JK SCIENCE pISSN: 0972-1177

CASE REPORT

# Sickle-beta Thalassemia: A Rare Case Report in a Young Child

**Safia Rana, Neha Parashar, Sabina Khan**

## Abstract

**Introduction:** Sickle cell disease (SCD) results from a point mutation in the beta globin gene located on chromosome 11. The combination of inheriting a sickle cell allele from one parent and a beta thalassemia allele from the other parent leads to sickle cell-beta thalassemia, often exhibit a more severe type of SCD thus health issues. **Case:** A female child, aged 2 years and 11 months, came in with fever, weakness, and decreased appetite, history of frequent infections. The patient's hemoglobin level was 4.8 g/dL, mean corpuscular volume measured 65.7 fl. HPLC analysis revealed a peak in the S-window at 68.7%, along with an increase in Hb levels. **Conclusion:** Timely diagnosis and treatment of patients with with sickle-beta thalassemia can aid in minimizing complications.

## Key Words

Sickle Cell Disease, Sickle-beta Thalassemia, Hemolytic Anemia, Recurrent Infections

## Introduction

The inheritance of both the sickle cell mutation and the beta-thalassemia (-Thal) mutation results in a condition known as Hb S/β thalassemia, characterized by compound heterozygosity.[1] In India, the reported prevalence of sickle-beta thalassemia is 0.37%. Individuals with Hb S beta thalassemia may remain asymptomatic or experience occasional vaso-occlusive crises.[2] Screening and identifying sickle cell disease should occur within the initial months of life.[3] Sickle cell disease can result in both acute and chronic complications.[4] The current case illustrates the complications associated with sickle cell beta-thalassemia that can arise in a child at a young age.

## Case Report

A 2-year-11-month-old girl presented withfever, lethargy, and appetite lossand repeated infections. The patient's hemoglobin (Hb) was 4.8 g/dL, mean corpuscular volume (MCV) was 65.7 fl, mean corpuscular hemoglobin (MCH) was 21.3 pg, and haematocrit was 15.5%. Her total leucocyte count was 16,970/mm$^3$. Absolute counts of neutrophils were 12,422/mm$^3$, lymphocytes were 2,834/mm$^3$ and monocytes 1,527/mm$^3$.

The peripheral smear demonstrated a microcytic hypochromic pattern with the presence of sickle cells, target cells, and occasional teardrop cells. Fragmented erythrocytes and polychromatophils, nucleated red blood cells were noted at a frequency of 200 per 100 white blood cells. The corrected reticulocyte count was 5%. High-performance liquid chromatography (HPLC) was recommended, which revealed a peak in the S-window (68.7%), along with elevated levels of Hb F (13.1%) and Hb A2 (5.5%). Family screening and clinical correlation were recommended.

Her father was from Africa and lived there, but he had never been evaluated for a hemoglobinopathy. He had never received a blood transfusion before. Her mother was from Maharashtra. It was a recognized instance of

**Department of Pathology, Hamdard Institute of Medical Sciences and Research, Jamia Hamdard, New Delhi, India.**
**Correspondence to**: Dr. Sabina Khan, Professor & Head, Department of Pathology, Hamdard Institute of Medical Sciences and Research, Jamia Hamdard, New Delhi, India.
**Manuscript Received**: 3.01.2026; **Revision Accepted**: 7.04.2026;
**Published Online First**: 10th July, 2026.
**Open Access at**: https://journal.jkscience.org


**Cite this article as:** Rana S, Parashar N, Khan S. Sickle-beta Thalassemia: A Rare Case Report in a Young Child. JK Science 2026;28(3):192-5.

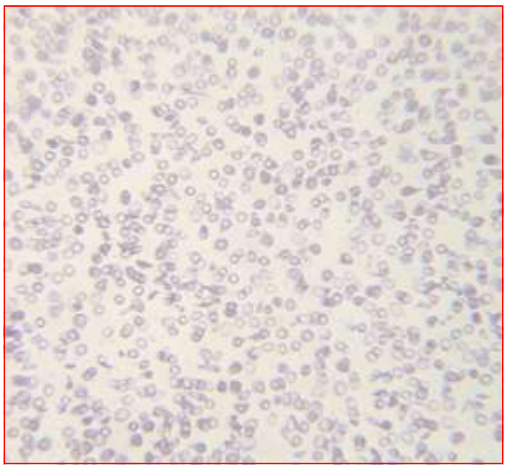

**Figure 1: Peripheral smear [Giemsa x 400]**

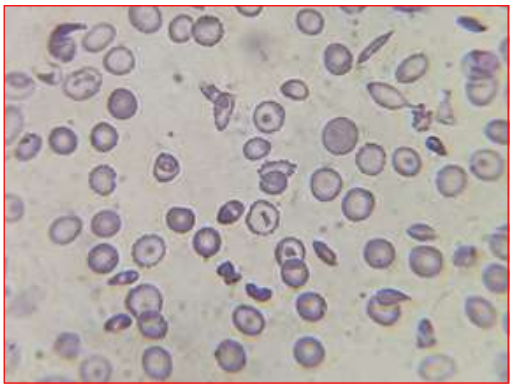

**Figure 2: Peripheral smear [Giemsa x 1000]**

the beta thalassemia trait. The younger sister was also given HPLC advice, which revealed Hb A2 of 5.0%, Hb F of 2.7%, and Hb A of 78.8%, all of which were indicative of a beta thalassemia trait. Therefore, a diagnosis of Compound Heterozygous sickle-beta thalassemia was proposed based on the patient's peripheral smear, complete blood count, family history, and HPLC. Molecular studies were also recommended for the patient.

## Discussion

One prevalent hemoglobinopathy is sickle cell disease. Every year, it affects around 300,000 newborns. The majority of births take place in sub-Saharan Africa and India. The majority of impacted babies born in low- and middle-income countries do not have access to testing for sickle cell disease and other hemoglobinopathies. Without a definitive diagnosis, these babies may die from difficulties in their early years of life. With early diagnosis and low-cost remedies, most deaths can be avoided.[3]

In a systematic review by Rao P. *et al*., the prevalence

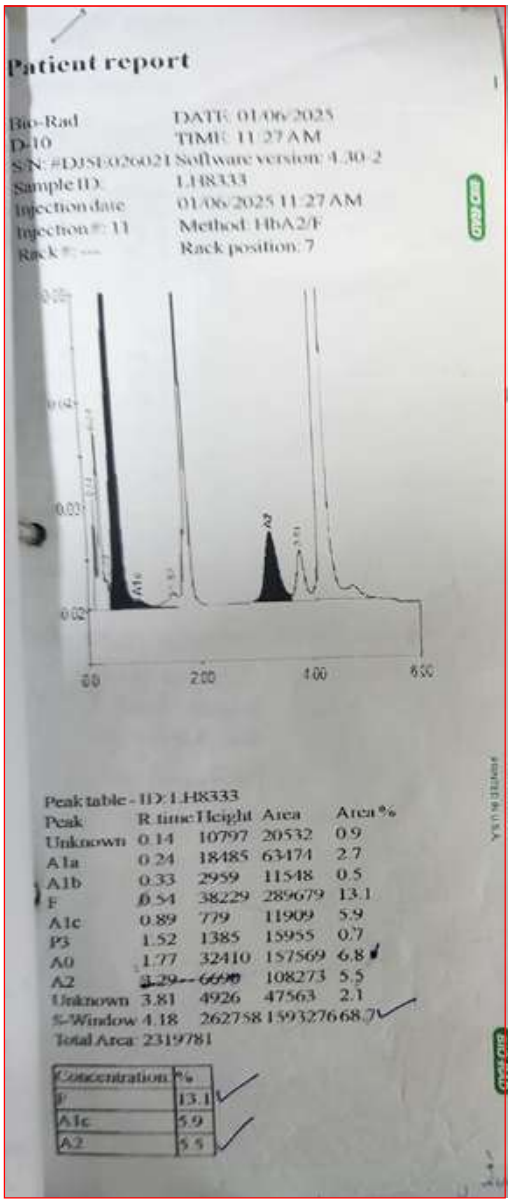


**Figure 3: HPLC Chromatogram**

of sickle cell disease, sickle cell trait, and HbS-beta-thalassemia was noted to be 1.17% (95% CI:0.79%–1.75%), 5.9% (95% CI:3.8%–8.88%) and 0.37% (95% CI:0.17%–0.83%) respectively. Uttar Pradesh and Odisha were found to have the highest prevalence of sickle-beta thalassemia, also reported in Gujarat, Maharashtra and West Bengal.[2] The present case was ofNewDelhi and hadmixed ethnicity. Global human migration can change the distribution of sickle cell disease.[2] Thus, it is important to consider the possibility of sickle-beta thalassemia in patients with recurrent

infections and severe anaemia, even in areas with low prevalence of sickle cell beta-thalassemia.

Foetal haemoglobin (Hb F) levels decrease and sickle haemoglobin (Hb S) starts to predominate during the first six to twelve months of life.[5] At this point, sickle cell disease clinical symptoms start to appear, and a diagnosis needs to be made.[3] Pallor, discomfort, fever, lethargy, and jaundice are all possible symptoms of sickle cell disease. Acute consequences include infections, severe anemia, vaso-occlusive events, pregnancy problems, venous thromboembolism, and stroke.[4] The patient in this instance was two years and eleven months old. She had a fever and was diagnosed with acute anemia.

Sickle β-chain mutations, including sickle β-thalassemia (Hb Sβ0 or Hb Sβ+). The sickle cell disease phenotype is seen in hemoglobin C (Hb SC illness) or thalassemia. Severe symptoms are linked to Hb SS and Hb Sβ0, while Hb SC and Hb Sβ+ are typically present with milder symptoms.[6] At the age of two years and eleven months, the patient in this instance arrived at the hospital with a fever and severe anemia. Molecular tests were recommended when HPLC revealed that she had sickle-beta thalassemia. It can be challenging to differentiate sickle-beta thalassemia from sickle cell anemia with α thalassemia. The diagnosis may be made with the aid of laboratory measures. (Table 1)[1]

**Table 1: Laboratory Differentiation of Sickle Cell Anaemia, Sickle cell anaemia/á-thalassemia and Hb S/β°- thalassemia.**

| Diagnosis | Level variation | | | Mean Hb F (%) |
|---|---|---|---|---|
| | Haemoglobin (g/dl) | MCV (fl) | Hb A2 (%) | |
| SCA | 7-8 | 85-95 | 2.5-3.5 | 5 |
| SCA/α-thalassemia | 8-10 | 70-85 | 3.5-4.5 | 5 |
| Hb S/β° thalassemia | 8-10 | 65-75 | 4-6 | 9 |
| **Present case** | **4.8** | **65.2** | **5.5** | **13.1** |

The MCV and Hb A2 in this instance were similar to the sickle-beta thalassemia reference range given by Figueiredo M. Nevertheless, compared to the reference values stated by Figueiredo M.[1], the Hb level was lower and the Hb F was higher. Clinical severity may be lessened in sickle-beta thalassemia due to elevated Hb F levels.In a study by Mukherjee *et al* 21 sickle-beta thalassemia cases in western India were examined, participants ranged in age from six months to eighteen years. The majority of cases exhibited low hemoglobin levels along with lower MCV and MCH. In the tribal population, the mean Hb was 8.7 ± 0.60 g/dl, while in the non-tribal group, it was 9.1 ± 2.04 g/dl. A 15-year-old boy had the lowest hemoglobin level in their sample, measuring 4.9 g/dl.[7] Yadav R *et al*. found that the mean hemoglobin level of sickle-beta thalassemia in female children under the age of 15 was 7.4 ± 1.5 g/dl.[8] The patient in this instance had a hemoglobin level of 4.8 g/dl and severe anemia (<7 g/dl).

A kid with severe anemia may experience stunted growth, agitation, and sluggishness. Patients with hemoglobinopathies who have hemoglobin levels below 7 g/dl twice in a row, at least two weeks apart, may require frequent blood transfusions. But there shouldn't be any aggravating factors for anemia, such as bleeding or infection.[5,9]

The patient had history of recurring illnesses and fever. Due to immunological deficits brought on by malnutrition, reduced adaptive immunity, splenic dysfunction, and problems in the opsonization of encapsulated organisms, sickle cell disease patients are vulnerable to infections.[6] Infectious pathogens can directly destroy red blood cells or produce hemolyticanemia through immune-mediated processes.[1] The patient may need frequent blood transfusions and has to be monitored.[5] Based on HPLC, sickle-beta thalassemia was proposed as the diagnosis. For additional molecular testing and care, the patient had to be referred to a higher facility.

The global burden illness 2019 study states that 48.7 million disability adjusted life years (DALYS) are caused by sickle cell disease. In 2023, the National Sickle Cell Anaemia Elimination Mission was initiated. It was started with the goal of eradicating sickle cell disease as a public health issue in India prior to 2047. Sickle cell disease can be eradicated through patient management, screening, and prevention.[4]

## Conclusion

Though the prevalence of sickle-beta thalassemia in India is low, the global migration has altered the distribution of the disease in India. Early diagnosis of sickle beta thalassemia is imperative for elimination of the disease. Patients diagnosed with sickle-beta thalassemia can present with severe anaemia and recurrent infections at an early age.

**Financial Support and Sponsorship : Nil**
**Conflict of Interest: Nil**

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