore throat or intake of any medications. Her menstrual cycle was regular but having menorrhagia from last 2 months. She gave past history of fever and vomiting followed by decreased urine output 9 months back. She received IV fluids and three units of blood at that time but further details not available. There was no history of diabetes or hypertension. On examination, patient was conscious and oriented. She had pallor and fever of 102°F. There was no icterus, clubbing, cyanosis, bony tenderness, lymphadenopathy or pedal edema. Her Blood pressure was 110/76 mm Hg, respiratory rate 20/min, pulse regular 110/min and SPO_2 98%. In respiratory system, B/L air entry was present with decrease air entry in right basal area, per abdomen was soft with no hepatosplenomegaly. On investigation, urine examination showed specific gravity 1.010, Ph: 5.0 and protein traces, pus cells: 2-3 pus cells per high power field, RBC:10-13 per high power field, and epithelial cells 0-1 per high power field. Cast or crystal not seen. Her hemoglobin was 4.5g/dl, total leucocyte count 3.0×10^3/µl, platelet count 91.0×10^3/µl and picture normocytic normochromic. No polychromasia or

nucleated RBCs seen the peripheral smear. Reticulocyte count was 2.2%.

Renal function test (RFT) revealed urea 120 mg/dl, creatinine 3.8 mg/dl, serum potassium 5.8 meq/l, serum sodium 133.0 meq/l, serum calcium 9.1 mg/dl and serum phosphorus 3.3mg/dl. Liver function test (LFT) showed aspartate aminotransferase 87 IU, alanine aminotransferase 72 IU, alkaline phosphatase 125U/l, total protein 6.5 g/dl, serum albumin 3.8 g/dl, total bilirubin 1.8 mg/dl with indirect bilirubin 1.3 mg/dl. On aerobic culture blood no growth was seen after 48 hours incubation at 37° Celsius. On radiological investigations, CECT (Chest and Abdomen) showed mild right hydropneumothorax and minimal left pleural effusion with basal atelectasis. In abdomen, it showed slightly bulky right kidney with few wedge shape hypodensities with minimal hydronephrosis with prominent ureter with enhancing wall. Minimal perinephric fat stranding was also seen with thickening of pararenal fascia? acute pyelonephritis. Liver was enlarged in size (span 20.0cm) and showed normal homogenous attenuation with no focal lesion. The intrahepatic biliary radicals were not dilated. Portal vein was normal. No ascites was seen.

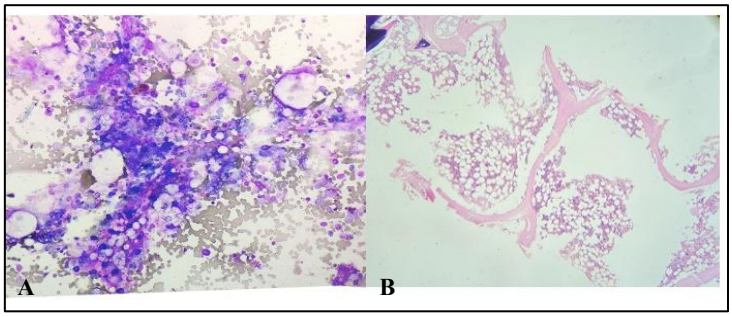

Figure 1: (A) bone marrow aspirate revealing low cellularity with serous atrophy (Leishman stain 20X) and (B) bone marrow biopsy section comprised of hypocellular marrow (H and E 5X).

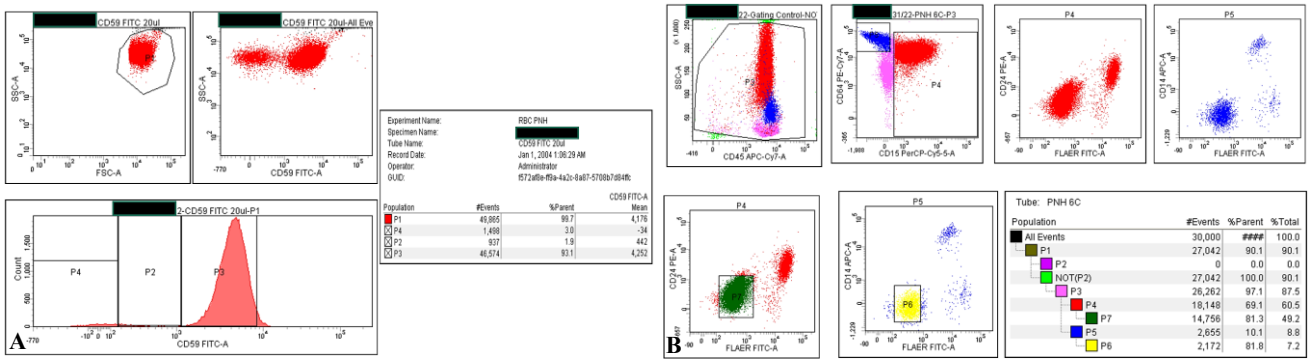

Figure 2 (A): flowcytometric image of RBCS: SSC versus CD59 gating dot plot and histogram showed reduced/ absent expression of CD 59 in RBCS and (B): SSC vs CD45 vs CD15 and CD64 showed reduced/ absent expression of FLAER in neutrophils and monocytes.

She was managed conservatively by giving IV fluids, 4 packed red cell transfusion along with steroids and antibiotics. RFT started normalizing in one week (urea: 60 mg/dl, creatinine: 1.8 mg/dl, serum potassium: 4.5 meq/l, serum sodium 138.0 meq/l). Meanwhile patient was investigated further for cause of acute renal failure. Pleural fluid was reddish colour and turbid.

Total protein was 2.8g/dl traces, glucose 84.0 mg/dl, chloride 106.0meq/l, globulin high elevation with cell counts $0.45\times10^3/\mu l$ with predominance of lymphocytes (95%). No microorganism seen on Gram stain. Pleural fluid adenosine deaminase (ADA) was 7.00U/l. Coagulation profile, osmotic fragility, hemoglobin electrophoresis and G6PD levels were normal. Direct and indirect coomb's tests were negative. Thyroid profile was within the normal limits. Serum Anti-Nuclear Antibody/ factor (ANA/ANF) by the IFA (HEP-2) end point revealed ANA Hep-2 positive; pattern cytoplasmic discrete dots, intensity 1+, primary titer/ dilution 1:100 and end point titer/ dilution:100. Viral markers were negative, C peptide fasting was 10.68 ng/ml, folate 15.47 ng/ml and tissue transglutaminase antibody 0.77 units. Bone marrow aspiration and biopsy revealed hypoplastic marrow with no iron stores (Grade 0) (Figure 1: A and B). In view of no apparent cause sample for PNH was processed. Flow cytometry analysis showed evidence of PNH clone based upon analysis of a variety of GPI linked antibodies on monocytes, granulocytes and RBCs as there is absent expression of FLAER (Proaerolysin conjugated with fluorescein) in 82.8% monocytes and 83.3% granulocytes. 6-9% RBC's showed absent expression of CD59 (Figure

2: A and B). Finally, the patients were diagnosed as case of PNH with acute gastroenteritis with acute kidney injury with right sided hydropneumothorax.

DISCUSSION

The main clinical manifestations of PNH are related to abnormalities in hematopoietic function including haemolytic anaemia, hypercoagulability, bone marrow aplasia or hypoplasia, and progression to myelodysplasia and/or acute leukaemia. The occurrence of acute and chronic kidney injury are observed in recurrent haemolysis as observed in PNH.[2] The renal involvement is generally benign, ranging from concentration defects to acute renal failure requiring dialysis and to chronic kidney disease. Intravascular haemolysis in PNH lead to two types of renal disease. Acute kidney injury by sudden acute haemolytic episode associated with massive hemoglobinuria may occur due to any cause (often in association with gastroenteritis). The mechanisms behind this phenomenon include release of heme which is liable iron, toxic to tubular cells and cause acute tubular necrosis, hypovolemia, nitric oxide depletion leading to renal ischemia, oxidative stress by reactive oxygen species, mitochondrial membrane damage and tubular obstruction by uric acid crystals. Rarely AKI may present with microscopic haematuria. Chronic haemolysis results in iron deposition in the kidneys leading to chronic kidney disease.[3-7]

In our case a young female, with history of fever, diarrhea, decreased urine output, and menorrhagia, normocytic normochromic anemia with normal reticulocyte count, hypoplastic marrow with absent iron stores, mild increase in indirect bilirubin, negative coomb's test and nonspecific autoimmune markers, the case was discussed for all the possible causes. Finally sample for PNH by flowcytometry was processed which showed evidence of PNH clone. Flow cytometry to study the expression of CD55, CD59 and FLAER is the choice of test because of its high sensitivity and specificity in confirming the diagnosis of PNH. The renal biopsy of these cases may show acute tubular necrosis and intense deposition of hemosiderin in renal tubular cells.[6-8] Although we have not performed a renal biopsy in our case, we believe that the patient had tubular injury secondary to haemolysis. Some studies implicated renal failure as the leading cause of death in 8–18% PNH patients while other studies report that renal failure is responsible for an eight-fold increase in mortality. Despite its frequency, renal dysfunction' pathophysiology is still elusive and is often neglected by physicians when treating PNH patients.[7-9]

CONCLUSION

PNH presenting as recurrent acute renal failure is extremely rare. Reported to highlight a rare, but potentially reversible cause of acute renal failure. This type of case needs high index of suspicion. Early diagnosis and treatment will help in preventing repeated episodes of AKI and thus chronic kidney disease.

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