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Study of Clinico-Hematological Profile in Various Hemoglobinopathies at a Tertiary Care Centre in Central India

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Abstract: ***Background: Hemoglobinopathies are among the most common inherited hematological disorders worldwide and constitute a major public health problem in India. Their prevalence and clinical manifestations vary considerably across different geographical regions and ethnic populations. Aim: To evaluate the clinico-hematological profile of patients with various hemoglobinopathies and to assess their prevalence at a tertiary care centre in Central India. Materials and Methods: This observational, descriptive cross-sectional study included 100 patients with suspected or confirmed hemoglobinopathies attending a tertiary care centre in Central India over an 18-month period. Detailed demographic, clinical, and hematological data were collected. Complete blood counts, peripheral smear examination, and High-Performance Liquid Chromatography (HPLC) were performed for diagnosis and characterization of hemoglobinopathies. Results: Among the 100 patients studied, 54% were females and 46% were males. Sickle Cell Disease was the most common hemoglobinopathy, accounting for 69% of cases, followed by Sickle Cell Trait (10%), Thalassemia Trait (6%), HbS + Beta Thalassemia (6%), and Beta Thalassemia Minor (5%). The mean hemoglobin concentration was 8.59 g/dL, and the mean RBC count was 3.40 million/µL. HPLC analysis showed mean HbS, HbF, and HbA fractions of 56.14%, 13.68%, and 18.72%, respectively. Clinically, jaundice (70%), blood transfusion history (65%), joint pain (60%), and previous hospitalization (55%) were the most common findings. Conclusion: Sickle cell disorders constitute the predominant hemoglobinopathies in Central India and are associated with significant clinical morbidity and hematological abnormalities. HPLC remains an invaluable tool for accurate diagnosis and classification. Early detection, genetic counseling, and comprehensive management strategies are essential to reduce disease burden and improve patient outcomes.***

Keywords: Hemoglobinopathies, Sickle Cell Disease, Thalassemia, HPLC, Hematological Profile, HbS, Central India, Anemia.

1. Introduction

Hemoglobinopathies constitute a heterogeneous group of inherited disorders characterized by abnormalities in the structure, synthesis, or function of hemoglobin. These disorders are among the most common monogenic diseases worldwide and represent a significant public health burden, particularly in developing countries where carrier frequencies are high and healthcare resources are often limited [1]. Hemoglobinopathies broadly include thalassemia syndromes and structural hemoglobin variants such as sickle cell disease, hemoglobin E disease, hemoglobin D disease, and their compound heterozygous forms [2].

The global prevalence of hemoglobinopathies has increased considerably due to population growth, migration, and improved survival of affected individuals. It is estimated that nearly 7% of the world's population carries a gene for a hemoglobin disorder, and over 300,000 affected infants are born annually [3]. India bears a substantial share of this burden, with a wide geographical variation in the distribution of hemoglobinopathies. The prevalence of β-thalassemia trait in India ranges from 3% to 17%, while sickle cell disorders are highly prevalent among tribal populations and certain communities of Central, Western, and Eastern India [4,5].

Hemoglobinopathies manifest with diverse clinical presentations ranging from asymptomatic carrier states to severe transfusion-dependent anemia and multisystem complications. Patients may present with pallor, jaundice, hepatosplenomegaly, growth retardation, recurrent infections, bone deformities, vaso-occlusive crises, and organ dysfunction depending upon the type and severity of the underlying disorder [6]. The phenotypic expression is influenced by several factors, including genetic modifiers, co-inheritance of other hemoglobin variants, environmental factors, and access to healthcare facilities [7].

Hematological evaluation plays a crucial role in the diagnosis and characterization of hemoglobinopathies. Complete blood count parameters, peripheral blood smear examination, red cell indices, reticulocyte counts, and hemoglobin fraction analysis by high-performance liquid chromatography (HPLC) or electrophoresis provide valuable information for identifying different hemoglobin disorders [8]. Early and accurate diagnosis is essential for initiating appropriate management strategies, genetic counseling, prenatal diagnosis, and prevention programs aimed at reducing disease burden [9].

Central India represents one of the regions with a high prevalence of various hemoglobinopathies due to its unique ethnic composition and genetic diversity. Despite the significant burden of these disorders, comprehensive data regarding their clinico-hematological characteristics remain limited in many tertiary care settings. Understanding the spectrum of clinical manifestations and associated hematological profiles is important for improving diagnostic accuracy, optimizing patient management, and formulating effective public health interventions [10].

The present study was undertaken to evaluate the clinico-hematological profile of patients with various hemoglobinopathies and to assess the prevalence of these disorders in Central India. It also aimed to analyze the spectrum of hematological parameters and clinical manifestations associated with different hemoglobinopathies, thereby identifying regional variations in disease presentation. Through a comprehensive assessment of clinical features and laboratory findings, the study sought to enhance

the understanding of the burden, pattern, and diversity of hemoglobinopathies in the population of Central India.

2. Materials and Methods

Study Design: Observational, descriptive cross-sectional study.

Study Population: Patients of all age groups and genders presenting to the outpatient or inpatient departments with suspected or confirmed hemoglobinopathies and undergoing diagnostic evaluation at a tertiary care center in Central India.

Sample Size: A total of 100 patients with suspected or confirmed hemoglobinopathies were included in the study.

Study Duration: 18 months (Academic Year 2022–2025).

Study Place: Tertiary Care Center, Central India.

Inclusion Criteria:

- Patients of all age groups and both genders presenting with symptoms suggestive of anemia or hemoglobinopathy, or referred for routine screening.
- Patients with clinical suspicion or family history of hemoglobinopathy.
- Subjects willing to participate and provide informed consent for investigations.

Exclusion Criteria:

- Patients with incomplete clinical or laboratory records.
- Patients with isolated iron deficiency anemia without evidence of hemoglobinopathy.
- Patients with chronic liver disease, renal disease, or other conditions significantly affecting hematological parameters.
- Patients with a history of blood transfusion within the preceding 3 months.

Statistical Analysis: We put the data into Microsoft Excel and then used SPSS software version 27.0 (SPSS Inc., Chicago, IL, USA) and GraphPad Prism version 5 to look at it. Mean ± standard deviation was used to show continuous variables, and frequencies and percentages were used to show categorical variables. The unpaired t-test was utilized to examine continuous variables between independent groups, whereas the paired t-test was employed for comparisons within the same group. The Chi-square test or Fisher's exact test was used to look at categorical variables, depending on which one was better. A p-value of less than 0.05 was seen to be statistically important.

3. Result

Table 1: Demographic Characteristics of the Study Population (n = 100)

Parameter	Male (n=46)	Female (n=54)
Number of Patients	46 (46%)	54 (54%)
Mean Age (Years)	23.91 ± 13.72	26.31 ± 11.33

Table 2: Types and Prevalence of Hemoglobinopathies Identified (n = 100)

Hemoglobinopathy	Number of Cases	Percentage (%)
Sickle Cell Disease (SS Pattern)	69	69
Sickle Cell Trait (AS Pattern)	10	10
Beta Thalassemia Minor	5	5
Thalassemia Trait (including Delta-Beta Thalassemia)	6	6
HbD Punjab (Heterozygous)	2	2
HbS + Beta Thalassemia (Double Heterozygous)	6	6
HbS + HbD (Double Heterozygous)	2	2
HbE + Beta Thalassemia	1	1
Sickle Cell Trait + Alpha Thalassemia	1	1

Table 3: Hematological Profile of the Study Population

Parameter	Mean Value
Hemoglobin (g/dL)	8.59
RBC Count (million/µL)	3.4
MCV (fL)	83.11
MCH (pg)	27.54
MCHC (g/dL)	32.13
RDW (%)	20.89
Mentzer Index	30.42
Total Leukocyte Count (×10³/µL)	7.94
Granulocyte Percentage (%)	49.37
Platelet Count (/µL)	2,80,000

Table 4: Average Hemoglobin Fractions on HPLC Analysis

Hemoglobin Fraction	Mean Percentage (%)
HbA	18.72
HbA_2	3.42
HbF	13.68
HbS	56.14
HbD	1.87

Table 5: Clinical Profile of Patients with Hemoglobinopathies

Clinical Feature	Number of Patients (Approx.)	Percentage (%)
History of Jaundice	70	70
History of Joint Pain	60	60
Previous Blood Transfusion	65	65
Previous Hospital Admission	55	55

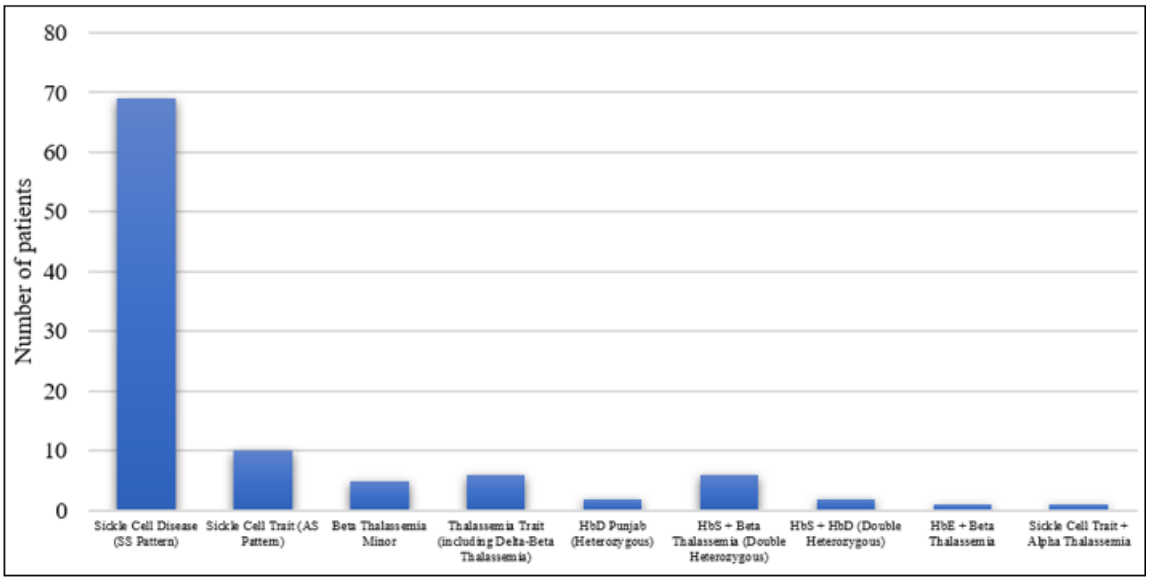

Figure 1: Types and Prevalence of Hemoglobinopathies Identified

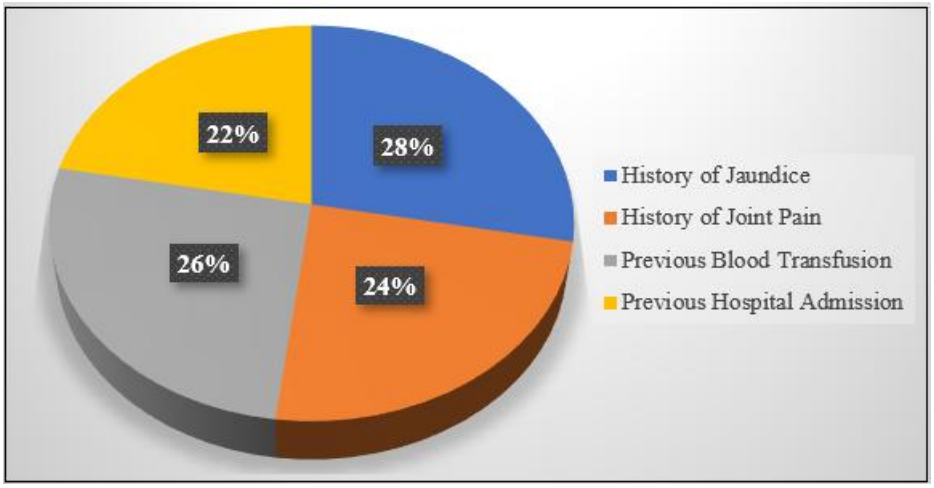

Figure 2: Clinical Profile of Patients with Hemoglobinopathies

A total of 100 patients diagnosed with various hemoglobinopathies were included in the study. Among them, 54 (54%) were females and 46 (46%) were males, indicating a slight female predominance. The mean age of female patients was 26.31 ± 11.33 years, whereas the mean age of male patients was 23.91 ± 13.72 years. The findings suggest that hemoglobinopathies were predominantly observed among young adults, with females constituting a marginally larger proportion of the study population. This age distribution reflects the chronic nature of these inherited disorders, which are frequently diagnosed during childhood or adolescence and continue to require medical attention into adulthood.

Sickle Cell Disease (SS pattern) was the most prevalent hemoglobinopathy identified in the study, accounting for 69 cases (69%). Sickle Cell Trait (AS pattern) was the second most common disorder, observed in 10 patients (10%). Thalassemia-related disorders collectively constituted a significant proportion of cases, including Beta Thalassemia Minor in 5 patients (5%) and Thalassemia Trait, including delta-beta thalassemia, in 6 patients (6%). Double heterozygous states were also encountered, with HbS + Beta Thalassemia present in 6 cases (6%) and HbS + HbD identified in 2 cases (2%). Rare variants included HbD Punjab heterozygosity in 2 patients (2%), HbE + Beta Thalassemia in 1 patient (1%), and Sickle Cell Trait associated with Alpha Thalassemia in 1 patient (1%). These findings demonstrate that sickle cell disorders constituted the overwhelming majority of hemoglobinopathies in the study population, highlighting their substantial burden in Central India.

The hematological analysis revealed significant abnormalities consistent with chronic hemolytic anemia and inherited hemoglobin disorders. The overall mean hemoglobin concentration was 8.59 g/dL, indicating moderate anemia among the study participants. The mean RBC count was 3.40 million/µL, reflecting reduced erythrocyte levels commonly associated with hemoglobinopathies. The mean MCV was 83.11 fL, while the mean MCH and MCHC were 27.54 pg and 32.13 g/dL, respectively.

The mean RDW was markedly elevated at 20.89%, suggesting significant anisocytosis and variability in red blood cell size. The Mentzer Index was 30.42, while the mean total leukocyte count was 7.94 $\times 10^3$/µL and the mean granulocyte percentage was 49.37%. The average platelet count was 280,000/µL, which remained within normal physiological limits.

Disease-specific analysis demonstrated that patients with Sickle Cell Disease had a mean hemoglobin level of approximately 8.2 g/dL, reflecting chronic hemolytic anemia. In contrast, patients with Beta Thalassemia Minor exhibited relatively higher hemoglobin levels ranging between 10 and 12 g/dL. Individuals with HbD and HbE variants showed variable hemoglobin values depending on the associated genetic abnormalities and disease severity.

High-Performance Liquid Chromatography (HPLC) analysis revealed distinct hemoglobin fraction patterns characteristic of the various hemoglobinopathies included in the study. The mean HbA fraction was 18.72%, while the mean HbA_2 level was 3.42%. The mean HbF concentration was elevated at 13.68%, reflecting increased fetal hemoglobin production in many patients, particularly those with sickle cell disease and thalassemia syndromes.

The mean HbS fraction was 56.14%, representing the predominance of sickle hemoglobin among the study population and correlating with the high prevalence of Sickle Cell Disease. The mean HbD fraction was 1.87%, reflecting the relatively lower frequency of HbD Punjab and related compound heterozygous states. These HPLC findings played a crucial role in confirming the diagnosis and classification of the various hemoglobinopathies encountered during the study.

Clinical evaluation revealed a substantial burden of disease-related symptoms and complications among the study participants. A history of jaundice was reported by approximately 70 patients (70%), making it the most common clinical manifestation observed. This finding reflects the chronic hemolysis associated with many hemoglobinopathies, particularly sickle cell disease.

Joint pain was reported by approximately 60 patients (60%), largely attributable to vaso-occlusive episodes and musculoskeletal complications seen in sickle cell disorders. A history of blood transfusion was present in approximately 65 patients (65%), indicating the significant transfusion requirements associated with severe forms of hemoglobinopathy. Furthermore, around 55 patients (55%) had a history of hospital admission due to disease-related complications, highlighting the considerable morbidity and healthcare burden imposed by these disorders.

Overall, the clinical profile demonstrated that hemoglobinopathies in this population were associated with frequent hemolytic manifestations, recurrent painful episodes, substantial transfusion dependency, and repeated hospitalizations, particularly among patients with sickle cell disease and compound heterozygous hemoglobin variants.

4. Discussion

In the present study, females constituted a slightly higher proportion of the study population (54%) compared to males (46%), with a mean age of 26.31 ± 11.33 years among females and 23.91 ± 13.72 years among males. The majority of patients belonged to the young adult age group, reflecting the chronic nature of hemoglobinopathies and their continued clinical impact beyond childhood. Similar observations were reported by Atla et al., who found a female predominance and a higher concentration of cases in the second and third decades of life among patients undergoing hemoglobinopathy screening in South India [11]. Nagose et al. also reported that most patients with hemoglobinopathies were young individuals between 10 and 35 years of age, emphasizing that these inherited disorders commonly become clinically evident during adolescence and early adulthood [12]. In contrast, Balgir observed a predominantly pediatric population among tribal communities in Odisha, which may be attributable to earlier disease detection through community-based screening programs and the higher burden of severe hemoglobinopathies in children within tribal populations [13]. The demographic findings of the present study therefore suggest that hemoglobinopathies continue to represent a significant healthcare concern among young adults in Central India, necessitating long-term monitoring and management.

The most striking finding of the present study was the predominance of Sickle Cell Disease (69%), followed by Sickle Cell Trait (10%), while thalassemia-related disorders and compound heterozygous states accounted for a smaller proportion of cases. This distribution reflects the unique epidemiological profile of Central India, where the sickle cell gene is highly prevalent among various tribal and non-tribal populations. Similar findings were reported by Patel et al., who documented a high prevalence of sickle cell disease among patients attending tertiary healthcare facilities in Central India, with sickle cell syndromes accounting for the majority of hemoglobinopathy cases [14]. In contrast, Goswami et al. from West Bengal observed a predominance of sickle cell trait rather than sickle cell disease, suggesting regional variations in gene frequency and referral patterns [15]. Studies from Eastern India have demonstrated a higher prevalence of HbE-related disorders and β-thalassemia syndromes compared to sickle cell disease [16]. The identification of rare variants such as HbD Punjab, HbS-HbD double heterozygosity, HbE-β thalassemia, and sickle cell trait with alpha-thalassemia in the present study highlights the genetic heterogeneity of hemoglobinopathies in India. Such diversity underscores the importance of comprehensive diagnostic evaluation, including HPLC analysis, for accurate characterization of these disorders.

The mean hemoglobin concentration in the present study was 8.59 g/dL, indicating moderate anemia among the affected patients. The reduced RBC count and elevated RDW further reflected chronic hemolysis and ineffective erythropoiesis associated with various hemoglobinopathies. Similar hematological findings were reported by Chaturvedi et al., who observed mean hemoglobin levels ranging from 7.8 to 9.1 g/dL among patients with sickle cell disease and thalassemia syndromes [17]. Nagose et al. also reported low hemoglobin values accompanied by elevated RDW and microcytosis among patients with hemoglobinopathies, particularly those with coexisting thalassemia [12]. The mean MCV of 83.11 fL in the present study was consistent with the coexistence of normocytic sickle cell disease and microcytic thalassemia syndromes. Elevated RDW (20.89%) indicated significant anisocytosis, a feature commonly associated with chronic hemolytic states and mixed red cell populations. The disease-specific observation that β-thalassemia minor patients

maintained relatively higher hemoglobin levels compared to sickle cell disease patients is consistent with the generally milder clinical course of heterozygous thalassemia syndromes [18]. These findings collectively support the utility of routine hematological indices as valuable screening tools in the evaluation of suspected hemoglobinopathies.

HPLC analysis demonstrated a mean HbS fraction of 56.14%, elevated HbF levels (13.68%), and a relatively low HbA fraction (18.72%), findings that correlate with the high prevalence of sickle cell disease in the study population. Elevated HbF levels are known to exert a protective effect by reducing sickling episodes and disease severity. Similar HPLC patterns have been reported by Pant et al., who demonstrated that patients with sickle cell disease exhibit markedly elevated HbS fractions accompanied by variable increases in HbF levels [19]. Chaturvedi et al. also documented increased HbA_2 levels in β-thalassemia carriers and characteristic mixed chromatographic peaks in compound heterozygous conditions [17]. The mean HbA_2 value of 3.42% observed in the present study was consistent with the presence of thalassemia trait cases. The identification of HbD Punjab and compound heterozygous variants further emphasizes the diagnostic accuracy of HPLC in differentiating complex hemoglobinopathies. These findings reinforce the role of HPLC as the gold standard screening and confirmatory technique for hemoglobinopathy diagnosis in resource-limited settings.

The clinical profile of the study population revealed that jaundice (70%), blood transfusion history (65%), joint pain (60%), and hospitalization (55%) were common manifestations. The high prevalence of jaundice reflects chronic hemolysis, particularly among patients with sickle cell disease and compound heterozygous states. Comparable findings were reported by Mondal et al., who observed jaundice in approximately two-thirds of patients with clinically significant hemoglobinopathies [20]. Joint pain was reported by 60% of patients in the present study and is consistent with the vaso-occlusive phenomena characteristic of sickle cell disease. Similar frequencies have been documented by Goswami et al., who reported musculoskeletal pain as one of the most frequent clinical presentations among sickle cell patients [15]. The substantial proportion of patients requiring blood transfusions and hospital admissions indicates the considerable disease burden and healthcare utilization associated with hemoglobinopathies. Frequent transfusion requirements are generally linked to severe anemia, recurrent crises, and complications such as acute chest syndrome and splenic sequestration. The present findings therefore highlight the significant morbidity associated with hemoglobinopathies in Central India and underscore the need for early diagnosis, genetic counseling, comprehensive disease management, and strengthened preventive strategies.

5. Conclusion

The present study highlights the significant burden of hemoglobinopathies in Central India, with Sickle Cell Disease emerging as the most prevalent disorder, followed by Sickle Cell Trait and various thalassemia syndromes. The study population was predominantly composed of young adults, with a slight female predominance. Hematological evaluation revealed moderate anemia, reduced red blood cell counts, elevated red cell distribution width, and characteristic alterations in red cell indices. HPLC proved to be an effective diagnostic tool for the identification and differentiation of various hemoglobinopathies, including rare and compound heterozygous variants. Clinically, jaundice, joint pain, blood transfusion dependency, and recurrent hospitalizations were common manifestations, reflecting the substantial morbidity associated with these disorders. The findings emphasize the need for early diagnosis, routine screening programs, genetic counseling, and comprehensive disease management strategies. Strengthening awareness and preventive measures at the community level can contribute significantly to reducing the disease burden and improving the quality of life of affected individuals in Central India.

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