



## PREVALENCE OF VARIOUS HEMOGLOBINOPATHIES USING HIGH-PERFORMANCE LIQUID CHROMATOGRAPHY (HPLC): A RETROSPECTIVE CROSS SECTIONAL INSTITUTIONAL STUDY

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### ABSTRACT

**Background:** Hemoglobinopathies are widely used in the world and form an important health burden to the population in India especially in the northeastern part where HbE and b-thalassemia are very common. “High-performance liquid chromatography (HPLC)” is a dependable and effective reference “tool of screening and” characterizing hemoglobin variants.

**Objectives:** The aim of the study was to find the prevalence and the distribution of different hemoglobinopathies” through HPLC in a tertiary care center and to evaluate the demographic and hematological profile of hemoglobinopathies.

**Materials and Methods:** This was a retrospective cross-sectional study done in the Department of Pathology, Dhubri Medical College, and” a time span of one year. There were 237 patients with hemoglobin variant analysis carried out using HPLC. The laboratory records provided demographic information, hematological parameters and the HPLC results. Cases were grouped into b-thalassemia trait (BTT), HbE trait, HbE disease, compound heterozygous conditions, sickle cell trait (SCT), borderline findings, Hb Lepore trait and normal phenotypes. The analysis was done using descriptive statistics.

**Results:** Among 237 cases (138 (58.2) were abnormal hemoglobins and 99 (41.8) were normal. The women formed 58.6 percent and men 41.3 percent. The abnormality that occurred most frequently was HbE trait (19.8%), with b-thalassemia trait (12.2%), HbE disease (8.9%), borderline results (11.0%), and compound heterozygous states (5.1%). One sickle cell trait (0.4) and two Hb Lepore trait (0.8) were detected. Most of the cases were in the age bracket of 21- 30 years.” The distribution based on community was more prevalent in Muslims (53.1) than in Hindus (46.8). Almost 45.5 percent of the patients were found to have a hemoglobin level that was less than 7 g/dL which means that there was a high level of anemia in the study population.

**Conclusion:** Hemoglobinopathies are very common among the population under study and HbE related disorders are the most common one. HPLC is a useful screening technique used in the early detection and classification of hemoglobinopathies. Premarital and antenatal screening is suggested to help reduce the burden of diseases in Northeast India by means of regional screening programs.

**Keywords:** Hemoglobinopathies, HPLC, Hbe Trait, B-Thalassemia Trait, Northeast India, Anemia, Screening.

### INTRODUCTION

Hemoglobinopathies are a family of inherited diseases that are associated with” the defect in the structure or production of hemoglobin that cause high morbidity and mortality across the globe.<sup>1,12,13</sup> They encompass thalassemias and structural forms of hemoglobin e.g. HbS, HbE and HbD. India is also among the most significant concentrations of hemoglobinopathies with a carrier ratio of 3-17 percent of b-thalassemia and a high level of HbE in the northeast states.<sup>1,2,8</sup>

Northeast India, Assam included, is known to be one of the hot spots of HbE and its associated disorder because of ethnic diversity and endogamy.<sup>7,8</sup> These diseases are leading causes of chronic anemia, dependency of transfusion and health care burden.<sup>12</sup> Laboratory screening is essential to genetic counseling, prevention of severe disease, and proper clinical management through the early identification of the disease.<sup>6,12</sup>

“High-performance liquid chromatography (HPLC) is a fast, automated, and sensitive technique” of identifying and determining the quantity of hemoglobin fractions including HbA, HbA2, HbF, and abnormal hemoglobin.<sup>4,5,11</sup> HPLC is a more preferable diagnostic modality in the tertiary care lab as compared to conventional electrophoresis because it gives the lab better resolution and reproducibility.<sup>4,11</sup>



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Although the prevalence rate of hemoglobinopathies has been known to be high in northeast India, there is insufficient institutional data of western Assam.<sup>8,9</sup> Thus, the current research was conducted to determine the prevalence and range of hemoglobinopathies in Dhubri tertiary care center with HPLC and examine their demographic and hematological profiles.<sup>4,5</sup>

## MATERIALS AND METHODS

### Study Design and Setting

The study was a cross-sectional study which was carried out in Central Clinical Laboratory, Department of Pathology, Dhubri Medical College, Assam, over a period of one year.”

### Study Population

The selection included 237 patients whose blood samples were analysed to determine the hemoglobin variant using HPLC within the study period.

### Inclusion Criteria

Every patient with the HPLC testing of hemoglobinopathies.

Access to full-demographic and laboratory data.

### Exclusion Criteria

Incomplete records

Samples of the same patient (first sample used).Data

Collection

Data were extracted out of laboratory HPLC records and comprised:

Age

Gender

Community

Hemoglobin level

RBC indices (MCV, MCH, MCHC)

HPLC parameters (HbA0, HbA2, HbF)

### Final Diagnosis

Normal Type HbA (95-98%) and HgA2 in the reference range (2-3.4%) and HgF less than 1 % in adults with no unusual forms of hemoglobin identified in HPLC.

b-Thalassemia Trait (BTT): HbA2 ( $\geq 4$ ) band supportive red cell indices.

HbE Trait (Heterozygous HbE): This is the presence of separate HbE peak where HbA is the dominant peak.

HbE Disease (Homozygous HbE): HbE with greater than 90 %

Compound Heterozygous States: Presence of multiple abnormal peaks.

Borderline (Indeterminate): With no conclusive signs of b-thalassemia trait, HbA2 of the borderline range (around 3.5- 3.9) and red cell indices are correlated.

Sickle Cell Trait (SCT): Both HbA and HbS will be present on the HPLC with HbA being the major in comparison to HbS.

**Hb Lepore Phenotype:** HbA2 window HPLC shows a typical peak of Hb Lepore variant,

### Statistical Analysis

The descriptive statistics were used to analyze the data. Categorical variables were computed in frequencies and percentages” and evaluated in terms of age, gender, hemoglobin levels, and communities.

### Ethical Considerations

The Institutional Ethics Committee of Dhubri Medical College gave its approval to this study in terms of ethics. Confidentiality of the patients was highly taken into consideration since retrospective lab data were only used.

## RESULTS

Among the 237 cases that were analysed, 138 (58.2) were found to have abnormal hemoglobin patterns and 99 (41.8) had normal HPLC patterns.

### Gender-wise Distribution

The highest number of cases were among the females with 139 cases (58.6), and in the male population, the number is 98 cases” (41.3). In the abnormal cases 55.7 percent were females and 44.2 percent males, meaning that a rate of detection was slightly higher in females.

### Various Hemoglobinopathies Distribution

The most frequently occurring abnormality was HbE trait (19.8) in 47 cases, and b-thalassemia trait (12.2) in 29 cases. HbE disease was identified in 21 (8.9) and 12 (5.1) cases as the state of compound heterozygous. In 26 cases (11.0%), borderline results were found. Uncommon ones were one instance of sickle cell trait (0.4%), and two instances of Hb Lepore trait (0.8%).

### Age-wise Distribution

Hemoglobinopathy cases were most in 21-30 years followed by 11-20 years. The cases of HbE trait and BTT were also very large in pediatric cases (1-10 years), which implies early onset of hereditary diseases.

### Hemoglobin Level and Diagnostic Classifications.

The percentage of patients with hemoglobin levels less than 7 g/dL was huge (45.5%), which indicates severe anemia. Among them, BTT, HbE trait, HbE disease and compound heterozygous states usually were linked to low hemoglobin levels. Hemoglobin of greater than 10 g/dL was found in only 13.5% of cases.

### Community-wise Distribution

Community-wise analysis showed that, 126 cases (53.1) were under Muslim community and 111 cases (46.8) were under Hindu community. HbE trait and BTT were common in both communities with somewhat more cases of borderline and compound heterozygous among Muslims.

## TABLES

1. Gender Wise Distribution of Hemoglobinopathies”

“Gender”	“Total no. of cases”	Total percentage (%)	Cases with abnormal Hb patterns	Percentage (%) of cases with abnormal Hb patterns
Male	98	41.3%	61	44.2%
Female	139	58.6%	77	55.7%
Total	237	100%	138/237	58.2%

2. Gender Wise Distribution of Various Hemoglobinopathies

GENDER	BTT	HbE trait	HbE disease	Compound heterozygous	SCT	Borderline results	Hb lepore trait	Normal study	Total
Male	11	20	9	6	0	14	1	37	98
Female	18	27	12	6	1	12	1	62	139
Total cases	29	47	21	12	1	26	2	99	237

3. “Age Group- Wise Distribution of Hemoglobinopathies”

Age group (years)	(BTT)	HbE trait	HbE disease	Compound heterozygous for HbE	Sickle Cell Trait (SCT)	Borderline results	Hb lepore trait	Normal study
1-10	5	12	2	2	1	4	0	16
11-20	5	11	6	3	0	10	1	18
21-30	8	10	5	5	0	5	0	28
31-40	2	3	4	1	0	1	0	11
41-50	4	4	2	0	0	4	0	15
51-60	4	3	1	1	0	1	1	6
>60	1	4	1	0	0	1	0	5
Total	29	47	21	12	1	26	2	99

4. Hemoglobin Levels and Distribution of Diagnostic Categories”

Hb (%)	Total cases (n)	% of cases	BTT	HbE trait	HbE disease	Compound heterozygous for HbE	SCT	Borderline	Hb Lepore	Normal study
<7	108	45.5%	28	18	18	12	0	10	2	20
7-10	97	40.95%	0	24	3	0	1	12	0	57
>10	32	13.5%	1	5	0	0	0	4	0	22
Total	237	100%	29	47	21	12	1	26	2	99

5. Community Wise Distribution of Hemoglobinopathies

Community	Total cases (n)	% of cases	BT T	Hb E trait	HbE disease	Compound heterozygous for HbE	SC T	Borderlin e	Hb Lepor e	Norma l study
Muslims	126	53.1%	13	19	11	8	1	20	1	53
Hindus	111	46.8%	16	28	10	4	0	6	1	46
Total	237	100%	29	47	21	12	1	26	2	99

**DISCUSSION**

Hemoglobinopathies are one of the significant social and economic burdens in India because of their heterogeneous irregularities based on ethnicities, geography and social practices.<sup>1,11</sup> The current hospital-based research study in the Dhubri Medical College during a year offers an insight into the range of hemoglobinopathies in Western Assam where the

published information is still scarce.<sup>8</sup> This high percentage of aberrant forms of hemoglobin in our sample indicates the abundance of inherited hemoglobin diseases in tertiary care referral facilities.<sup>4,5</sup>

In the current study, there were many cases with abnormal hemoglobin variants and this is similar to other tertiary care based studies in India where

samples are mostly referred to be examined with regards to anemia and suspected hemoglobinopathies.<sup>4,5</sup> These results have been echoed by Sachdev R et al<sup>4</sup> and Rao S et al<sup>5</sup> who found a high rate of abnormal hemoglobin fractions in their hospital based HPLC studies in North India.<sup>4,5</sup> Such studies have higher levels of detection and this is probably because of referral bias since most of the cases involved are clinically suspected or screened groups.<sup>4,5</sup>

The factor that is leading to the high prevalence of hemoglobinopathies, as was observed in our study is the fact that inherited hemoglobin-based disorders in India are very widespread.<sup>1</sup> Mohanty D et al.<sup>2</sup> in their multicentric study on six Indian cities showed a strong regional difference in the “prevalence of b-thalassemia and other hemoglobinopathies, highlighting the effect of ethnic and” geographical differences.<sup>2</sup>

On comparison with other studies done in the Northeast of India, we find that there is a similarity in the hemoglobin variant distribution. In an HPLC-based investigation in the state of Upper Assam, Baruah MK et al.<sup>8</sup> reported the HbE-related disorders as the primary hemoglobinopathy and b-thalassemia trait and other variants as the subsequent ones.<sup>8</sup> Moreover, Deka R et al.<sup>7</sup> have reported that HbE is very common among the northeastern people and that is why disorders related to HbE are mostly observed in the current study.<sup>7</sup> These parallels suggest that the range of hemoglobinopathies in Western Assam follows the conventional epidemiological picture of the Northeast India.

However, Western and Northern Indian studies have shown a distribution pattern otherwise. The most frequent abnormality was the b-thalassemia trait reports by Madan N et al.<sup>3</sup> in India with HbE being somewhat less prevalent in the North and West of the region.<sup>3</sup> Similarly, Chatterjee T et al.<sup>14</sup> in Eastern India found a wide range of hemoglobinopathies with b-thalassemia trait having a significant contribution.<sup>14</sup> These regional variations can be explained by the difference in ethnic composition, genetic background, and marriage practices across “different parts of the country”.<sup>2,7</sup>

Regarding the public health, the current results highlight the importance of specific screening measures in the areas of high prevalence. It has been pointed out by Colah R et al.<sup>1</sup> that antenatal screening, premarital counseling, and early detection programs have an important role in preventing the burden of severe hemoglobinopathies in India.<sup>1</sup> The immense carrier pool and the chronic morbidity of these disorders necessitate early diagnosis of these disorders by HPLC in tertiary care hospitals such as the Dhubri Medical College in disease prevention and disease management.<sup>4,11</sup>

The greatest strength of the study is that the study gives region-specific data of Western Assam where

there is a dearth of literature on the spectrum of hemoglobinopathies. Nevertheless, the study was a hospital-based study and thus, the findings may not be generalizable to the population since it may refer to referral bias and community-based screening as the limitation of external validity, which is also observed in the research by Rao S et al.<sup>5</sup> and Sachdev R et al.<sup>4</sup>

In general, the range of hemoglobinopathies identified in the current research is similar to the rest of Indian tertiary care studies and clearly reflects the epidemiological pattern of Northeast India, which is a greater prevalence of HbE-related diseases and mixed hemoglobin variants, as reported by Baruah MK et al.<sup>8</sup>, and Deka R et al.<sup>7</sup>. This underscores the need to implement region-specific diagnostic and preventive measures to alleviate the prevalence of hemoglobinopathies in this region in the country.<sup>1</sup>

#### Limitations

As a retrospective study that is lab-based, the results might not indicate the actual prevalence in the community. Absence of molecular confirmation and full clinical correlation are other drawbacks.

#### CONCLUSION

The current research indicates a high incidence of hemoglobinopathies in patients undergoing HPLC testing in Dhubri Medical College wherein HgE trait has been the most frequent trait and b-thalassemia trait and HgE disease comes in the next sequence. Considerable health imposition of anemia and early age activity highlights the importance of routine screening, genetic counseling and public health awareness interventions in Northeast India. Early detection and classification of hemoglobinopathies in tertiary care is an effective and reliable modality that HPLC can perform.

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#### Conflict of Interest

None declared.

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