

Sickle Cell Disease With Ulcerative Colitis in An Ethiopian Child

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Abstract: Sickle cell disease (SCD) is a hereditary blood disorder characterized by abnormal hemoglobin, resulting in various clinical manifestations. Recognizing the complex clinical picture of SCD is crucial for physicians to effectively diagnose and manage the disease. While typical presentations may be absent, it is important to consider the possibility of SCD in patients presenting with ulcerative colitis (UC). The concurrent occurrence of UC and SCD is extremely rare. Therefore, a comprehensive investigation is warranted for individuals at risk of developing SCD with UC. In this report, we presented the first documented case of a child in Ethiopia with both SCD and UC.

Keywords: sickle cell disease, ulcerative colitis, children, Ethiopia

Introduction

Sickle cell disease (SCD) is a genetic blood disorder characterized by the abnormal shape and rigidity of red blood cells (RBCs), leading to impaired blood flow and various clinical symptoms. The crescent-shaped RBCs can cause blockages in small blood vessels, resulting in pain, infections, ulcers, and swelling of the hands and feet.¹

SCD is caused by a mutation in the β -globin gene and is a life-threatening condition affecting multiple systems, including the gastrointestinal system.² Abdominal pain is a common manifestation of SCD, but the coexistence of inflammatory bowel disease (IBD) and hemoglobinopathy, such as sickle cell anemia, is rare. To date, only one concurrent case of sickle cell anemia and ulcerative colitis has been reported in sub-Saharan Africa.^{3,4}

The spleen is particularly vulnerable in sickle cell anemia, and evidence of hyposplenism, characterized by fibrosis and atrophy, can typically be observed within the first five years of life. In our patient, functional asplenia was present, leading to anatomical asplenia. Dysfunctional spleen function may also be seen in other conditions, such as ulcerative colitis, Crohn's disease, and celiac disease.^{5,6}

Case Report

We present the case of a 13-year-old female child who initially presented at the age of 3 years with complaints of easy fatigability, multiple joint pain, decreased appetite, and irritability lasting for one month. She was initially evaluated at a nearby health center and subsequently referred to Tikur Anbessa Specialized Hospital (TASH) for further investigation due to severe anemia of unknown etiology. The patient had received multiple transfusions of whole blood and packed red blood cells. She was then linked to the Haemato-oncology clinic but was lost to follow-up for approximately 10 years.

At the current age of 13 years, the patient returned to TASH with a complaint of easy fatigability, intermittent joint pain, decreased appetite, and lightheadedness persisting for the past 3 years. Her fatigue had progressively worsened, initially occurring with walking but later affecting even ordinary activities. Additionally, she reported a history of intermittent diarrhea and constipation, with periods of remission lasting for 2 years. The diarrhea episodes were described as yellowish to brownish in color and occasionally mixed with blood, occurring 3–4 times per day and alternating with episodes of constipation. The patient also experienced intermittent colicky abdominal pain, which worsened with changes

in bowel habits. She had sought medical attention at various healthcare facilities and received unspecified oral medications as well as multiple blood transfusions. Subsequently, she was referred to the pediatric gastrointestinal clinic at TASH for further evaluation.

Physical examination at TASH revealed a pulse rate of 100 beats per minute, respiratory rate of 22 breaths per minute, and axillary temperature of 36.6°C. Anthropometric measurements indicated a weight of 22 kg, height of 125 cm, and a body mass index for age between −2 and −3 standard deviations (SD) on the Z-score, indicating moderate acute malnutrition. The patient exhibited pale conjunctivae and palmar pallor. Based on the clinical presentation and findings, a diagnosis of moderate acute malnutrition and anemia associated with chronic diarrhea secondary to inflammatory bowel disease (IBD) was made.

This case highlights the challenges encountered in the management of a patient with coexisting sickle cell disease and ulcerative colitis. The prolonged period of loss to follow-up underscores the importance of consistent medical care and regular monitoring for individuals with complex medical conditions. Further investigations and targeted treatment plans will be implemented to address the patient's specific needs and improve her overall health and quality of life.

Following the investigations conducted, the case report provides the following findings and management:

Complete Blood Cell Count

The patient's white blood cell count was elevated at 14,500/mm³, with a differential count showing 36.5% neutrophils and 49.4% lymphocytes. The platelet count was 92,900/mm³, indicating thrombocytopenia. Hemoglobin level was 6.6 g/dL, hematocrit was 20.4%, and mean cell volume (MCV) was 91.5 femtoliters. Mean cell hemoglobin was 29.6%, indicating hypochromic red blood cells, and red cell distribution width was elevated at 28.4%.

Stool Examination and Serology

Stool examination did not reveal any significant abnormalities. Renal and liver functions were normal, and serology for hepatitis B surface antigen, hepatitis C virus, and HIV were negative.

Lactate Dehydrogenase and Peripheral Morphology

Lactate dehydrogenase (LDH) level was elevated at 273 mg/dL, indicating increased cell turnover. Peripheral morphology of red blood cells showed mild anisochromia (normochromic to hypochromic), mild anisopoikilocytosis with frequent polychromatophilic red blood cells, a few fragmented red blood cells ("boat-shaped"), and sickled red blood cells [Figure 1](#).

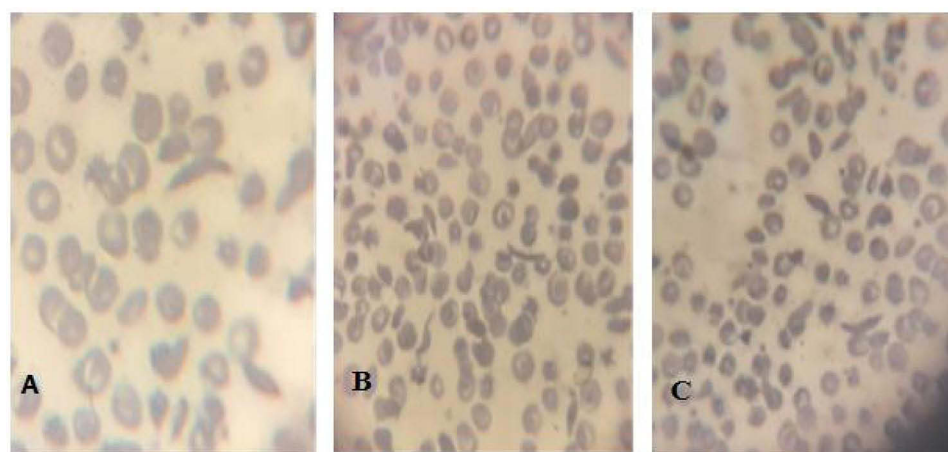


Figure 1 Peripheral smear from a patient with sickle cell anemia shows multiple sickle cells. (A) Sickle shaped and boat shaped RBCs. (B) Hypochromic and anisochromic RBCs. (C) Anisopoikilocytosis with polychromatophilic RBCs.

Abdominal Ultrasound

Abdominal ultrasound revealed diffused circumferential thickening of the cecum to sigmoid colon, indicating bowel wall inflammation. The spleen was not visible (absent), indicating functional asplenism.

Diagnosis

Based on the above findings, a diagnosis of moderate acute malnutrition, severe anemia, inflammatory bowel disease (ulcerative colitis), and asplenia secondary to sickle cell disease was made.

Treatment

The patient received packed red blood cell transfusion to address the severe anemia. Hydroxyurea, a medication that can help reduce sickling of red blood cells, was initiated. However, despite treatment, the patient continued to experience bloody diarrhea.

Colonoscopy with Biopsy

A colonoscopy was performed, revealing edematous and erythematous mucosae with continuous irregular rectal and sigmoid ulcers, as well as multiple erosions and ulcers throughout the colon. Biopsy of the colon showed expanded lamina propria with inflammatory infiltrates, basal plasmacytosis, lymphoid nodule formation, marked crypt dropout, minimal fibrosis, low-grade dysplasia areas, and polypoid changes. Separate ulcer surfaces and fibrinopurulent and granulation tissue fragments were also observed [Figure 2](#).

Diagnosis of Ulcerative Colitis

Based on the colonoscopy findings, the patient was diagnosed with ulcerative colitis. Treatment with prednisolone, a corticosteroid to reduce inflammation, and amoxicillin prophylaxis (likely to prevent infection) was initiated.

Clinical Improvement

On subsequent follow-up assessments, the patient showed noticeable clinical improvement, suggesting a positive response to treatment.

The case report underscores the complexity of managing a patient with multiple coexisting conditions, including sickle cell disease, ulcerative colitis, severe anemia, and moderate acute malnutrition. The treatment approach involved

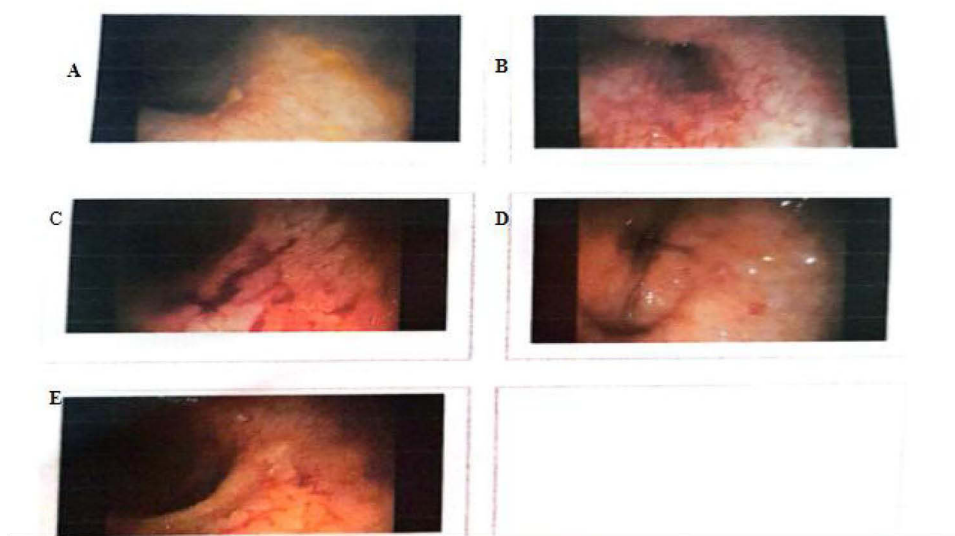


Figure 2 (A) Multiple rectal erythematous mucosae. (B) Multiple rectal edematous erosions. (C) Continuous irregular rectal sigmoid ulcers (D) Lymphoplasmacytic eosinophilic and minimal neutrophilic inflammatory infiltrations with basal plasmacytosis. (E) Low grade dysplasia areas of surface villi prosection and polypoid changes.

addressing the specific conditions with appropriate interventions, including transfusion, hydroxyurea, prednisolone, and amoxicillin prophylaxis. Regular follow-up and monitoring will be crucial to ensure continued improvement and long-term management of the patient's conditions.

Discussion

Indeed, the combination of a thorough medical history, physical examination, and diagnostic tests plays a crucial role in establishing accurate diagnoses. While hemoglobin electrophoresis and DNA testing are the gold standard for confirming sickle cell disease,⁷ the peripheral morphology findings in this case, along with the clinical and laboratory manifestations, strongly support the diagnosis. It's unfortunate that financial constraints limited the availability of comprehensive testing.

The management approach for sickle cell disease aims to prevent vaso-occlusive crises, manage anemia, and address associated complications. Hematopoietic stem cell transplantation is considered a curative option, but it may not be feasible or readily accessible for all patients due to various factors, including availability of suitable donors. In this case, the patient demonstrated clinical improvement with the prescribed management, which included packed red blood cell transfusion and the initiation of hydroxyurea. Hydroxyurea is commonly used in sickle cell disease to reduce the frequency and severity of vaso-occlusive crises.

Ulcerative colitis, on the other hand, is a chronic inflammatory bowel disease characterized by inflammation and ulceration of the colon and rectum. While colonoscopic biopsy is the definitive method for diagnosing ulcerative colitis, the clinical presentation, along with the colonoscopy findings of edematous and erythematous mucosae, ulcers, and erosions, strongly support the diagnosis in this case. The patient showed a positive response to oral prednisolone therapy, which is a common approach for inducing remission and controlling inflammation in ulcerative colitis. Maintenance treatment with 5-Aminosalicylates, which are anti-inflammatory agents, is often used to prevent relapse and reduce disease activity.

It's important to note that sickle cell disease is associated with increased susceptibility to various complications, such as infections, acute chest syndrome, stroke, and leg ulcers. The presence of sickle cell disease and ulcerative colitis in this patient adds complexity to the management, requiring a multidisciplinary approach involving hematologists, gastroenterologists, and other specialists to address the specific needs and potential complications associated with both conditions.

Regular monitoring, follow-up visits, and an individualized treatment plan will be essential in managing the patient's conditions, optimizing disease control, and promoting overall well-being.

Conclusion

Yes, based on the presented information, it appears to be a case of a 13-year-old female with sickle cell disease and ulcerative colitis. The peripheral morphology findings, combined with the CBC indices, support the diagnosis of sickle cell disease. The colonoscopy findings and biopsy results confirm the presence of ulcerative colitis. The additional finding of an absent spleen on abdominal ultrasound is consistent with the complications of sickle cell disease, specifically splenic infarction.

It's worth noting that the patient presented with abdominal pain, which was attributed to ulcerative colitis. This highlights the importance of considering and investigating additional causes of symptoms, even in individuals with known chronic conditions. The coexistence of sickle cell disease and ulcerative colitis can present unique challenges in terms of management and requires a multidisciplinary approach.

Overall, this case underscores the complexity of managing patients with multiple coexisting conditions and the importance of a comprehensive evaluation to provide appropriate treatment and care.

Consent

Informed consent has been duly obtained from the patient's legal guardian, and assent has been obtained from the patient, allowing for the publication of this case report and any accompanying images. Institutional consent or IRB (Institutional review board letter is not required for case report publication.)

Disclosure

The authors report no conflicts of interest in this work.

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