A RARE PRESENTATION OF SICKLE CELL ANEMIA AS BILATERAL AVASCULAR NECROSIS OF HIP JOINT IN A YOUNG FEMALE

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***Abstract*--Avascular necrosis (AVN) is a condition that occurs due to obstruction of blood flow to the joints, resulting from various causes like embolic, thrombotic or traumatic events . However Sickle cell disease (SCD) is an underrecognized yet significant cause of AVN occurring in approximately 10% of scd patients .This condition arises from a state of decreased blood supply to hip joint due to abnormally sickle shaped red blood cells undergoing destruction in spleen and hence decreased oxygen carrying capacity and oxygen supply to the hip joints. SCD is one of the most common causes of hemoglobinopathies worldwide.(1) This case report presents an unusual instance of AVN of hip joints as the initial presentation of SCD ,implying the importance of investigating for hemoglobinopathies in patients presenting with unexplained AVN.**

I. INTRODUCTION

Sickle cell disease (SCD) refers to a group of hemoglobinopathies that include mutations in the gene encoding the beta subunit of hemoglobin. The first description of SCA 'like' disorder was provided by Dr. Africanus Horton in his book The Disease of Tropical Climates and their Treatment (1872). However, it was not until 1910 when Dr. James B Herrick and Dr. Ernest Irons reported noticing 'sickle-shaped' red cells in a dental student (Walter Clement Noel from Grenada).[2]In 1949, independent reports from Dr. James V Neel and Col. E. A. Beet described the patterns of inheritance in patients with SCD. In the same year, Dr. Linus Pauling described the molecular nature of sickle hemoglobin (HbS) in his paper 'Sickle Cell Anemia Hemoglobin.' Ingram Vernon, in 1956, used a fingerprinting technique to describe the replacement of negatively charged glutamine with neutral valine and validated the findings of Linus Pauling.[3] . Bone involvement is the commonest clinical manifestation of sickle cell disease both in the acute setting such as painful vaso-occlusive crises, and as a source of chronic, progressive disability such as avascular necrosis. Management of these problems is often difficult because of the diagnostic imprecision of most laboratory and imaging investigations and because of the lack of evidence for most surgical procedures in sickle cell disease.(4) This case report presents an unusual instance of AVN of hip joints as the initial presentation of SCD ,implying the importance of investigating for hemoglobinopathies in patients presenting with unexplained AVN.

II. CASE REPORT

A 29-year-old female presented with bilateral hip joint pain - increased severity with standing position and movement ,ocassional sob grade 2-3 and fatigue. Examination revealed a thin built individual with high volume Pulse and normal rate. Pallor was present. systemic examination revealed haemic murmur. Other examination findings were normal. Laboratory tests showed anemia with:Hb: 8.1 g/dL, hct -24.2, Mcv- 68 and peripheral smear - predominantly microcytic hypochromic with moderate anisopoikilocytosis, with few pencil cells ,target cells and nucleated rbc. Sickling test- positive . ultrasonography of abdomen and pelvis showed autospleenectomy. Further evaluation for hb electrophoresis identified HbF - 14.2, HbA -11.1, HbA2-3.6. The patient was diagnosed as Sickle cell anemia with beta thalassemia trait. Treatment with hydroxyurea was initiated, followed by pneumococcal vaccines for preventive measures.

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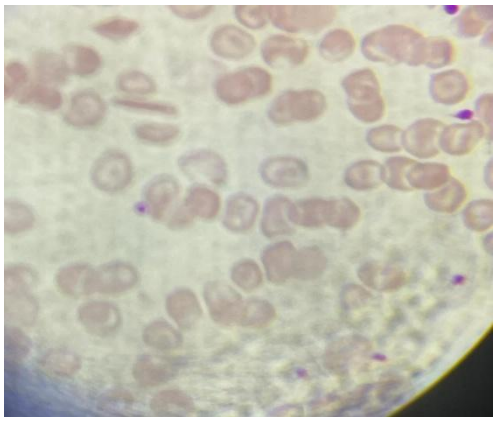

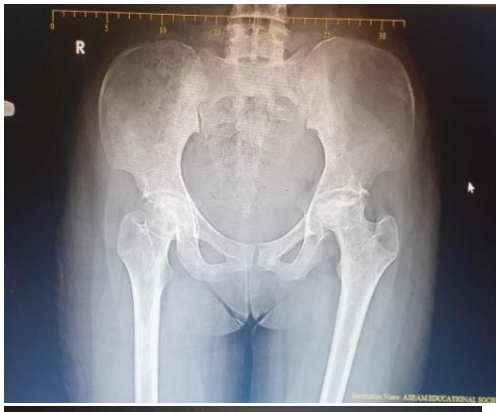

III. DISCUSSION

Sickle cell disease (SCD) is an inherited disorder of hemoglobin, characterized by formation of long chains of hemoglobin when deoxygenated within capillary beds, resulting in sickle-shaped red blood cells, progressive multiorgan damage, and increased mortality. (5) After building an electrophoresis machine, Pauling was able to separate normal adult hemoglobin (α2β2, HbA) from abnormal sickle hemoglobin ($\alpha 2\beta 2^S$, HbS) and describe SCD at a molecular level for the first time. Our understanding of sickle pathophysiology has also been greatly helped by the use of humanized sickle mouse models, which has provided new insights on adhesion, inflammation, and interactions of the sickled RBCs with their microenvironment—vasculature, neutrophils, monocytes, platelets, and the upregulation of vasculature cyto-adhesion molecules.Molecules such as P- and E-selectin, fundamental in the adhesion and activation of white blood cells, specially neutrophils, to the vasculature have been found to represent an important component of the pain crisis pathophysiology and have become therapeutic targets. (6) Avascular necrosis , infantile dactylitis, leg ulcers , sickle chronic lung disease , renal failure, retinopathy predicted an increased likelihood of developing a more lethal form of organ damage and earlier death End-stage renal disease (glomerulosclerosis), chronic pulmonary disease with pulmonary hypertension, retinopathy, and cerebral microinfarctions are manifestations of arterial and capillary microcirculation obstructive vasculopathy (7). Sickle cell disease is one of the most common causes of hemoglobinopathies worldwide.this case report presents an unusual instance of AVN of hip joints as initial presentation of SCD, implying the importance of investigating for hemoglobinopathies in patients presenting with unexplained AVN. This patient was diagnosed as sickle cell anemia with beta-thalassemia trait. Treatment with hydroxyurea was initiated(8), followed by pneumococcal vaccines for preventive measures.other treatment options include total hip replacement or bone reconstruction surgeries if activities of daily living are affected causing significant functional disability.(9)

IV.CONSET

The consent was taken and signed by respective patient.

V. CONFLICTS OF INTEREST

The authors have no conflicts of interest.

VI. CONCLUSION

This study highlights the importance of early recognition of unusual presentations of AVN with the help of multidisciplinary approach .

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