Original Article

Hemoglobinopathies in a tertiary care hospital in Bangladesh: a retrospective study

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ABSTRACT

Background: *A hereditary condition known as hemoglobinopathies causes one of the globin chains of the hemoglobin molecule to have an abnormal structure. Worldwide, inherited hemoglobin abnormalities are common. Studies on thalassemia and hemoglobinopathies are scarce in Bangladesh. Even though this country had not done much research on the subject, the number of people with inherited hemoglobin problems is alarming. This study was conducted to assess the forms of inherited hemoglobin abnormalities among the patients of a tertiary care hospital.*

Methods: *A total of 431 individuals were recruited in this study. The samples were collected from both outdoor and indoor patients of BIRDEM General Hospital, BIRDEM General Hospital (Women and Children), Bangladesh Institute of Health Sciences (BIHS) from January to June 2024. The patients were referred to these hospitals from different regions of the country. The blood samples collected from them were analyzed at the Department of Laboratory Medicine, BIRDEM General Hospital Dhaka.*

Results: *Out of 423 cases, 268 (63.3%) blood samples showed normal findings and 155 (36.7%) blood samples showed different forms of haemoglobinopathies in hemoglobin electrophoresis. Out of 155 abnormal cases, 58 (37.4%) were males and 97 (62.6%) were females. The most common form was HbE trait (20.09%) followed by â-thalassemia trait (13.71%). Moreover HbE disease was found in 0.95%, Hb D Punjab trait 0.95%, E-β-thalassemia in 0.47%, β-thalassemia major in 0.47%.*

Conclusion: *It is expected that public health officials will find the current data useful in adjusting current facilities to lower the number of thalassemia carriers and haemoglobinopathies in Bangladesh.*

Key words: *hemoglobinopathies, thalassemia, thalassemia carriers.*

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INTRODUCTION

Inherited hemoglobin diseases are becoming a global public health concern. These conditions constitute a heterogeneous group of mendelian disorders known as hemoglobinopathies[1]. They are distinguished by structurally aberrant hemoglobin variations and thalassaemia, which is caused by partial or complete suppression of normal hemoglobin molecule peptide chains[2,5]. One of three main conditions can cause aberrant hemoglobin: structural flaws in the hemoglobin molecule, thalassemias, which are reduced production of one of the hemoglobin molecule's two subunits, or inappropriate association of otherwise normal subunits. The majority of these are caused by one or more globin chains having a single amino acid substition[3].

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Every year, an estimated 320,000 newborns with a clinically significant hemoglobin disease are born worldwide (83% with sickle cell disorders and 17% with thalassaemias)[4]. Almost 80 percent of these births take place in underdeveloped nations. Clinical signs of hemoglobinopathies can result from homozygous or compound heterozygous conditions between certain variations, placing a financial, social and psychological strain on the family, particularly in low-resource environments[5]. According to global statistics, hemoglobin problems account for approximately 3.4% of deaths in children under the age of five[6].

Due to high rate of international migration, several types of haemoglobinopathies are spreading to non-endemic parts of the world. Most information on thalassemia in South Asia comes from studies which had been conducted in India[7]. Beta-thalassemia carriers are estimated to constitute between 2.78 to 4% of the population in India, or approximately 30 to 48 million people and between 5 to 7% of the population in Pakistan, or around 5 to 12 million people[8].

There is a dearth of population-based data on the prevalence of hemoglobinopathies in Bangladesh. According to the sole published study on the prevalence of thalassemia among Bangladeshi school children (n = 735), the beta-thalassemia trait was 4.1% and the HbE trait was 6.1% [5,8]. According to a different estimate from the World Health Organization (WHO), 4% of Bangladeshis are hemoglobin E (HbE) carriers and 3% of the population are beta-thalassemia carriers[9].

There are about 700 known variations of hemoglobin that involve genes from both the alpha and beta gene clusters. From the alpha-globin gene cluster, or the beta-globin gene cluster, two distinct variations can occasionally be inherited. They refer to this state as compound heterozygosity[10]. Therefore, inherited diseases of hemoglobin production are a major global source of morbidity and mortality[11]. Patients, their families and even their communities are heavily burdened by them. Although they are mostly incurable, prenatal diagnosis, genetic counseling and population screening can help to prevent them. The purpose of the study is to assess the different forms of hereditary hemoglobin problems and their range and distribution of hemoglobinopathies in Bangladeshi community.

METHODS

Study subjects: A total of 431 individuals were recruited in this study. The samples were collected from both indoor and outdoor patients of BIRDEM General Hospital, BIRDEM General Hospital (Women and Children), BIHS during January to June 2024. The patients were referred to these hospitals from different regions of the country. The blood samples collected from them were analyzed at the Department of Laboratory Medicine, BIRDEM General Hospital, Dhaka.

Blood samples: 3 ml intravenous blood samples were collected using EDTA (ethylene diamine tetra acetic acid) as anticoagulants by disposable syringes and needles from each individual by maintaining aseptic precaution.

Haemoglobin electrophoresis: Haemoglobin electrophoresis was carried out by automated minicap sebia flex piercing instrument by capillary electrophoresis.

Data processing and analysis: After collection of all the required data, these were checked, verified for consistency and tabulated using the SPSS (v. 21) software. Statistical significance is set as 95% confidence level at 5% acceptable error level. Data were presented as the proportion of valid cases for discrete variables and as means ± standard deviations and/or medians with interquartile ranges for continuous variables. Differences and/or relation in baseline characteristics were compared using necessary statistical test. A p value of <0.05 was considered significant.

RESULTS

Out of 423 cases, 268 (63.3%) blood samples showed normal findings and 155 (36.7%) blood samples showed different forms of hemoglobinopathies in hemoglobin electrophoresis. Table I depicted the electropherograms of different types of haemoglobinopathies. The most common form was HbE trait (20.09%) followed by β-thalassemia trait (13.71%). Moreover HbE disease was found in 0.95%, Hb D Punjab trait 0.95%, E-β-thalassemia in 0.47%, β-thalassemia major in 0.47%.

Table II shows distribution of the study population by age, it was observed that almost half (50.59%) population was between 30-60 years. The minimum age was found 1 year and maximum was 80 years.

It is apparent from the table III that majority of the cases of haemoglobinopathy belong to reproductive age group 30-60 years (51.61%) followed by less than 10 years (20.64%) and only 41(10.96%) cases were found who were > 60 years of age.

Table I. Distribution of patients with hemoglobinopathies (n=423)

Findings of hemoglobin electrophoresis	Male	Female	Total	Percentage (%)
Normal findings	122	146	268	63.3
β-thalassemia trait	21	37	58	13.71
β-thalassemia major	02	00	02	0.47
Hb E Trait	24	61	85	20.09
Hb E Disease	01	03	04	0.95
E-β-thalassemia	01	01	02	0.47
Hb D Punjab trait	03	01	04	0.95
Total	174	249	423	100(%)

Table II. Distribution of the study population by age (n=423)

Age (Years)	Number of patients	Percentage(%)
≤10	103	24.34
10-30	65	15.36
30-60	214	50.59
>60	41	9.69

Table III. Distribution of age of the study population by findings in hemoglobin electrophoresis (n=423)

Age (Years)	Number of haemoglobinopathies	Number of normal findings
≤10	32 (20.64%)	71 (26.49%)
10-30	26 (16.77%)	39 (14.55%)
30-60	80 (51.61%)	134 (50.00%)
>60	17 (10.96%)	24 (8.95%)
Total	155 (100%)	268 (100%)

Table IV shows sex distribution of the study patients, Out of 155 abnormal cases, 58 (37.42%) were males and 97 (62.58%) were females. Here, P value was found <0.05, which was significant.

Table IV. Distribution of the study population by sex (n=423)

Sex	Haemoglobinopathy found in hemoglobin electrophoresis	Normal findings in hemoglobin electrophoresis	Total	P value
Male	58	102	160	<0.05
Female	97	166	263	
Total	155	268	423	

p-value is determined by chi-square test. P <0.05 is significant.

DISCUSSION

Haemoglobinopathies are hereditary erythrocyte production abnormalities that are common in the Mediterranean region, the Middle East, the Indian subcontinent, and some regions of Southeast Asia. Although the precise number of hemoglobinopathies and their range in Bangladesh is unknown, it appears to be rising[2,12]. The sickness could develop into a pandemic if it keeps spreading vertically[13]. The only method to avoid the disease is to identify carriers and raise public awareness of this new epidemic, as currently no viable therapy is found[14]. This study, therefore, provides an informative data on the pattern of hemoglobinopathies in Bangladesh.

Present study was conducted on 423 individuals. The samples were collected from both indoor and outdoor patients of BIRDEM General Hospital, BIRDEM General Hospital (Women and Children), BIHS during January

to June 2024. Among the 423 cases studied, 268(63.3%) cases had a normal Hb electrophoresis pattern and 155 (36.7%) had haemoglobinopathies (Table I). Out of 155 abnormal cases, 58 (37.42%) were males and 97 (62.58%) were females.Sharma et al; 2020 found haemoglobinopathy among male to female ratio of 2.5:1[14].On the other hand, Rakib et al; 2023 found no association between the type of thalassaemia with age and sex of the subjects[15].

Most common haemoglobinopathies observed were Hb E trait (20.09%), β- thalassemia trait (13.71%), Hb E disease (0.95%), E-β-thalassemia (0.47%) and β-thalassemia major (0.47%) . Several other studies also have documented such high frequency of these haemoglobin formation disorders in neighbouring and other Southeast Asian countries[16]. Genetic counselling for couples at risk for oûspring with homozygous β-thalassemia may be done at this stage.

Hb E-β-thalassemia was detected in 0.46% of the overall study population. E-β-thalassemia patients inherit one gene for β-thalassemia and one gene for Hb E disease. Many other studies also reported E-β-thalassemia as the commonest form of thalassemia in southeast Asia[3,17].

Hb E trait was detected in 20.09% and Hb E disease in 0.95% of the overall study population. Homozygotic and heterozygous conditions of a β-chain variation, in which glutamic acid replaces a lysine residue, are known as HbE illness and E trait. Additionally, this monogenic disease is widely distributed across the Indian subcontinent, the Middle East, and the Mediterranean. On chromosome 11, the gene that regulates the formation of delta chains is situated extremely near to the beta gene. The deletion of one gene may have an impact on the other[17].

Those with African descent and Indian tribal people are most likely to suffer from sickle cell disease. In the USA, the sickle gene carrier rate is 1 in 10, and in Canada, where the majority of the black population is made up of people from the Caribbean and Africa, it might be greater. Conversely, Hb D is primarily found in Iran, Pakistan, and northwest India[18]. In this study, we found Hb D Punjab trait in 0.95% of study population.

Conclusion

In this study, the majority of the patients had Hb E trait. However, other disorders like β-thalassemia trait, Hb E disease and β-thalassemia major were not uncommon. A statewide screening program including more advanced methods such as polymerase chain reaction (PCR) followed by direct sequencing, genetic counseling and public awareness-raising is required to successfully stop the disease's spread in Bangladesh.

Authors' contribution: TN prepared the study design, collected data, writing the manuscript, SM helped in draft and laboratory work and GSS participated in overall supervision. All authors read and approve the final version for submission.

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