

Original Research Article

Prevalence of haemoglobinopathies in anaemic children attending the outpatient department of paediatrics in a tertiary care hospital in Dhaka

Farjana Afroze Jui*, S. Serjina Anwar, Mafuza Khanum,
Saleh Mohammad Hasibul Hasan

Department of Paediatrics, Kurmitola General Hospital, Dhaka, Bangladesh

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*Correspondence:

Dr. Farjana Afroze Jui,

E-mail: farjana7665@gmail.com

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ABSTRACT

Background: Anaemia in children is a significant public health issue, particularly in developing countries, where nutritional deficiencies and haemoglobinopathies contribute substantially. Understanding the prevalence and underlying causes are essential for targeted interventions. The aim of the study was to evaluate the prevalence of haemoglobinopathies in anaemic children attending a tertiary care hospital in Dhaka.

Methods: This observational study was conducted over six months, from July 2024 to December 2024, at the Outpatient Department of Paediatrics in Kurmitola General Hospital, Dhaka, Bangladesh. Data were collected from children diagnosed with anaemia during study period. Demographic information, essential clinical features and laboratory findings, including haemoglobin electrophoresis and serum iron profile were analyzed to determine types of anaemia and prevalence of Haemoglobinopathies among them.

Results: Out of 120 anaemic children, 71 (59.2%) were male, and 49 (40.8%) were female, with a male-to-female ratio of 1.45:1. The majority (71.7%) were aged 1–3 years. Consanguineous marriages were reported among 5.8% of the parents. Among haemoglobinopathies HbE Trait was 21.6% while Thalassemia Major, Thalassemia Minor, HbE Disease, and HbE with β -Thalassemia were 1.7%, 4.2%, 1.7% and 4.2% respectively. Iron Deficiency Anaemia (IDA) was the most prevalent condition, accounting for 50% of cases. Additionally, 16.7% of cases were due to other causes, underscoring the diverse etiology of anaemia in children. Notably, 57.5% of Hb E trait cases also had iron deficiency anaemia.

Conclusions: Haemoglobinopathies significantly contribute to childhood anaemia, especially after nutritional deficiencies. The findings highlight the need for routine haemoglobinopathy screening and formulation of public health strategies to address genetic & nutritional causes of anaemia.

Keywords: Anaemia, Iron deficiency anaemia, Haemoglobinopathies, HbE trait, Paediatrics

INTRODUCTION

Anaemia is a major global health issue and a leading contributor to illness and death among children. It is characterized by a reduced number of red blood cells or a haemoglobin level lower than the standard range for a child's age and sex.^{1,2} Adequate haemoglobin is crucial for transporting oxygen to tissues, and its deficiency can

result in symptoms such as lethargy, fatigue, weakness, and shortness of breath. According to the World Health Organization (WHO), the global prevalence of anaemia in children aged 6–59 months is approximately 39.8%.¹

Haemoglobinopathies are the most prevalent single-gene autosomal recessive disorders globally, with wide geographical variation. In Southeast Asia, α -thalassaemia,

β -thalassemia, haemoglobin (Hb) E, and Hb constant spring (CS) are common.³ These hereditary haemoglobin disorders are broadly classified into two groups: thalassemia and haemoglobinopathies.⁴

Thalassemia is characterized by reduced or absent production of one or more globin chains that form the haemoglobin tetramer, making them the most common autosomal recessive disorders worldwide.⁴

Among more than 300 structural variants, Hb E is the second most prevalent haemoglobin disorder globally, caused by a mutation in the beta-globin gene that substitutes glutamic acid for lysine at position 26 in the beta-globin chain.⁵ These inherited haemoglobin disorders are characterized by chronic haemolytic anaemia and often require life-long blood transfusion for survival.⁶

Hb E- β thalassemia is a significant health issue across Southeast Asia, particularly in Thailand, Indonesia, Sri Lanka, Northeast India, and Bangladesh.⁷ National Thalassemia Survey-2024 reveals HB E Trait 65.8%B thalassemia trait 18.65, HB E dis (4.7%) are the leading haemoglobinopathies over the country.⁸

In Bangladesh, E- β thalassemia is the most common haemoglobinopathy among anaemic children, accounting for 68.5% of cases, followed by β -thalassemia major (31%) and Hb E disease (0.5%) among the admitted patient in Thalassemia center of Dhaka Sishu Hospital.⁹

The pathophysiology involves ineffective erythropoiesis, apoptosis, oxidative damage, and shortened red cell survival.¹⁰ While regular blood transfusions can prevent complications associated with ineffective erythropoiesis, they may result in iron overload, causing toxicity in the heart, endocrine glands, and other organs, leading to increased morbidity and mortality.¹¹

The socio-economic impact of thalassemia is profound, affecting not only the child and family but also placing a significant burden on the healthcare system and the community at large.¹² Bangladesh, a South Asian country overall of 6-12% population carry the thalassemia gene.¹³

Although there is no national database, Khan et al estimated that there are approximately 100,000 thalassemia patients in the country, with an annual birth rate of 1,040 β -thalassemia major cases and 6,443 Hb E- β thalassemia cases.¹² Additionally, WHO estimates 4.8 million people carry HB E, which is 4% of population in Bangladesh.^{1,2}

With the improvement of health services children mortality rate is reduced due to infectious disease but still thalassemia is leading cause of many hospitals admission. Prevention is most cost effective than treatment.

Anaemia is a major cause of childhood growth failure as well as poor development. So accurate diagnosis is needed for treatment. Developing a successful prevention and treatment protocol for anaemia requires accurate regional data on the prevalence and socio-demographic distribution of haemoglobinopathies and other causes of anaemia in children.

The objective of this study was to assess the current prevalence and distribution of haemoglobinopathies among anaemic children in Bangladesh. By understanding the burden and distribution of these disorders, this study seeks to inform strategies for prevention and management, ultimately reducing the health burden in the country.

METHODS

This observational study was conducted over six months, from July 2024 to December 2024, at the Outpatient Department of Paediatrics in Kurmitola General Hospital, Dhaka, Bangladesh. A total of 120 newly diagnosed anaemic children were enrolled based on predefined inclusion and exclusion criteria. Children aged 1year–12 years with newly diagnosed anaemia according to WHO guidelines were included, while those younger than 1 year or older than 12 years, previously treated for anaemia, previously diagnosed with haemoglobinopathy, or suffering from chronic diseases were excluded.

Data were collected using a structured proforma, which documented demographic information such as age and sex, as well as clinical details including haemoglobin levels (Hb%) and the types of anaemia or haemoglobinopathies diagnosed. Haemoglobin levels were measured at the time of enrolment, and additional laboratory tests were performed to identify specific haemoglobinopathies or other underlying causes of anaemia.

The collected data were entered into a predesigned datasheet and analyzed using SPSS software version 19. Descriptive statistics such as frequencies and percentages were used to summarize the findings, while relevant clinical characteristics were examined to identify the prevalence and distribution of haemoglobinopathies and other causes of anaemia within the study population. The study was approved by the Institutional Ethics Committee.

This methodology ensured a systematic approach to data collection and analysis, enabling a comprehensive assessment of haemoglobinopathies as a cause of anaemia in children attending the outpatient department.

RESULTS

Figure 1 shows the sex distribution of study participants, where out of 120 individuals, 71 (59.2%) were male, and 49 (40.8%) were female. The male-to-female ratio was

1.45:1, with males forming the majority of the participants in this study.

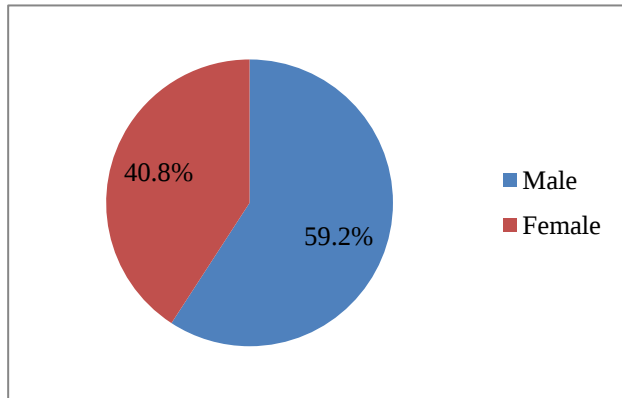


Figure 1: Sex distribution of our study participants (N=120).

The majority of the participants (71.7%) were in the 1–3 years age group, followed by 18.3% in the 4–6 years group. Only 5.8% and 4.2% of the participants belonged to the 7–9 years and 10–12 years age groups, respectively. This indicates that most of the anaemic children in the study were in the younger age group (1–3 years).

Table 1: Age distribution of our study participants (n=120).

Age group (in years)	Frequency (N)	(%)
1–3	86	71.7
4–6	22	18.3
7–9	7	5.8
10–12	5	4.2
Total	120	100

Table 2: Prevalence of consanguineous marriage among parents of anaemic children (n=120).

Category	Frequency (N)	(%)
Consanguineous marriage	7	5.8
Non-consanguineous marriage	113	94.2
Total	120	100

Table 3: Distribution of haemoglobinopathies and other causes (n=120).

Condition	Frequency (N)	%
Iron deficiency anemia (IDA)	60	50
HbE trait	26	21
Thalassemia major	2	1.7
Thalassemia minor	5	4.2
HbE disease	2	1.7
HbE with β -Thalassemia	5	4.2
Other causes	20	16.7
Total	120	100

Table 2 presents the prevalence of consanguineous marriage among the parents of anaemic children in the study. It shows that 5.8% of the parents had consanguineous marriages, while the majority (94.2%) was non-consanguineous.

Table 3 shows the most commonly identified condition was iron deficiency anemia (IDA), accounting for 50% of the cases. HbE Trait was observed in 21% of participants. Other conditions included thalasaemia major (1.7%), thalassemia minor (4.2%), HbE disease (1.7%), and HbE with β -Thalassemia (4.2%). Additionally, 16.7% of cases were attributed to other causes.

Table 4: Association of IDA with Hb E trait children (n=26).

Category	Frequency (N)	%
Without IDA	11	42.5
With IDA	15	57.5
Total	26	100

Table 4 shows 57.5% of HbE trait patients also had iron deficiency anaemia while 42.5% patients had no IDA.

DISCUSSION

Anaemia in children is a global public health issue with significant morbidity and mortality, particularly in low- and middle-income countries. Our study revealed a prevalence of anaemia primarily in younger children, with 71.7% of participants aged 1–3 years, followed by 18.3% in the 4–6 years group, and much lower proportions in older age groups (5.8% in 7–9 years and 4.2% in 10–12 years). This predominance of anaemia in younger children aligns with previous studies, which attribute it to higher nutritional demands and vulnerability to nutritional deficiencies during early childhood.^{14–16}

The sex distribution in our study showed that males formed the majority of participants (59.2%) with a male-to-female ratio of 1.45:1). This male predominance is consistent with findings from other regional studies, such as those by Khan et al, reported similar trends, potentially reflecting healthcare-seeking behavior that favors male children in certain socio-cultural contexts.¹² The gender distribution in our study revealed a higher prevalence of thalassemia syndromes among males, with a male-to-female (M/F) ratio consistent with findings from other studies.^{9,10} This may reflect either a genuine higher incidence of thalassemia among males in this region or sociocultural gender biases, where male children receive more attention and healthcare access.

Consanguineous marriage was reported in 5.8% of anaemic children which was supported by a study conducted by Hosen et al who reported 6.64% of consanguine marriage in Bangladesh.¹⁷ Among the haemoglobinopathies, HbE Trait, with or without IDA

was observed in 21% of participants, while other conditions included thalassemia major (1.7%), thalassemia minor (4.2%), HbE disease (1.7%), and HbE with β -Thalassemia (4.2%). These findings underscore the significant contribution of haemoglobinopathies to the anaemia burden in Bangladesh.¹⁸ Iron deficiency anaemia (IDA) was the most common condition identified in our study, accounting for 50% of the cases. This is in line with global data and local studies, which consistently highlight IDA as the leading cause of anaemia in children.

Our study also noted that 16.7% of anaemia cases were attributed to other causes, reflecting the multifactorial nature of anaemia in children. Previous studies, such as by Paul et al, in Bangladesh, have similarly highlighted the diversity of causes, ranging from nutritional deficiencies to genetic and environmental factors.¹⁹

The significant burden of haemoglobinopathies observed in our study, particularly the high prevalence of HbE Trait with or without IDA, aligns with earlier reports by Tahura et al, emphasizing the importance of targeted screening and prevention strategies in regions where such haemoglobinopathies are endemic.⁹

Rare haemoglobinopathies, such as HbE disease and HbE with β -thalassemia, were identified in our study. While these were less common, they represent clinically significant conditions that warrant attention. The study by Corrons et al., highlights the importance of advanced diagnostic tools for rare haemoglobinopathies, a sentiment echoed by our findings, as these conditions often present with severe anaemia and systemic complications.²⁰

In our study Hb-E trait was the most common haemoglobinopathy consistent with the findings of Hasan et al., who reported Hb-E trait (10.67%) as predominant disorder in a study conducted in ICDDR. Hb-E trait was also found as a leading hereditary haemoglobin disorder in 36% of tribal population of Bangladesh in a study conducted by Shannon et al. In another study from Olivieri et al, revealed Hb-E disorders were most common form of haemoglobinopathies in south east Asia including Bangladesh.^{7,22}

This study found E-B thalassemia was second most common haemoglobinopathy in anaemic children. While a study conducted in thalassemia center of Dhaka Sishu Hospital by Tahura et al reported E-B thalassemia was the major portion about 68.50% of Thalassemic patients.⁹ Probably this difference was due to different study place and population.

Thalassemia major and HbE disease were the least common among anaemic children. This contrasts with findings by Aziz et al, who reported 3.06% Thalassemia Major in BD and 1.4% HbE disease in BD. Another study by Noor et al., also reported this finding.^{23,24} They found

0.16% Hb E disease in Bangladesh. We also found that more than half of the children of HbE trait also had IDA. These findings were included in HbE trait. Our study showed haemoglobinopathies were the important cause of anaemia in children. So, we should give more emphasize on diagnosis of it for better management of anaemia.

Future studies should be conducted to explore the genetic mutations responsible for Thalassemia and other hemoglobinopathies in Bangladesh to develop targeted screening and diagnostic tools.

This study was conducted based on laboratory findings of anaemia. So, sample size was limited which may not fully represent the prevalence and characteristics of hemoglobin disorders in the general population. Additionally, factors like consanguinity and other socio-demographic aspects could not be explored in depth due to data constraints.

CONCLUSION

Our study emphasized the distribution of haemoglobinopathies as a cause of anaemia in children (1–12 years). Hb E trait was the most prevalent haemoglobinopathy which was followed by HbE-B thalassemia and thalassemia minor. Thalassemia major and HbE disease were least common among anaemic children.

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Ethical approval: The study was approved by the Institutional Ethics Committee

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