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(CASE REPORT)

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Early presentation of Sickle Cell Disease in a two-month-old infant: A case report

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Abstract

Sickle Cell Disease (SCD) is an inherited haemoglobin disorder that commonly manifests clinically after the first six months of life due to the protective effects of fetal haemoglobin. Early symptomatic presentation in young infants is uncommon and may pose diagnostic challenges, particularly in settings where newborn screening programs are limited. We report the case of a two-month-old male infant from Pwani Region, Tanzania, who presented with severe anaemia and fever. Laboratory investigations revealed severe anaemia, evidence of haemolysis, and haemoglobin electrophoresis findings consistent with homozygous sickle cell disease (HbSS). The infant responded well to blood transfusion, supportive treatment, and subsequent comprehensive sickle cell care. This case highlights the importance of considering sickle cell disease in young infants presenting with anaemia and fever, even before the age at which symptoms are typically expected to occur. Early diagnosis and initiation of comprehensive care are essential to reduce morbidity and improve outcomes.

Keywords: Sickle Cell Disease; Severe Anaemia; Early Infancy; Haemoglobin Electrophoresis; Newborn Screening

1. Introduction

Sickle Cell Disease (SCD) is one of the most common inherited haemoglobin disorders worldwide and constitutes a major public health challenge in sub-Saharan Africa. It results from an autosomal recessive mutation in the β-globin gene, leading to the production of haemoglobin S (HbS), an abnormal variant of adult haemoglobin [1,2]. Under conditions of deoxygenation, infection, or physiological stress, red blood cells containing haemoglobin S (HbS) undergo sickling due to HbS polymerization, resulting in chronic haemolytic anaemia, vaso-occlusive events, and progressive organ damage [1-3].

Clinical manifestations of sickle cell disease are generally uncommon during the first few months of life because fetal haemoglobin (HbF) inhibits haemoglobin S polymerization and red blood cell sickling, thereby providing partial protection against disease manifestations [2,3]. Consequently, most children become symptomatic after six months of age as HbF levels decline [2,3]. However, some infants may present earlier with severe anaemia, recurrent infections, or other complications, particularly in settings where newborn screening programs are unavailable [4,6].

Early diagnosis is essential because timely initiation of preventive care, parental education, vaccination, infection prophylaxis, and regular follow-up can significantly reduce morbidity and mortality in children with sickle cell disease [4,8]. We report a case of a two-month-old infant diagnosed with sickle cell disease after presenting with recurrent severe anaemia and fever, highlighting the importance of maintaining a high index of suspicion for sickle cell disease even in very young infants.

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2. Case Presentation

We report a case of a two-month-old male infant who was referred to Tumbi Regional Referral Hospital in Kibaha, Tanzania, with a history of fever and severe anaemia. The infant had previously been admitted to a district hospital at five weeks of age with severe pallor and fever, where he received one packed red blood cell transfusion before being discharged after clinical improvement.

The child was born at term through spontaneous vaginal delivery following an uneventful pregnancy and delivery. He had a birth weight of 3.2 kg and no history of neonatal complications. The infant was on exclusively breastfeeding and had received age-appropriate immunizations. There was no known family history of sickle cell disease.

At presentation, the mother reported recurrent episodes of fever, progressive pallor, reduced activity, and poor feeding for one week. Physical examination revealed a severely pale and jaundiced infant who appeared ill and febrile. His temperature was 38.4°C, heart rate 168 beats per minute, respiratory rate 48 breaths per minute, and oxygen saturation 96% on room air.

Laboratory investigations revealed severe anaemia with haemoglobin of 5.0 g/dL, haematocrit of 11.9%, red blood cell count of 1.36×10^{12}/L, white blood cell count of 5.18×10^{9}/L, and platelet count of 143×10^{9}/L. Red blood cell indices showed an MCV of 87.5 fL and MCHC of 31.1 g/dL. Differential count demonstrated neutrophils of 45% (1.99×10^{9}/L) and lymphocytes of 50.1% (2.20×10^{9}/L) **(Table 1).**

Peripheral blood smear demonstrated sickle cells, anisocytosis, poikilocytosis, microcytosis, hypochromia, schistocytes, and target cells. White blood cell and platelet morphology were normal **(Table 2),** and blood culture showed no bacterial growth.

Because severe symptomatic anaemia at two months of age is uncommon, haemoglobin electrophoresis was performed. Results demonstrated HbS 93%, HbF 4.9%, HbA_2 2%, and HbA 0%, confirming homozygous sickle cell disease (HbSS) **(Table 3).**

Table 1 Full blood count results at presentation demonstrating severe anaemia with preserved white blood cell and differential counts.

Parameter	Result	Reference Range
WBC	5.18×10^{9}/L	$5.0–19.5 \times 10^{9}$/L
Neutrophils	45% (1.99×10^{9}/L)	20–50%
Lymphocytes	50.1% (2.20×10^{9}/L)	40–70%
Hb	5.0 g/dL	10.5–13.5 g/dL
MCV	87.5 fL	74–108 fL
MCHC	31.1 g/dL	32–36 g/dL
HCT	11.9%	33–39%
RBC	1.36×10^{12}/L	$3.5–5.5 \times 10^{12}$/L
Platelet Count	143×10^{9}/L	$150–450 \times 10^{9}$/L

Table 2 Peripheral Blood Smear

Parameter	**Findings**
Red Cell Morphology	Sickle cells present, with anisocytosis,poikilocytosis,microcytosis,hypochromia,schistocytes, and target cells seen
WBC Morphology	Normal
Platelet Morphology	Normal

Table 3 Abnormal Hemoglobin studies Hb Variants

S/N	**Test Name**	**Specimen**
1	Hb-Electrophoresis	Whole Blood
Doctor's Review *Two months old infant presenting with history of recurrent hospital admissions for fever and severe anaemia and had previously received one blood transfusion during an earlier admission.*		
Results		
S/N	Parameter	Observed Value
1	HbA	0 %
2	HbA_2	2%
3	HbF	4..9 %
4	HbS	93 %
5	Impression	Highly suggestive of Sickle Cell Disease (Homozygous HbS)

The infant was admitted and managed with two packed red blood cell transfusions, intravenous fluids, antipyretics, and supportive care. Following stabilization, he was initiated on folic acid supplementation and oral penicillin prophylaxis according to sickle cell management protocols [4,8]. His clinical condition improved significantly, with resolution of fever and correction of anaemia.

The child was subsequently enrolled in the paediatric haematology clinic for regular follow-up. At nine months of age, hydroxyurea therapy was initiated according to institutional protocols. At the time of writing, he remains clinically stable on hydroxyurea, folic acid, and penicillin prophylaxis, with no further episodes of severe anaemia requiring transfusion.

3. Discussion

Sickle cell disease typically becomes clinically apparent in infancy, usually after 4–6 months of age, as fetal haemoglobin levels decline [2,3]. Clinical presentation of sickle cell disease in a two-month-old infant is therefore unusual and highlights the variability in disease presentation [2,3]. In many low-resource settings where newborn screening programs are not routinely available, diagnosis often occurs only after the onset of classical symptoms [6,9].

The early presentation observed in this case may be explained by the markedly low fetal haemoglobin level (4.9%) despite the young age of the infant. Fetal haemoglobin inhibits erythrocyte sickling and is generally protective during infancy. An earlier decline in fetal haemoglobin (HbF) levels may contribute to earlier clinical manifestations of sickle cell disease, as HbF inhibits HbS polymerisation and reduces disease severity [2,3].

Similar findings were reported by Muzazu et al. in Zambia, who described a 2 months old infant presenting with severe symptoms who was subsequently diagnosed with HbSS disease [10]. Their report emphasized that clinicians should

maintain a high index of suspicion for sickle cell disease in infants presenting with unexplained severe anaemia regardless of age.

Severe anaemia is a major contributor to morbidity and mortality among children with sickle cell disease in sub-Saharan Africa [5,9]. Recurrent haemolysis, infections, nutritional deficiencies, and splenic sequestration crises may contribute to profound reductions in haemoglobin concentration in children with sickle cell disease [2,3,5,9]. In the present case, the infant had already required a previous transfusion and subsequently presented with a haemoglobin level of only 5 g/dL, indicating significant disease severity.

Early diagnosis facilitated the timely initiation of comprehensive sickle cell care, including folic acid supplementation, penicillin prophylaxis, parental counselling, and regular specialist follow-up, all of which are associated with improved clinical outcomes [4,8]. These interventions have been shown to significantly reduce childhood mortality and improve long-term outcomes [4,8]. Furthermore, hydroxyurea therapy initiated at nine months of age is expected to increase fetal haemoglobin production, reduce haemolysis, decrease disease-related complications, and improve quality of life [7]

This case also shows the importance of strengthening newborn screening programs in countries with a high burden of sickle cell disease. Early identification of affected infants facilitates prompt initiation of preventive interventions before the occurrence of severe complications [4,6]. In regions where newborn screening is unavailable, clinicians should consider haemoglobin electrophoresis in infants presenting with recurrent anaemia, jaundice, or unexplained febrile illnesses.

4. Conclusion

This case demonstrates that sickle cell disease may present much earlier than traditionally expected, even during the first few months of life. The occurrence of recurrent severe anaemia and fever in a two-month-old infant led to the diagnosis of homozygous sickle cell disease despite the presumed protective effect of fetal haemoglobin. Clinicians should maintain a high index of suspicion for sickle cell disease in young infants presenting with unexplained anaemia, jaundice, or recurrent infections. Expansion of newborn screening programs and early enrolment into comprehensive sickle cell care services are essential to improve outcomes and reduce disease-related morbidity and mortality in resource-limited settings.

Compliance with Ethical Standards

Acknowledgement

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Disclosure of Conflict of Interest

No conflict of interest to be disclosed.

Statement of Ethical Approval

Ethical approval for publication of this case report was obtained from the relevant institutional authorities at Tumbi Regional Referral Hospital. Written informed consent was obtained from the patient's parents, and all procedures were conducted in accordance with institutional protocols.

Statement of Informed Consent

Written informed consent was obtained from the parents. Confidentiality and anonymity of the patient were strictly maintained throughout the preparation and publication of this report.

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