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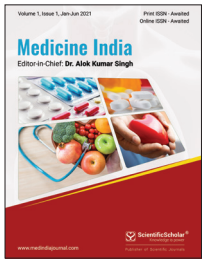

Case Report

A rare presentation of sickle cell disease diagnosed for the first time in a 60-year-old female: A misdiagnosis or missed diagnosis

Darshankumar Manubhai Raval[1], Vaishnavi Mahendrasinh Rathod[2]

[1]Department of Infectious Diseases, Mayo Clinic, Jacksonville, Florida, United States, [2]Department of General Medicine, Baroda Medical College, Vadodara, Gujarat, India.

***Corresponding author:**
Vaishnavi Mahendrasinh Rathod,
Department of General Medicine, Baroda Medical College, Vadodara, Gujarat, India.

vaishnavirathod40@gmail.com

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ABSTRACT

Sickle cell disease (SCD) mainly manifests in early childhood. During the first 6 months of life, infants are largely protected by high levels of hemoglobin F; soon thereafter, the disease becomes evident in form of various crises such as vaso-occlusive crisis, hemolytic crisis, and acute chest syndrome, and in rare instances aplastic crisis. Therefore, the majority of patients with SCD are diagnosed in childhood or adulthood, whereas those with sickle cell trait usually remain asymptomatic. However, here, we are presenting a rare case in which a patient has been diagnosed to have SCD for the 1st time at the age of 60 years while being treated for dengue and severe anemia. Our case is one of the rare presentations of SCD, as she never required any blood transfusions even during childbirth and without any sickle crises; therefore, she might not be diagnosed with SCD till her presentation to our hospital. This indicates that advanced age should not be considered as an exclusion criterion for the suspecting and diagnosis of SCD. This case outlines the need for awareness and incorporation of screening for sickle cell anemia in childhood or adulthood as same thalassemia screening programs. The early diagnosis by screening programs will ensure appropriate treatment and therefore improve the quality of life in such patients.

Keywords: Sickle cell disease in elderly, Splenomegaly in elderly, Dengue fever, Severe anemia, Sickle cell crisis, Elderly population

INTRODUCTION

Sickle cell disease (SCD) is a type of hemoglobinopathies, characterized by atypical hemoglobin molecules called hemoglobin S (HbS), distorting the shape of red blood cells into a sickle, or crescent shape.[1,2]

Being an autosomal recessive disorder, both copies of the gene in each cell have to be mutated to manifest the disease clinically. Each carrier parent of an individual with a disease carries one copy of the mutated gene, but they usually do not develop symptoms and signs of the condition, hence called sickle cell trait.[2]

Although SCD is present since birth, most infants do not develop any problems from the condition until 5 or 6 months of age.[3] Keeping this perspective in mind, we report a patient who was diagnosed for the 1st time to have SCD at the unusual age of 60 years.

CASE REPORT

A 60-year-old female without any major medical illness, presented to our hospital, from a tribal area of Gujarat, with complaints of fever, dizziness, breathlessness, fatigue for 5 days, and vomiting for a day. There were no complaints of bleeding from any site, cough, cold, chest pain, joint pain, loss of consciousness, or palpitation. She had a history of jaundice 5–6 times without any significant family history of any disease. The patient had four normal vaginal deliveries without any complications and was in a menopausal state for the past 10 years.

On general examination, she was vitally stable except for the presence of pallor and icterus. The systemic examination revealed a loud second heart sound without any murmur, reduced air entry with diffuse crepitation in bilateral lung fields, and non-tender splenomegaly (Hackett's Grade 2) without hepatomegaly.

The routine investigations were suggestive of severe anemia and thrombocytopenia with sepsis, indirect hyperbilirubinemia, hyperkalemia, raised transaminases, and negative for malarial parasite with normal urine and coagulation profile [Table 1]. On serological investigations, dengue NS1 antigen was found to be positive, with other viral markers (Hepatitis B, hepatitis C, hepatitis A, hepatitis E, and human immunodeficiency virus) and widal which were negative. The thyroid function test, stool investigations, blood and urine cultures, and direct and indirect Coomb's test did not show any abnormality.

A working diagnosis of dengue fever with hepatitis was made, and the patient was treated as per standard guidelines for dengue fever along with antibiotics for sepsis, and prophylactic nasal oxygen for severe anemia.

However, history, symptoms, or signs were not suggestive of hemorrhage, so we did not consider dengue fever as a cause of severe anemia. To find out the cause of severe anemia, further investigations were done, which were suggestive of positive sickle solubility test, SCD on high-performance liquid chromatography (HPLC) test (Hemoglobin F - 25%, hemoglobin subunit alpha 2-3.4%), hemolysis (raised lactate dehydrogenase hydrogenase), and macrocytic hypochromic red blood cells with sickle cells on peripheral smear. Radiological investigations, such as chest X-ray, showed bilateral mild pleural effusion, whereas ultrasonography of the abdomen revealed moderate splenomegaly and mild hepatomegaly with normal liver texture. This is also an unusual finding as in most patients of SCD; the spleen is usually atrophied by adolescence due to multiple episodes of splenic infarcts.

The patient continued to be treated for both dengue fever with hepatitis and SCD, with fluid therapy, analgesics, antibiotics, and transfusion of packed cell volume. Close monitoring was done to look for signs of shock, hemolysis, hemorrhage, as well as fluid overload. The patient improved and became asymptomatic after a week of admission with returning of blood parameters toward the normal range. After 10 days of hospitalization, the patient was discharged with advice to take plenty of oral fluid and to continue medications for SCD to prevent a further sickle crisis.

Table 1: Serial routine blood investigations.

Investigations	On admission	Day 3	Day 7	Day 10
Hemoglobin (mg/dL)	3.90	11.20	11.40	11.30
White blood cell (per cumm)	36300	22400	14000	10500
Neutrophil (%)	62	56	66	70
Lymphocyte (%)	36	42	32	28
Monocyte (%)	01	01	01	01
Platelet count (per cumm)	59000	65000	78000	123000
Reticulocyte count (%)	1	5	–	6
Malarial parasite	Negative	–	–	–
Bilirubin–total, direct, indirect (mg/dL)	4.8, 1.5, 3.3	9.3, 2.5, 6.8	5.6, 2.2, 3.4	3.6, 1.4, 2.2
Urea (mg/dL)	71	74	48	36
Creatinine (mg/dL)	1.28	0.84	0.70	0.68
S. Protein (g/dL)	6.5	6.0	6.4	6.7
S. Albumin (g/dL)	3.5	3.0	3.6	3.8
Alanine transaminase (IU/L)	1467	2192	1120	560
Aspartate transaminase (IU/L)	2754	4860	2167	643
S. Potassium (mEq/L)	5.8	4.9	4.4	4.1
S. LDH (U/L)	1959	3848	1580	597
S. TSH (U/L)		3.1		
S. Iron Study		S. Iron–40 mg/dL S. Ferritin–116 mg/dL TIBC–360.g/dL Transferrin Saturation–32% s/o Mild Iron Deficiency Anemia		
S. Vitamin B-12 (pg/mL)		110		

S. Protein: Serum protein, S. Albumin: Serum Albumin, S. Potassium: Serum potassium, S. TSH: Serum thyroid stimulating hormone, S. LDH: Serum lactate dehydrogenase hydrogenase, S. Iron: Serum iron, S. Vitamin: Serum vitamin, S. Ferritin: Serum ferritin, TIBC: Total iron-binding capacity

DISCUSSION

SCD is one of the most common monogenic disorders globally with an autosomal recessive inheritance.[4] The history and physical examination of sickle cell patients range from being asymptomatic to a broad range of presentations, depending on the type of complication and the body system affected such as vaso-occlusive crisis, acute chest syndrome, infections, pulmonary hypertension, cerebrovascular

accidents/stroke, pulmonary embolism, eye complication, renal complication, splenic sequestration, priapism, cholelithiasis, osteonecrosis, and aplastic crisis. Patients are completely asymptomatic during the first 6 months of life due to the presence of fetal hemoglobin which gradually decreases and HbS begins to predominate.[5] Most cases of SCD are diagnosed in childhood and adulthood.

The case reports by Yaranal *et al*.,[5] and Sood *et al*.,[6] presented a 55-year-old patient with bilateral lower limb paraplegia, and a 46-year-old patient with fat embolism syndrome, respectively, in previously undiagnosed SCD. Similarly, our patient was also a 60-year-old female who had no history of crisis related to SCD, whereas sickle crisis is a usual presenting complaint of patients in early childhood and adolescence according to the literature.[7] For those without significant symptoms or with mild symptoms such as intermittent pain, these syndromes are often missed by providers due to the mild anemia, lack of robust hemolysis, and non-specific smear. Microcytosis in the presence of normal iron stores is frequently the only abnormality suggestive of hemoglobinopathy in these instances.[6]

Our patient was also positive for dengue NS1 antigen; however, the presence of severe anemia was not explained by dengue fever in absence of symptoms and signs of hemorrhage. Considering the high prevalence of SCD in a tribal area of Gujarat state,[8] and with a history of multiple episodes of jaundice in the past, we considered for sickling solubility test, which turned out to be positive, and further, investigation by HPLC showed SCD hemoglobinopathy. The patient had four normal vaginal deliveries, for which she never required a blood transfusion, to add to this, she never had any major sickle cell crisis episodes, which explains the patient's undiagnosed state of SCD. Although the usual presentation of SCD is in childhood or early adulthood, our suspicion and correlation from the residence, history, examination, and initial routine investigations have led us to the diagnosis of the patient, which could have been missed or misdiagnosed otherwise.

CONCLUSION

We have reported an unusual case of SCD, the 1st time diagnosed at the age of 60 years in a female coming from a tribal area of Gujarat State. After correlating the data from the residence, history of multiple episodes of jaundice, examination, and routine investigations, we reached the diagnosis of SCD, which was previously undiagnosed. The diagnosis of SCD guided us in the correct management of the patient's condition. Therefore, through this case, we want to bring forward the possibility of undiagnosed state SCD in the elderly population. The advanced age should not be used as an exclusion criterion for the diagnosis of SCD, which could lead to misdiagnosis or a missed diagnosis of the condition. Moreover, we also want to emphasize the need for screening programs for sickle cell anemia in the childhood and adolescent population, especially in tribal areas of India. By creating awareness in the tribal population, we can diagnose the condition earlier and treat the condition appropriately, therefore improving the quality of life of patients with SCD.

Author contributions

Darshankumar Manubhai Raval: (1) Final approval of the version to be published. (2) Aptitude to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.
Vaishnavi Mahendrasinh Rathod: (1) Concept and design of the study, acquisition of data, or analysis and interpretation of data. (2) Drafting the article or revising it critically for important intellectual content.

Declaration of patient consent

Patient's consent not required as patients identity is not disclosed or compromised.

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Conflicts of interest

There are no conflicts of interest.

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