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Persistent splenomegaly in an adult female with homozygous sickle cell anemia

SOMPAL SINGH, DEEPAK KUMAR SINGH, RUCHIKA GUPTA, SONU NIGAM, & TEJINDER SINGH

Department of Pathology, Maulana Azad Medical College, New Delhi, India

Abstract

Sickle cell anemia (SCA) is associated with repeated episodes of erythrostasis in the spleen, which lead to thrombosis and infarction of the spleen resulting in "autosplenectomy" which is usually complete by 8 years of age.

We present a case of a 22-year-old female who presented with complaints of fever, bone pain and joint swelling. On examination she had pallor, icterus and moderate splenomegaly. Her hemoglobin was 7.5 g/dl. Peripheral smear showed many sickled red cells. Slide test for sickling was positive with 2% sodium metabisulphite. Hemoglobin electrophoresis revealed a single band in the hemoglobin S, D, and G region. No band was seen in the HbA & HbA_2 region. HbF level was 0%. USG showed an enlarged spleen with few defined hypoechoeic lesion.

We present this case because of rarity of association of homozygous SCA with splenomegaly in this age group, the confusion that echogenic lesions in spleen can create and to emphasize the risk of sequestration crises, which remains in such cases.

Keywords: *Sickle cell anemia, splenomegaly, hypoechoic, autosplenectomy*

Introduction

Sickle cell anemia (SCA) is an autosomal recessive disorder with production of abnormal "sticky" hemoglobin, which binds with other hemoglobin molecules, causing red cell deformity and irreversible sickling of red cells [1,2]. Resultant erythrostasis is responsible for ischemia and infarction throughout the body, including autosplenectomy. Because of autosplenectomy, the spleen is no longer palpable after 8 years of age in most of the patients [2].

We present a rare case of homozygous SCA with persistent splenomegaly in an adult patient with focal hypoechoeic lesions in the spleen. The significance of splenomegaly and focal hypoechoeic splenic lesions are discussed.

Case report

A 22-year-old female patient, resident of Bihar, presented with complaints of episodic joint swelling and bone pain for the past 10 years and fever with jaundice for the past one month. There was a history of an episode of jaundice one year previously. Family history revealed that her brother had died of similar complaints at 10 years of age. On physical examination she was thin built with mild pallor and icterus. Abdominal examination revealed non-tender splenomegaly, 5 cm below left costal margin. Ultrasound examination showed splenomegaly with diffusely altered echo texture and few well-defined hypoechoeic lesions. The liver was normal in size and echo texture. A single gallbladder calculus, 4 mm in size was also observed.

Her hemoglobin was 7.5 g/dl, a total leukocyte count of 9000/mm^3, with P_{56}, L_{50}, M_2, E_3. Platelet count was 1.5 lakhs/mm^3. MCV was 89 fl with MCHC of 33.8 g/dl. The peripheral blood film showed moderate anisopokilocytosis, with normocytic normochromic RBC along with many polychromasia, some macrocytes, sickled red cells and target cells (Figure 1). The reticulocyte count was 11%. The slide

Correspondence: T. Sing, Director-Professor & Head, Department of Pathology, Maulana Azad Medical College, New Delhi 110002, India. E-mail: sompal151074@yahoo.com

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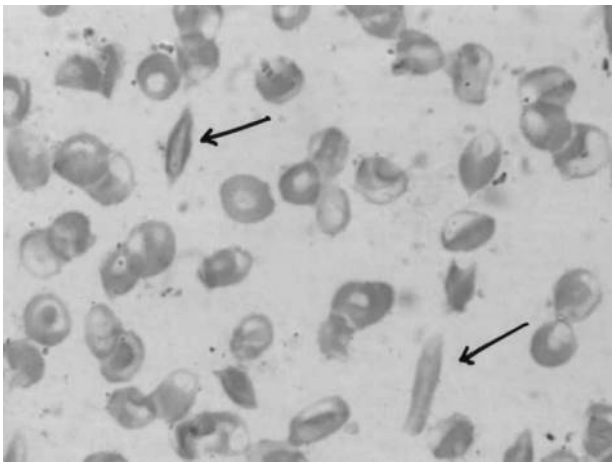

Figure 1. Peripheral smear showing normocytic normochromic RBCs along with irreversibly sickled RBCs (long arrows), Giemsa, 400x.

test for sickling with 2% sodium metabisuphite was positive. Starch gel hemoglobin electrophoresis at pH 8.6 showed single band in a hemoglobin S, D, and G region with no band in the HbA and HbA_2 region (Figure 2). HbF was 0% and HbA_2 was 2.0%. Laboratory investigations revealed total serum bilirubin of 5.4 g/dl (direct 4.2 g/dl), total serum proteins — 6.2g m/dl, serum albumin — 4.7 g/dl and serum globulin — 1.5 g/dl (A:G ratio–3.3:1). Her brother had not being worked up before his death, and hence his diagnosis was not known.

Since double heterozygotes for HbSD do not show sickling on peripheral smear and HbS/β thalassemia show $HbA_2 > 3.5\%$ [2], the final diagnosis of homozygous SCA (HbSS) with persistence of splenomegaly was made. The patient refused further treatment and went back to her native village.

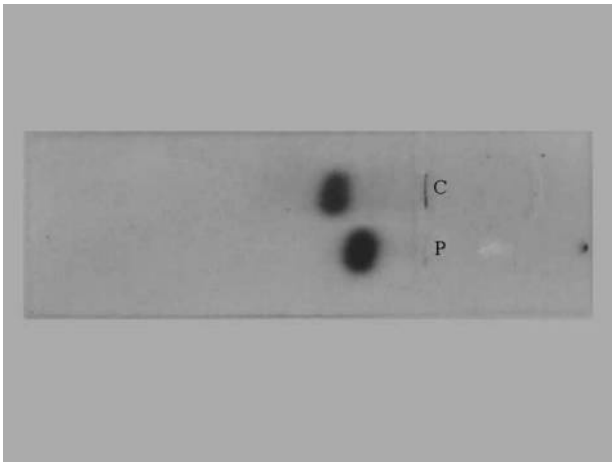

Figure 2. Hemoglobin electrophoresis demonstrating a slow-moving band in the region of HbS and the absence of band in region of HbA and HbA_2 (C and P denote point of application of control and patient, respectively), starch gel electrophoresis, pH 8.6.

Discussion

Homozygous SCA usually presents with signs and symptoms of pallor, fever, and abdominal and joint pains within the first year of life [3]. First presentation in adult age group is rare [4]. Our case also presented with complaints of joint swelling and bone pains for past 10 years.

Abnormal red cell rheology leads to erythrostasis in SCA, because of which there is ischemia and infarction throughout body, involving spleen, bone marrow, bone, lungs, etc. [1]. Process of autosplenectomy due to repeated episodes of infarction starts at 2 years of age and is complete by the age of 8 years. Although splenomegaly is known in children, spleen is not palpable in adults [2,4]. However, our patient had palpable spleen 5 cm below costal margin, which is rare at the age of 22 years. Recent study suggests that the persistence of spleen may be due to chronic malaria or improved clinical care [5]. This does not explain persistent splenomegaly in our case as our patient did not show evidence of chronic malarial infection and was not under any treatment.

Causes of persistent splenomegaly in sickle cell disease include HbSD, HbSβ thalassemia and splenic sequestration crisis. The former two were excluded in our case due to the positive sickling, $HbA_2 < 3.5\%$ and 0% HbF [2]. Splenic sequestration crisis is defined by a decrease in steady state hemoglobin concentration of at least 2 g/dl, evidence of marrow erythropoiesis and an acutely enlarged spleen [4]. Sequestration crises usually present in infants and young children whose spleens are chronically enlarged before autoinfarction and fibrosis have occurred leading to autosplenectomy. It is characterized by sudden occurrence of massive splenomegaly at the expense of blood volume. Circulatory collapse and even death can occur within few hours [6]. High index of suspicion is needed as it may present with sudden drop of hemoglobin even without palpable spleen [7]. Although persistence of spleen is rare in adult cases of sickle cell disease, chances of sequestration crises in such cases do remain a possibility.

Walker et al. reported the presence of focal hypoechoic lesion in spleen of SCA in children [8]. Our patient also showed similar lesions in spleen. The differential diagnoses of focal hypoechoic splenic lesions comprise of splenic abscess, lymphoma [8,9] and metastasis [8,10]. Our patient did not have any features of these conditions. Walker et al. emphasized the benign course of such lesions with no need for further investigation for malignant disease [8]. These hypoechoic lesions on ultrasonography have imaging characteristics of normal spleen on MRI, thus allowing for a correct diagnosis. They are probably "preserved functioning splenic tissue" in an otherwise fibrosed and calcified spleen [11].

To conclude, homozygous SCA is a rare, though possible diagnosis in an adult with anemia and sickled red cells on smear, and such adult patients who present with splenomegaly need regular follow-up for early diagnosis and treatment of any complication or crisis.

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