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CASE REPORT

Multisystem Involvement in a Newly Diagnosed Adult with Homozygous Sickle Cell Disease

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Abstract

Background: Sickle cell disease is a haemolytic disorder usually diagnosed in paediatric age group. In regions where proper screening lacks it goes undiagnosed in childhood and presents itself in adulthood with multisystem complications, diagnosis and treatment of these patients in adulthood becomes challenging. The incidence of sickle cell disease Indian adults is ~1.1% and prevalence of sickle cell trait is ~5.9% making it very hard to diagnose and needs a vigilant approach. **Case Presentation:** A 31-yr old female conscious oriented came walking by herself with chief complaints of pain in her b/l upper and lower limbs since 1 week with fatigue and headache since 15 days. No seizure, vomiting or other signs of increased ICP. CT Brain-normal. Lab investigations showed microcytic anaemia, indirect hyperbilirubinemia, positive sickling test and leucocytosis. Haemoglobin electrophoresis confirmed homozygous sickle cell disease. She was treated with hydration, oxygen, analgesic and hydroxyurea. On day 4 she developed acute hypoxemic respiratory failure for which invasive ventilation was needed. 2D Echocardiography revealed moderate pulmonary hypertension with severe tricuspid regurgitation, right atrial and ventricular dysfunction with mild pericardial effusion. Bone marrow aspiration revealed hypercellular marrow with erythroid hyperplasia. Also, on day 7 she developed status epilepticus for which MRI Brain showed acute infarct in rt splenium of corpus callosum in the pericallosal branch of posterior cerebral artery, suggesting embolic stroke also CSF study was normal. With adequate blood transfusions, antibiotic, antiepileptics supportive management patient improved and was discharged in stable condition. **Conclusion:** This case is an example of considering SCD even in adults with Vaso-occlusive symptoms with unexplained anaemia, as it as a much grave and multisystemic complications where diagnosis and treatment becomes challenging and can lead to life threatening events.

Keywords: Sickle cell disease, Pulmonary hypertension, Embolic stroke, Vaso-occlusive crisis, Haemolytic crisis, Thromboembolic crisis

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Introduction

Sickle cell disease (SCD) is an inherited disorder of haemoglobin structure, where there is point mutation in the beta globin gene (chr 11) by which valine replaces glutamic acid at 6th position leading to formation of Hb S [4]. HbS under low oxygen tension and dehydration has tendency to polymerize into long rigid fibers which distort the shape of RBC leading to sickle shape RBC, which are fragile and are lysed in extravascular and intravascular systems [1]. Also they obstruct the small capillaries causing microvascular occlusion. This disease is usually diagnosed in childhood, delayed detection in adulthood is seen in regions lacking screening leading to severe complications like pulmonary hypertension, cerebrovascular accidents haemolytic crisis due combination of sickling, endothelial injury, hypercoagulable state, Vaso-occlusion, thromboembolism. This report highlights this unusual presentation of homozygous SCD in a young adult who developed acute thromboembolic events affecting the lungs, heart, brain within course of single admission.

Case presentation

A 31-yr old female, Komal More, presented with generalized pain in all her limbs since 1 week insidious in onset gradually progressive associated with headache and fatigue since 15 days. No power loss, seizure, syncope, vomiting or signs of increase ICP. On presentation (day1) her vitals were stable. Laboratory findings showed haemoglobin 8.3 g/dl, mean corpuscular volume (MCV) 68fl, white blood cell count 17810/microL, platelet count 375000/microL, total bilirubin 2.6 mg/dl (direct 1.1, indirect 1.5), SGOT 38IU/L, SGPT 34IU/L, creatinine 1.2 mg/dl. Thyroid function tests revealed TSH 16.24 microIU/mL, with normal T3/T4, suggestive of sick euthyroid syndrome. Peripheral smear showed anisopoikilocystosis with microcytosis.

Suspecting sickle cell anaemia, a sickling test was done which turned out to be positive. Haemoglobin electrophoresis showed a peak in HbS window and elevated HbF, consistent with homozygous sickle cell disease. She was then treated with hydroxyurea, analgesics, hydration, oxygen therapy and was improving well for next 2 days.

On day 4 she developed acute onset breathlessness with persistent SpO2~60%, even non-invasive ventilation failed to improve oxygenation, she was then intubated providing invasive intubation. In an attempt to find the cause of respiratory distress 2D Echocardiography was done which revealed moderate pulmonary hypertension, severe tricuspid regurgitation, right atrial and right ventricular dysfunction and mild pericardial effusion. Post-intubation her hypoxemia decreased as revealed by ABG.

On the same day, (day 4) labs showed haemoglobin 4.8g/dl, WBC 40,210/microL, platelets 50,000/microL, MCV 64fl, total bilirubin 4.1 mg/dl(direct 0.9, indirect 3.2), SGOT 98IU/L, SGPT74 IU/L, D-dimer 3880ng/ml. Bone marrow aspiration was done to rule out bone marrow dysfunction which revealed hypercellular marrow with erythroid hyperplasia. Also, iron studies did not reveal iron deficiency. Also, no hepatomegaly or splenomegaly or haematuria. Accordingly, blood transfusions and antibiotic treatment was given.

By the evening of day 5, the patient regained consciousness, was extubated and maintained stable oxygenation.

On day 6 haemoglobin improved to 9.8g/dl, WBC 33,850/microL, platelets 68,000/microL, total bilirubin 2.8mg/dl(direct 1.0, indirect 1.8), SGOT 78IU/L, SGPT62IU/L.

On day 7, she developed multiple seizures progressing to status epilepticus for which CSF study was done, which was normal also MRI Brain was done which showed acute infarct in right splenium of corpus callosum in pericallosal branch of the posterior cerebral artery, suggestive of embolic stroke. With intensive supportive care, appropriate management with anticoagulant and antiplatelets, the patient was stabilized over the period of next 6 days and discharged on 14th day with clinically and vitally improved state.

Discussion

SCD complications arise due to multiple factors including haemolytic crisis, abnormal red cell structure, microvascular obstruction and thromboembolic episodes. Pulmonary hypertension being one of the serious cardiopulmonary complications, is thought to be because of repeated microvascular obstruction, nitric oxide depletion, and progressive vascular remodelling. The presence of severe tricuspid regurgitation and right sided failure in our patient suggest chronic pulmonary vascular disease likely precipitated by acute Vaso-occlusive crisis.

Cerebral infarction is another complication of SCD, with ischemic stroke affecting both children and adults [2]. In our case, the acute non haemorrhagic infarct in the splenium of the corpus callosum supplied by posterior cerebral artery points towards an embolic event, possibly due to patients hypercoagulable [6] and severe endothelial injury. Elevated D- dimer levels supports the active thrombus formation during hospitalization.

Bone marrow examination showed erythroid hyperplasia indication well-functioning marrow with ongoing compensatory haematopoiesis rather than marrow suppression, which then helped guide transfusion strategy. The absence of hepatosplenomegaly may reflect functional asplenia, a common long-term outcome in homozygous SCD.

This case highlights the clinical importance of considering SCD in adults presenting with unexplained fatigue, pain and anaemia. Early detection, monitoring the possible neurological and cardiovascular complications and timely institution of disease modifying therapy such as hydroxyurea [7] can be life saving and significantly improve prognosis [3].

Conclusion

Undiagnosed homozygous SCD presenting in adulthood can be life threatening due to its multisystem thromboembolic complications. A high index of suspicion and multidisciplinary approach involving prompt diagnosis, supportive therapy and targeted interventions proves to be life saving with good prognosis in such patients

Abbrevation

SCD	Sickle cell disease
b/l	bilateral
MRI	Magnetic resonance imaging
RBC	Red blood cell
SGOT	Serum glutamic oxaloacetic transaminase
SGPT	Serum glutamic pyruvic transaminase
CSF	Cerebro spinal fluid

Statements and Declarations

Conflicts of interest

The authors declare that they do not have conflict of interest.

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