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Original Research Article

Assessing Hemoglobinopathy Occurrence via High-Performance Liquid Chromatography in a Tertiary Care Setting

Darshanaben Kanabhai Gohel[1], Nishant Pujara[2], Sandip Patel[3]

[1]Junior Resident, Department of Physiology, GMERS Medical College, Himmatnagar, Gujarat, India
[2]Profesoor, Department of General Medicine, Gujarat Adani Institute of Medical Sciences, Bhuj, Kutch, Gujarat, India
[3]Assistant Professor, Department of General Medicine, GMERS Medical College, Dharpur,Patan, Gujarat, India

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Corresponding author: Dr. Sandip Patel
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Abstract

Background: Hemoglobinopathies are among the most common inherited disorders worldwide, with a significant burden in India. High-performance liquid chromatography (HPLC) has become the gold standard for detecting and classifying these disorders, offering precise quantification and identification of hemoglobin variants.

Aim: To estimate the prevalence and distribution of various hemoglobinopathies detected by HPLC in a tertiary care center in India.

Material and Methods: This cross-sectional observational study included 310 patients screened for the duration of one year. Patients of all age groups and both sexes, referred for hemoglobinopathy screening were enrolled. Detailed clinical data were collected and venous blood samples were analyzed using HPLC. Demographic details,age-wise distribution, and prevalence of hemoglobinopathies were recorded.

Results: Of the 310 patients, 125 (40.3%) were male and 185 (59.7%) were female. The majority of patients belonged to the 21–30 years age group (33.9%). HPLC analysis showed 235 (75.8%) had normal hemoglobin while 38 (12.3%) had β-thalassemia trait, 12 (3.9%) had sickle cell trait, 4 (1.3%) had sickle cell-β-thalassemia compound heterozygosity, and 2 (0.6%) had sickle cell homozygosity. Thalassemia trait was most commonly diagnosed in the 21–30 years group. Among thalassemia carriers, RDW was predominantly in the 16–20 range.

Conclusion: This study highlights the considerable burden of hemoglobinopathies, particularly β-thalassemia trait in the regional population. HPLC proved highly effective for screening and diagnosis. Strengthening screening programs, especially among young adults along with public awareness initiatives are essential to reduce the hemoglobinopathy burden in India.

Keywords: Hemoglobinopathies, HPLC, β-thalassemia Trait, Sickle Cell Disease, Prevalence.

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Introduction

Hemoglobinopathies are among the most common monogenic disorders worldwide, characterized by structural or quantitative abnormalities in hemoglobin molecules [1]. These disorders arise due to mutations in the globin genes, leading to conditions such as thalassemias and structural hemoglobin variants like sickle cell disease, hemoglobin D, hemoglobin E, and others [2,3].

Affecting millions globally, hemoglobinopathies are especially prevalent in tropical, subtropical, and Mediterranean regions, where the carrier rates can range from 1% to 17% depending on the population studied [4,5]. India bears a significant burden of hemoglobinopathies, with an estimated 42 million carriers of β-thalassemia and approximately 150,000 homozygous patients, making it one of the countries with the highest global prevalence [6,7]. The prevalence, however, varies greatly across different geographical regions, castes, and ethnic groups, highlighting the influence of consanguineous marriages, endogamy, and migration patterns on the distribution of these disorders [8,9]. Regions such as Gujarat, West Bengal, Odisha, Maharashtra, and certain tribal populations have reported particularly high prevalence rates of hemoglobinopathies [10].

High-performance liquid chromatography (HPLC) has revolutionized the screening and diagnosis of

hemoglobinopathies over the past few decades [11]. HPLC allows for rapid, accurate, and quantitative analysis of hemoglobin fractions, enabling the detection of both common and rare hemoglobin variants [12]. It offers advantages over traditional methods like hemoglobin electrophoresis, such as improved sensitivity, reproducibility, and the ability to provide precise hemoglobin A2 and fetal hemoglobin levels, which are critical for the identification of β-thalassemia carrier counselling and compound heterozygotes [13,14].

Early and accurate diagnosis of hemoglobinopathies is crucial to guide clinical management, reduce complications, prevent unnecessary blood transfusions and offer appropriate counselling to at-risk families [15]. Population-based screening, premarital counselling and prenatal diagnosis are recognized strategies for reducing the incidence of severe hemoglobinopathies [16,17]. However, despite national programs and guidelines, the implementation of large-scale screening remains challenging in many parts of India due to limited resources, lack of awareness, and regional disparities in healthcare access [18].

Regional studies, therefore, play a pivotal role in generating baseline data on the burden and spectrum of hemoglobinopathies, which can guide policymakers and clinicians in planning effective preventive and therapeutic interventions [19]. The present study aims to estimate the prevalence and spectrum of hemoglobinopathies using HPLC in a tertiary care centre of an Indian institute, thereby contributing valuable epidemiological insights that can help improve local screening programs and clinical care pathways.

Material and Methods

Study Design and Setting: This was a cross-sectional observational study conducted at the Department of Hematology in a tertiary care centre of an Indian institute. For the duration of 12 months.

Sample Size and Population: A total of 310 consecutive patients were included in the study. These patients were either referred for evaluation of anemia, abnormal blood counts, suspected hemoglobinopathy, or were undergoing routine screening. Individuals from all age groups and both sexes were enrolled after obtaining written informed consent.

Inclusion Criteria

- Patients of all ages and genders referred for hemoglobinopathy screening
- Patients with clinical suspicion of hemoglobinopathy (e.g., microcytic hypochromic anemia, unexplained hemolysis, jaundice, or splenomegaly)
- Individuals attending premarital, prenatal, or family screening programs

Exclusion Criteria

- Patients who had received blood transfusions within the past 3 months.
- Patients with incomplete clinical or laboratory data.
- Refusal to provide consent.

Ethical Considerations: The study protocol was reviewed and approved by the Institutional Ethics Committee. Written informed consent was obtained from all participants or guardians (in case of minors) prior to enrollment. Patient confidentiality was maintained all throughout the study.

Data Collection: A detailed clinical history, including demographic data, family history, consanguinity, history of anemia, jaundice, and transfusion requirement, was recorded for each participant. A thorough physical examination was conducted to assess signs of anemia, jaundice, splenomegaly or any other relevant clinical features.

Sample Collection and Laboratory Analysis: Venous blood samples (2–3 mL) were collected in EDTA vacutainers. Complete blood count (CBC) was performed using an automated hematology analyzer. Peripheral blood smear examination was carried out to assess red cell morphology. Reticulocyte counts and sickling tests were done where indicated. For hemoglobinopathy screening, high-performance liquid chromatography (HPLC) was performed using a Bio-Rad D-10 system (or equivalent model), following the manufacturer's guidelines. The hemoglobin fractions analyzed included HbA, HbF, HbA2, and abnormal variants such as HbS, HbE, HbD, and others. Interpretation of chromatograms and quantification of hemoglobin fractions were done using the instrument's integrated software.

Statistical Analysis: The data were entered into Microsoft Excel and analyzed using SPSS version 25. Descriptive statistics were calculated for demographic variables. The prevalence of various hemoglobinopathies was expressed as percentages. Continuous variables were presented as mean ± standard deviation (SD) and categorical variables as frequencies and percentages.

Results

Table 1 shows the gender distribution of the study population, with males accounting for 40.3% and females for 59.7% of the 310 patients.

Table 2 presents the age-wise distribution, highlighting that the majority of patients belonged

to the 21–30 years group (33.9%), followed by the 31–40 years group (16.1%), and the 11–20 years group (11.3%).

Table 3 illustrates the prevalence of hemoglobinopathies detected, where 235 individuals (75.8%) had normal hemoglobin patterns, while beta thalassemia trait was the most common abnormality (12.3%), followed by sickle cell trait (3.9%), sickle cell-beta thalassemia compound heterozygosity (1.3%), and sickle cell homozygosity (0.6%). Table 4 details the age-wise detection of thalassemia carriers, showing the highest number in the 21–30 years group (39.5%), followed by 11–20 years (15.8%) and 31–40 years (13.2%), indicating the importance of screening in young adults.

Table 5 displays the RDW range among thalassemia carriers, where most had RDW values between 16–20 (22 cases), followed by 12–16 (10 cases), and a smaller subset had RDW >20 (6 cases), reflecting the variability in red cell indices among carriers.

Table 1: Gender distribution of patients

Sex	Samples (%)
Male	125 (40.3%)
Female	185 (59.7%)

Table 2: Age-wise distribution of patients

Age group (years)	Samples (%)
1 to 10	8 (2.6%)
11 to 20	35 (11.3%)
21 to 30	105 (33.9%)
31 to 40	50 (16.1%)
41 to 50	40 (12.9%)
51 to 60	30 (9.7%)
61 to 70	25 (8.1%)
71 to 80	15 (4.8%)
81 to 90	2 (0.6%)

Table 3: Prevalence of various hemoglobinopathies in study population

Hemoglobin variant	Number
Normal	235
Beta thalassemia trait	38
Sickle cell trait	12
Sickle cell-beta thalassemia compound heterozygous	4
Sickle cell homozygous	2

Table 4: Age-wise distribution of thalassemia carriers at diagnosis

Age group (years)	Thalassemia trait (%)
1 to 10	2 (5.3%)
11 to 20	6 (15.8%)
21 to 30	15 (39.5%)
31 to 40	5 (13.2%)
41 to 50	4 (10.5%)
51 to 60	4 (10.5%)
61 to 70	2 (5.3%)

Table 5: Range of red cell distribution width (RDW) among thalassemia carriers

RDW Range	Number
12 to 16	10
16 to 20	22
Above 20	6

Discussion

The present study provides valuable insights into the prevalence and distribution of hemoglobinopathies detected by high-performance liquid chromatography (HPLC) in a tertiary care setting in India. Out of 310 patients screened, 24.2% were diagnosed with some form of hemoglobinopathy, with β-thalassemia trait being the most common (12.3%), followed by sickle cell

trait (3.9%) and compound heterozygous states such as sickle cell-β-thalassemia (1.3%). These findings align with prior reports from India, where β-thalassemia trait prevalence ranges from 3–17% in various regions [4,6,9]. The high proportion of cases in the 21–30 years age group underlines the importance of targeting young adults for routine screening and premarital counseling to prevent at-risk marriages [19,20]. Earlier studies have emphasized that mass screening in schools, colleges, and antenatal clinics is crucial in controlling the disease burden in high-prevalence areas [21]. The higher prevalence among females (59.7%) in our cohort likely reflects gender patterns in healthcare-seeking behavior, particularly in reproductive-age women undergoing antenatal screening [22].

HPLC has emerged as the diagnostic modality of choice due to its ability to detect both qualitative and quantitative hemoglobin disorders with high precision and reproducibility [11,12,23]. It offers distinct advantages over traditional electrophoresis, such as automated analysis, minimal hands-on time, and the ability to detect multiple abnormal hemoglobin variants in a single run [14].

This explains why it is now widely used for carrier detection, premarital counseling, and neonatal screening programs across India [18]. Interestingly, the RDW (red cell distribution width) analysis among thalassemia carriers in our study showed that the majority fell within the 16–20 range, consistent with the microcytosis and anisocytosis seen in thalassemia traits [5,24].

RDW can serve as a useful adjunct marker in screening programs, particularly in resource-limited settings where advanced testing may not be readily available. Several population-based studies have emphasized the importance of regional epidemiological data to design locally relevant control strategies [8,10,25]. Given the heterogeneity of hemoglobinopathies across India's diverse population, localized data as generated in this study are critical to informing public health programs.

Moreover, early detection enables timely genetic counseling and prevention of births with severe hemoglobinopathies, which carry significant emotional, financial, and health burdens.

Despite national thalassemia control programs, there remain challenges such as lack of awareness, limited access to screening in rural areas, and inadequate counseling services [17,19]. Our findings reinforce the need to strengthen these programs and expand them to reach a wider population base, especially targeting high-risk communities.

Conclusion

The present study highlights the significant burden of hemoglobinopathies, particularly β-thalassemia trait, in the regional population attending a tertiary care center. HPLC proved to be an invaluable tool for the detection and classification of hemoglobin variants. The findings underscore the necessity for routine population-based screening, especially among young adults, to enable early diagnosis, counseling, and prevention strategies. Strengthening public health interventions and awareness campaigns will be essential to reduce the incidence and burden of hemoglobinopathies in India.

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