A CASE OF PAROXYSMAL NOCTURNAL HEMOGLOBINURIA WITH MYELODYSPLASTIC SYNDROME

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Paroxysmal Nocturnal Hemoglobinuria (PNH): Rare acquired clonal hematopoietic stem cell disorder Caused by PIGA gene mutation ’! deficiency of CD55 & CD59 Leads to complement-mediated intravascular hemolysis Strong association with bone marrow failure syndromes, especially Myelodysplastic Syndrome (MDS). A 45-year-old male farmer 10-year history of recurrent anemia requiring repeated transfusions Presenting complaints: Generalized weakness (1 month) Early morning dark urine (15 days) Dysuria & frequency. Key Clinical Features suggested that chronic transfusion-dependent anemia Hemoglo-binuria (dark urine, RBC absent in urine) No bleeding, jaundice, or systemic illness. On examination patient had mild anemia. No organomegaly aqnd vitals stable. Clinical Problem List Recurrent anemia (10 years). Repeated blood transfusions Hemoglobinuria Suspected hemolysis, possible marrow pathology. Key investigations reavealed that Hb: 3.7–9.8 g/dL (chronic severe anemia) reticulocyte count: 5.17% LDH: Markedly elevated (3250 U/L), Coombs test: Negative, excludes autoimmune hemolysis which suggests that non-immune intravascular hemolysis. Urine Findings reddish urine no RBCs and hemoglobinuria which supports PNH diagnosis. Peripheral blood film showed that normocytic anemia anisocytosis No blasts Bone Marrow Study Hypercellular marrow Dysplasia in >10% cells Erythroid & megakaryocytic abnormalities, consistent with myelodysplastic Syndrome (MDS). Flow Cytometry (Key Diagnostic Test) FLAER (WBC panel): Negative RBC panel: CD55 deficiency: ~25% CD59 deficiency: ~25% confirmed PNH CLONE Final Diagnosis was Paroxysmal Nocturnal Hemoglobinuria (PNH) and Associated Myelodysplastic Syndrome (MDS) and Urinary Tract Infection Important Clinical Insights were PNH should be suspected in Coombs-negative hemolytic anemia hemoglobinuria without RBCs cytopenias or marrow failure strong overlap with: Aplastic anemia. Not all anemia with dark urine is hematuria—think hemoglobinuria and PNH. Early suspicion + targeted testing = life-saving diagnosis

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