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HbSE disease: A case report

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Abstract
HbSE disease is a rare hemoglobinopathy characterized by the coexistence of hemoglobin S (HbS) and hemoglobin E (HbE) in a double heterozygous state. This case report highlights the clinical presentation, diagnosis, and implications of HbSE disease in a 37-year-old male with hypertension and type 2 diabetes mellitus. The patient presented with persistent abdominal pain, and subsequent investigations revealed bilateral nephrolithiasis and a left-sided polycystic kidney. Hematological analysis showed mild anemia with microcytosis, and high-performance liquid chromatography (HPLC) confirmed the presence of HbS (56%) and HbE (32.7%). While HbSE disease is often considered a benign condition, it can present with vaso-occlusive complications under stressful conditions. The case emphasizes the importance of screening for hemoglobinopathies in patients with unexplained vaso-occlusive events, anemia, or microcytosis to prevent misdiagnosis and unnecessary investigations. HPLC remains the gold standard for diagnosis. Early detection and appropriate disease management strategies, including family screening, are essential to prevent complications and improve patient outcomes.

Keywords: HPLC, haemoglobin SE disease, anemia

Introduction
Sickle cell-haemoglobin E (HbSE) disease is a hemoglobinopathy. It refers to a double heterozygous state where Hemoglobin E (HbE) exists in combination with haemoglobin S (HbS). This case is presented because of its rarity in the clinical scenario.

Case details
A 37 year old male, known case of hypertension and type 2 diabetes mellitus, on treatment presented with complaints of pain abdomen for 7 months duration. On investigating for abdominal pain, an ultrasonogram of abdomen and pelvis was done and revealed bilateral nephrolithiasis and left sided polycystic kidney. Complete blood count revealed a haemoglobin of 10.8 gm%, RBC count of 4.5 million/cumm, Mean corpuscular volume (MCV) of 73.8 fl, Red cell distribution width of 15.4%. Peripheral smear showed microcytic hypochromic RBCs, with few target cells and occasional polychromatophils. (Fig 1). Sickling test was positive. (Fig 2). High performance liquid chromatography (HPLC) analysis showed HbA of 8.7%, HbE of 32.7%, HbF of 2.7% and HbS of 56%, suggestive of a double heterozygous state for HbS and HbE (Fig 3) The patient was advised family screening to rule out the existence of this hemoglobinopathy in other family members as well.

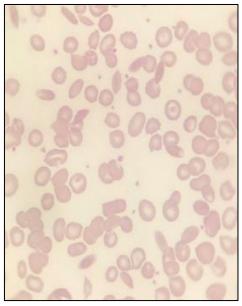

Fig 1: Peripheral smear

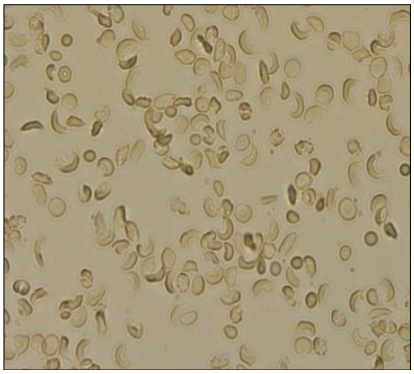

Fig 2: Sickling Test

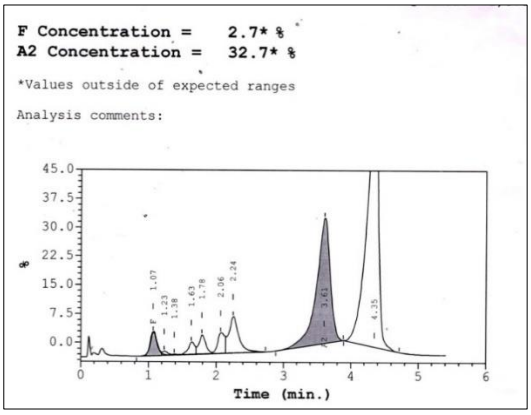

Fig 3: HPLC showing various hemoglobin concentrations

Discussion

HbSE disease is a rare genotypic variant of sickle cell disease [1]. It is usually considered a benign form. Patients with HbSE typically remain clinically asymptomatic till adulthood and may present at a later stage with vasoocclusive crisis in the setting of an acute stress event [2]. Patients may have mild anemia and microcytosis. Coinheritance of HbSE leads to a sickling disorder similar to sickle β thalassemia. HbS results due to substitution pf glutamine to valine, at sixth position in the betaglobin gene.HbE results due to substitution of lysine for glutamine at amino acid number 26, in the beta globin gene. HbSE variant occurs as a result of a combination of HbS and HbE genotypes [3].

Diagnosis of this entity occurs in screening programs or family studies of individuals with HbSE disease, or on routine evaluation of smears with significant microcytosis without anemia. HPLC is the method of choice to quantify various Hb fractions [4].

Conclusion

HbSE is a rare disease. Its knowledge is essential in cases which present with vasoccclusion to prevent misdiagnosis and provide better disease management. Screening for Haemoglobinopathies is recommended in patients with unexplained vasoocclusive events, mild anemia and mild microcytosis to prevent further unnecessary investigations and better patient management.

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