

CASE STUDY

AN UNUSUAL CASE OF SICKLE CELL DISEASE

S Rahar¹, S Marwah², S Singh¹

¹Senior Resident, Department of Pathology, PGIMER & Dr. RML Hospital, New Delhi

²Professor, Department of Pathology, PGIMER & Dr. RML Hospital, New Delhi

Received: 12 August, 2019/ Accepted: 20 August, 2019

ABSTRACT: Sickle cell disease is one of the common hemoglobinopathies in the world affecting many organs including spleen. Splenomegaly in adult patients with sickle cell disease is very uncommon, however, in this case patient presented first time in adulthood with acute abdomen which is unusual. This case is presented to emphasize that, although rare, splenomegaly can persist in adults with sickle cell disease and can be associated with severe and even life-threatening splenic sequestration

KEYWORDS: Sickle cell disease, Haemoglobinopathies, Splenomegaly, Splenic sequestration

INTRODUCTION:

Sickle cell disease is one of the common hemoglobinopathies in the world¹.

It can affect many organs, however, it commonly affects spleen. It is commonly enlarged during the first decade of life but then undergoes progressive atrophy leading to autosplenectomy. The clinical manifestations of SCD arise from the tendency of sickle haemoglobin to polymerize at reduced oxygen tensions and deform red cells into the characteristic rigid sickle cell shape.

CASE REPORT:

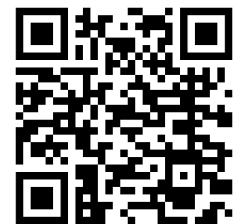
A 19 years old unmarried female with no significant illness since her childhood was admitted for pain abdomen since 5 days in left upper quadrant. Pain was acute in onset, severe, non radiating, no aggravating or relieving factors present. Patient also had fever since 5 days which was intermittent, high grade. It was not associated with chills and rigor. She had a history of easy fatigability for past one month. No history of bleeding/rash/burning micturition was present. On examination, icterus was present.

Corresponding Author:

Dr. Sadhna Marwah,

Professor,

Haematology, Department of Pathology, PGIMER & Dr. RML Hospital, New Delhi, PIN-110001



Per abdomen examination revealed tenderness in left hypochondrium, spleen was palpable 3 cm below left costal margin. Other systemic examination was normal.

Complete blood count investigation revealed hemoglobin-4.2g/dl, total leukocyte count-24000/ μ l, differential leukocyte count- P58%L20%E3%Myelocyte10%, Metamyelocyte7%, platelets-1.5lac/ μ l, PCV- 11.5%, MCV-75.3fl, MCH-28.2pg, MCHC-37.4%.

Peripheral smear examination showed RBCs-moderate anisopoikilocytosis with presence of microcytic hypochromic cells, sickle cells, target cells, spherocytes, tear drop cells, nRBCs- 52/100 WBC. WBCs showed neutrophilic leukocytosis with marked shift to left. Platelets were adequate on smear. (Fig 1,2) An impression of leukoerythroblastic picture with presence of sickle cells was given.

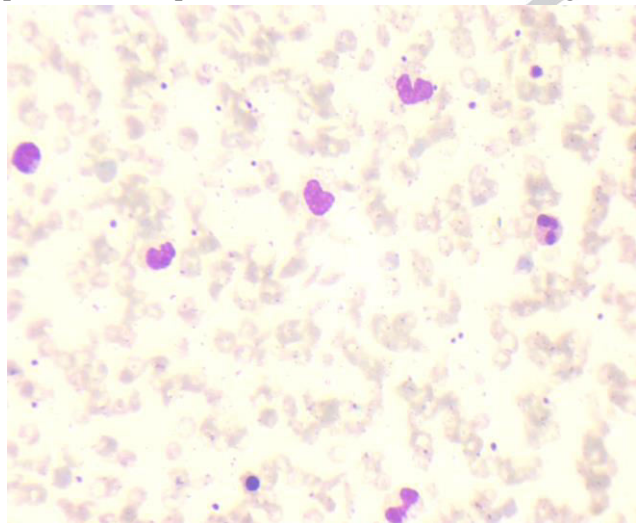


Fig 1. (100X, Giemsa)- Peripheral smear showing shift to left with presence of myelocyte, metamyelocyte and nRBC along with sickle cells.

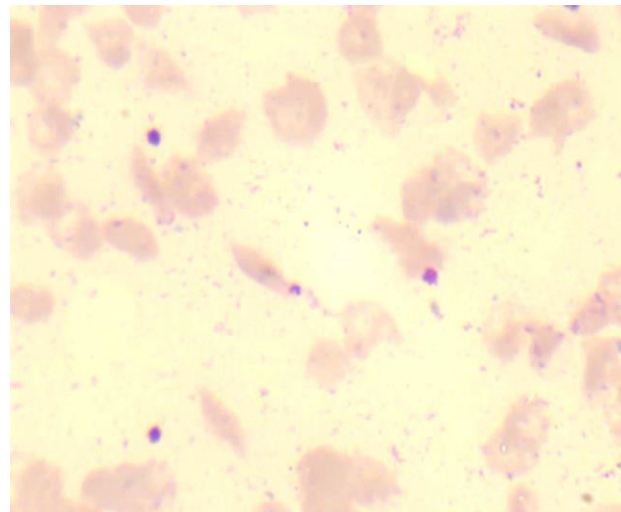


Fig 2. (400X, Giemsa)-Peripheral smear shows many sickle cells.

Liver function and kidney function test was deranged with laboratory values of bilirubin (Total/Direct/Indirect) -2.7/1.1/1.6mg/dl, SGOT-403U/L, SGPT-133U/L, ALP-62 U/L, urea-55mg/dl, Creatinine-1.1mg/dl, uric acid-6.6 mg/dl. Cardiac profile showed CPK-3375U/L, CK-MB-39U/L, LDH-155760 U/L. Ultrasound abdomen was done and showed hepatomegaly with normal echotexture, splenomegaly with altered echotexture, irregular hypoechoic area noted near the splenic hilum (Infarct). High performance liquid chromatography was done (Table 1)

Table 1. High performance liquid chromatography (HPLC) of patients.

	PATIENT	NORMAL
HbF	22.70%	<1.50%
Hb A	3.40 %	83.24-90.79%
HbA2	2.50 %	1.50-3.50%
Hb S	71.30%	0

Bone marrow aspiration was done which showed erythroid hyperplasia with M:E ratio of 1:2.5 with mixed erythroid reaction i.e. normoblastic and megaloblastic. Myeloid series cells were seen in all

stages of maturation. Megakaryocytes were adequate. Finally, sickling test was performed which came out to be positive.

Patient was given exchange transfusions, was started on hydroxyurea and supportive treatment. She showed slight improvement but after few days developed severe chest pain and bone pain. Later she expired due to renal failure, sequestration crisis and acute chest syndrome.

DISCUSSION:

The incidence of splenomegaly in sickle cell anemia appears to be around 10% after 10 years of age. Persistent splenomegaly in older children is rare and is frequently associated with hypersplenism². Sometimes sickle cell disease can present with lesser severity in comparison to the classic sickle cell disease. Multiple factors responsible have been evaluated to explain this progression like occurrence of Asian haplotype of bS globin gene, high levels of fetal hemoglobin (HbF), and frequent α -thalassemia³.

In a study, Sickle cell anemia (SCA) patients with persistence of splenomegaly were categorized into

- a) SCA with high fetal hemoglobin group when HbF levels were >20%,
- b) patients with sickle cell beta-thalassemia (S-bthal) identified by using criteria that combined microcytosis, hemoglobin A2 (HbA2) > 3.5%, presence of sickle cell hemoglobin (HbS) with the absence of hemoglobin A1 (HbA) and
- c) patients with associated Alpha-thalassemia, patients were evaluated for it by using the criteria combining microcytosis and HbA2 < 3.5% .

The HbF has inhibitory effect upon the process of RBC sickling by interfering with the formation of HbS polymers; it has been further observed that patients having HbF levels >20% are more likely to have milder sickling manifestations in sickle cell disease.

Higher HbF levels may have some contributory role in the late persistence of splenomegaly, probably by an ameliorating influence upon the sickling process. Other responsible factors postulated for the mild nature of sickle cell disease comprise the Asian haplotype of bS globin gene and the frequent co-occurrence of α -thalassemia. α -thalassemia co-occurrence in the patients can be evaluated by correlating hematological indices with Hb electrophoresis and HbA2 levels; the definitive diagnosis of associated α -thalassemia requires either globin-chain analysis or DNA studies. A large spleen predisposes the patient to the risks of sequestration crises, hypersplenism, infarction, and abscess formation.

Splenic enlargement does not imply its normal function, and the enlarged spleen may act only as a reservoir for blood with markedly deranged reticuloendothelial system (RES) function. Functional hyposplenism in the sickle cell disease can affect the patients over a wider age range and is frequently associated with late persistence of splenomegaly. Late, persistent splenomegaly has also been reported in the 5% of Jamaican (similar to North America), 19% of Greek, and 69% of Indian sickle cell disease patient populations. In the former two population groups, the predominant bS globin gene haplotype is of Benin type while Indian sickle cell disease has Asian haplotype of the bS globin gene. Sickle cell disease presents most commonly with anaemia. Although splenic sequestration crises are rare but can present as acute abdomen in sickle cell disease. The patient with sickle cell anaemia /sickle cell b⁰ thalassemia has more episode of pain and higher degree of anaemia than patients with haemoglobin SC disease/sickle b+ thalassemia. Acute pain in patients with sickle cell disease is caused by ischemic tissue injury resulting from the occlusion of microvascular beds by sickled erythrocytes during an acute crisis. Chronic pain occurs because of the destruction of bones, joints and visceral organs as a result of recurrent crises. An acute abdominal pain crisis can resemble an intra-abdominal process such

as cholecystitis / appendicitis / peritonitis/perforation/pancreatitis. Investigations like abdominal ultrasound scans and radiographs could be found useful to rule out other causes of acute abdomen.

CONCLUSION:

This patient presented first time in adulthood with acute abdomen which is unusual. Splenomegaly in adult patients with sickle cell disease is also uncommon and splenic sequestration crises are rare. This case is presented to emphasize that, although rare, splenomegaly can persist in adults with sickle cell disease and can be associated with severe and even life-threatening splenic sequestration.

REFERENCES:

1. Wintrobe clinical haematology. 13th ed;2013: 686-702.
2. Sommer A, Kontras S et al. Splenomegaly with Hypersplenism in Sickle Cell Anemia Treated by Radiation—Case Report ;Paediatrics.1971;48(3)
3. Chopra R, Rahman A et al. Fibrocongestive Splenomegaly in Sickle Cell Disease: A Distinct Clinicopathological Entity in the Eastern Province of Saudi Arabia. Am j Haemat.2005;180-185
4. Moll S, Orringer EP et al. Case report: splenomegaly and splenic sequestration in an adult with sickle cell anemia. Am J Med Sci. 1996 Dec;312(6):299-302.

Cite of article: Rahar S, Marwah S, Singh S. An unusual case of sickle cell disease. Int. J. Med. Lab. Res. 2019, 4(2):41-44

CONFLICT OF INTEREST: Authors declared no conflict of interest

SOURCE OF FINANCIAL SUPPORT: Nil

International Journal of Medical Laboratory Research (IJMLR) - Open Access Policy

Authors/Contributors are responsible for originality of contents, true references, and ethical issues.

IJMLR publishes all articles under Creative Commons Attribution- Non-Commercial 4.0 International License (CC BY-NC).

<https://creativecommons.org/licenses/by-nc/4.0/legalcode>