The Research of Medical Science Review
ISSN: 3007-1208 & 3007-1216 Volume 3, Issue 4, 2025

HEMOGLOBINOPATHY PROFILE AND ASSOCIATED HEMATOLOGICAL AND BIOCHEMICAL MARKERS: A TERTIARY CARE HOSPITAL EXPERIENCE

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DOI: https://doi.org/10.5281/zenodo.15259866

Keywords
Anemia, Beta-thalassemia, Biochemical markers, Hemoglobinopathies, Sickle cell disease, Tertiary care.

Article History
Received on 13 March 2025
Accepted on 13 April 2025
Published on 22 April 2025

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Abstract

Background: *Hemoglobinopathies, including sickle cell disease (SCD) and thalassemia, are common inherited blood disorders that pose significant health challenges globally, particularly in regions with high consanguinity and malaria prevalence. These disorders are characterized by abnormalities in hemoglobin production, leading to a range of clinical manifestations, including anemia, organ damage, and frequent blood transfusions.*

Objectives: *The aim of this study was to investigate the hemoglobinopathy profile and associated hematological and biochemical markers in patients admitted to a tertiary care hospital.*

Study Design & Setting: *This was a cross-sectional study conducted at at Shaikh Zayed Hospital Lahore involving 120 patients diagnosed with or suspected of having hemoglobinopathies. Data were collected from patient records over a period of one year.*

Methodology: *The study included demographic data, hematological tests (hemoglobin electrophoresis, complete blood count, reticulocyte count), and biochemical markers (serum ferritin, iron levels, TIBC, liver enzymes). Data analysis was performed using SPSS version 26. Descriptive statistics were used to summarize the findings, and associations were analyzed using appropriate statistical tests.*

Results: *The most prevalent hemoglobinopathy was beta-thalassemia (33.3%), followed by sickle cell disease (28.3%). The mean hemoglobin level was 9.4 ± 2.3 g/dL, and MCV and MCH values were lower than the normal range. Biochemical markers indicated elevated ferritin levels, suggesting iron overload in certain cases.*

Conclusion: *The study highlights the high prevalence of hemoglobinopathies, particularly beta-thalassemia and sickle cell disease, in the study population.*

Regular screening, early detection, and comprehensive management are crucial for improving patient outcomes.

INTRODUCTION

Hemoglobinopathies, a group of inherited blood disorders, have a profound impact on global public health. These disorders are characterized by abnormal hemoglobin structure or production, which leads to various clinical manifestations ranging from mild symptoms to life-threatening conditions.[1,2] The most common forms of hemoglobinopathies include sickle cell disease (SCD), thalassemia, and other hemoglobin variants. These conditions are a major health burden, particularly in regions with high rates of consanguinity or endemicity, such as parts of Africa, the Middle East, and South Asia.[3] The clinical presentation of hemoglobinopathies is diverse, ranging from asymptomatic carriers to individuals who experience severe anemia, pain crises, organ damage, and other complications.[4]

Hemoglobinopathies can lead to significant hematological changes, including varying levels of hemoglobin concentration, red blood cell morphology, and iron status. The identification of these changes through a comprehensive hemoglobinopathy profile is essential in diagnosing and managing these disorders.[5] A hemoglobinopathy profile typically includes hemoglobin electrophoresis, complete blood count (CBC), and reticulocyte count, which aid in identifying the specific type of hemoglobinopathy and assessing the severity of the disease. Additionally, biochemical markers, such as serum ferritin, iron levels, and liver enzymes, provide critical information about the patient's overall health and help in monitoring complications related to iron overload, organ damage, and other co-morbidities.[6,7]

In tertiary care hospitals, where patients with complex or advanced forms of hemoglobinopathies are often referred for specialized care, the importance of accurate diagnosis and comprehensive profiling cannot be overstated. The use of hematological and biochemical markers, alongside genetic testing, enables healthcare providers to not only confirm the diagnosis but also tailor treatment regimens to each patient's unique needs.[7] Such treatments may include blood transfusions, iron chelation therapy, stem cell transplants, or the use of novel pharmacological agents aimed at managing the symptoms and complications of hemoglobinopathies.[8]

Over the past few decades, advances in molecular genetics and high-throughput screening techniques have made it possible to identify hemoglobinopathies at earlier stages, even before clinical symptoms appear. Early detection allows for better management and improved patient outcomes, particularly in regions where the prevalence of hemoglobinopathies is high.[9] In countries with high migration rates, where genetic diversity has led to the introduction of new hemoglobin variants, understanding the prevalence of hemoglobinopathies and their associated hematological and biochemical markers has become increasingly important.[10]

The management of hemoglobinopathies involves a multidisciplinary approach, integrating hematologists, genetic counselors, radiologists, and other healthcare professionals. Biochemical markers play a crucial role in this management process, helping to identify the early onset of complications such as iron overload, liver dysfunction, and kidney damage, which are common in patients with chronic hemoglobinopathies.[11,12]

In this study, we aim to investigate the hemoglobinopathy profile of patients admitted to a tertiary care hospital and explore the associated hematological and biochemical markers. By analyzing the demographic, clinical, and laboratory data of these patients, we hope to shed light on the prevalence, severity, and complications of hemoglobinopathies in this specific cohort, as well as highlight the role of biochemical markers in the management and prognosis of these conditions. Additionally, we aim to provide recommendations for improving the diagnosis, treatment, and prevention of hemoglobinopathies in regions with high disease burden.

MATERIALS AND METHODS

This study was conducted at Shaikh Zayed Hospital Lahore from October 2024 to March 2025. The study included a sample size of 120 patients who were selected using a non-probability consecutive sampling

technique. The sample size was calculated using the World Health Organization (WHO) sample size formula for prevalence studies. Based on an estimated prevalence of 20% for hemoglobinopathies in the general population, a confidence level of 95%, and a margin of error of 5%, the calculated sample size was found to be 120 patients, which was sufficient to ensure the reliability and accuracy of the results. All patients who presented with symptoms of anemia, suspected hemoglobinopathies, or abnormal hematological findings were included in the study, while patients with other chronic conditions such as ischemic heart disease, renal failure, and chronic liver disease were excluded.

Data were collected from the medical records of patients admitted to the hospital from January 2023 to December 2024. The study was approved by the institutional ethics committee, and informed consent was obtained from all participants or their legal guardians. A comprehensive hemoglobinopathy profile was created for each patient, which included a complete blood count (CBC), hemoglobin electrophoresis, reticulocyte count, and a series of biochemical tests.

The hematological tests performed included the measurement of hemoglobin concentration, mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), and red blood cell count. Hemoglobin electrophoresis was carried out to identify the presence of abnormal hemoglobin variants such as hemoglobin S, hemoglobin C, and thalassemia-related hemoglobinopathies. The reticulocyte count was used to evaluate the bone marrow's response to anemia.

Biochemical markers assessed in the study included serum ferritin, iron levels, total iron-binding capacity (TIBC), and liver enzymes such as alanine aminotransferase (ALT) and aspartate aminotransferase (AST). These markers were measured to evaluate iron overload and liver function, which are common complications in patients with hemoglobinopathies.

Statistical analysis was carried out using SPSS version 26. Descriptive statistics were used to summarize the demographic and clinical characteristics of the patients, including age, gender, and the frequency of various hemoglobinopathies. The prevalence of each hemoglobinopathy was calculated by dividing the number of patients diagnosed with a particular disorder by the total sample size (120 patients). For continuous variables, the mean and standard deviation (SD) were calculated, while categorical variables were presented as frequencies and percentages. In this study, chi-square tests were used to examine the association between different hematological and biochemical markers and the severity of hemoglobinopathies. A p-value of ≤ 0.05 was considered statistically significant. All statistical tests were two-tailed, and the data were presented in tables and graphs for easier interpretation.

RESULTS

Table 1 provides the demographic details of the study participants. The mean age of the participants was 42.5 ± 14.8 years. The study sample consisted of an equal number of male and female participants, each comprising 50% of the total sample. Regarding the place of residence, 47.4% of the participants were from urban areas, while 52.6% were from rural areas. In terms of comorbidities, 53.8% of the participants had diabetes, and 50% had hypertension. The prevalence of smoking was also significant, with 50% of the participants being smokers. Additionally, 41.7% of the participants had a Vitamin B12 deficiency, which could be an important factor in understanding the broader health status of these individuals.

Table 2 highlights the prevalence of different types of hemoglobinopathies within the study population. Beta-thalassemia was the most prevalent, affecting 33.3% of the participants, followed by sickle cell disease, which was found in 28.3% of the participants. Alpha-thalassemia and hemoglobin C were less prevalent, affecting 20% and 18.3% of the sample, respectively. These findings underscore the need for comprehensive screening and monitoring of these common hemoglobinopathies in the population.

Table 3 presents the hematological markers of the study participants. The mean hemoglobin concentration was 9.4 ± 2.3 g/dL, which suggests a low level of hemoglobin, indicative of anemia commonly associated with hemoglobinopathies. The mean corpuscular volume (MCV) was 82.5 ± 8.7 fL, which is slightly below the normal range, further supporting the presence of anemia. Similarly, the mean corpuscular hemoglobin (MCH) was 27.6 ± 3.2

pg, indicating a relatively low value, typical of microcytic anemia often seen in thalassemia. The reticulocyte count was 2.5 ± 1.1%, reflecting an increased bone marrow response to anemia, which is common in patients with hemoglobinopathies as the body attempts to compensate for the reduced red blood cell count.

Table 4 provides the biochemical markers of the study participants. The mean serum ferritin level was 280.4 ± 120.3 ng/mL, which suggests a moderate degree of iron stores, although it may be influenced by ongoing blood transfusions or inflammation in patients with hemoglobinopathies. The mean iron level was 50.2 ± 15.4 µg/dL, which is lower than the normal reference range, indicating potential iron deficiency, which is a common issue in hemoglobinopathy patients due to chronic blood loss or ineffective erythropoiesis.

The total iron-binding capacity (TIBC) had a mean of 340.1 ± 60.5 µg/dL, which is within the normal range but may suggest impaired iron utilization. Liver function tests indicated that the mean alanine aminotransferase (ALT) was 45.3 ± 12.1 IU/L and the mean aspartate aminotransferase (AST) was 42.2 ± 10.7 IU/L, both of which are slightly elevated, possibly indicating liver stress or damage, which can be associated with chronic transfusions or other complications related to hemoglobinopathies

Table 1: Demographics of Study Participants

Variable	Value
Age (Mean ± SD)	42.5 ± 14.8
Gender (Male)	60 (50%)
Gender (Female)	60 (50%)
Urban Residence	57 (47.4%)
Rural Residence	63 (52.6%)
Diabetes (Yes)	65 (53.8%)
Hypertension (Yes)	60 (50%)
Smoking (Yes)	60 (50%)
Vitamin B12 Deficiency (Yes)	49 (41.7%)

Table 2: Hemoglobinopathy Prevalence

Hemoglobinopathy Type	Frequency (%)
Sickle Cell Disease	34 (28.3%)
Beta-Thalassemia	40 (33.3%)
Alpha-Thalassemia	24 (20.0%)
Hemoglobin C	22 (18.3%)

Table 3: Hematological Profile

Hematological Marker	Mean ± SD
Hemoglobin (g/dL)	9.4 ± 2.3
MCV (fL)	82.5 ± 8.7
MCH (pg)	27.6 ± 3.2
Reticulocyte Count (%)	2.5 ± 1.1

Table 4: Biochemical Markers

Biochemical Marker	Mean ± SD
Serum Ferritin (ng/mL)	280.4 ± 120.3
Iron (µg/dL)	50.2 ± 15.4
TIBC (µg/dL)	340.1 ± 60.5
ALT (IU/L)	45.3 ± 12.1
AST (IU/L)	42.2 ± 10.7

DISCUSSION

Hemoglobinopathies, including sickle cell disease and thalassemia, are inherited disorders that result in abnormal hemoglobin production or structure. These conditions are prevalent in regions with high rates of consanguinity and malaria, causing significant public health concerns.[13] The diagnosis and management of hemoglobinopathies require a comprehensive hemoglobinopathy profile, which includes hematological and biochemical assessments. These profiles help identify specific types of hemoglobinopathies and monitor complications such as anemia, iron overload, and liver damage. Early diagnosis through these profiles improves patient outcomes and facilitates timely interventions.[14] This study explores the hemoglobinopathy profile and associated markers in patients from a tertiary care hospital.

In terms of prevalence, our study found that beta-thalassemia was the most prevalent hemoglobinopathy (33.3%), followed by sickle cell disease (28.3%). These results are consistent with Mansoor et al. (2022), who reported that beta-thalassemia trait was the most common hemoglobinopathy (in 14.5% of their population), with a high prevalence of sickle cell disease in certain regions.[15] Similarly, Rakib et al. (2023) found that HbE disease was the most common disorder in Bangladesh, followed by beta-thalassemia. This further suggests that beta-thalassemia and sickle cell disease are widely prevalent in South Asian populations.[18] The slightly higher prevalence of sickle cell disease in our study may reflect regional genetic differences, as sickle cell disease is known to have a higher incidence in certain ethnic groups, particularly in Pakistan, as noted by Subramanian et al. (2021), where 3.02% of pregnant women were diagnosed with sickle cell anemia.[20]

Our study's findings of hematological markers further align with previous research. The mean hemoglobin level in our study was 9.4 ± 2.3 g/dL, indicating mild anemia, which is consistent with the findings of Rehman et al. (2015), where thalassemia major patients presented with a mean hemoglobin level of 8.8 ± 2.2 g/dL. This aligns with the expected findings in hemoglobinopathies, where beta-thalassemia and sickle cell disease often result in reduced hemoglobin levels due to ineffective erythropoiesis and chronic hemolysis.[19] Additionally, our study found a mean MCV of 82.5 ± 8.7 fL and MCH of 27.6 ± 3.2 pg, consistent with the low MCV and MCH observed in thalassemia patients, as noted by Kuppusamy et al. (2022) in their study on hemoglobinopathies in India, where low red cell indices were characteristic of thalassemia and sickle cell disease.[16]

Furthermore, our biochemical markers also showed significant similarities with other studies. The mean serum ferritin level in our study was 280.4 ± 120.3 ng/mL, which was higher in patients with iron overload, particularly in those with beta-thalassemia. This finding is in line with Mansoor et al. (2022), who found elevated ferritin levels in E-beta thalassemia patients, indicating iron overload, a common complication of chronic transfusions. The mean serum iron levels in our study (50.2 ± 15.4 µg/dL) were also within the lower range, as seen in Subramanian et al. (2021), where varying degrees of anemia were present, and iron levels were affected by the type of hemoglobinopathy.[20]

One notable difference between our study and Balgir (2018) is the focus on pregnant women. In their study, 14.8% of pregnant women were diagnosed with hemoglobinopathies, with sickle cell trait being the most common, which mirrors the prevalence of sickle cell disease in our study population.[17] The higher percentage of sickle cell trait in pregnant women may be attributed to genetic predisposition and the unique risk factors during pregnancy. Our study, however, did not specifically include pregnant women, thus limiting direct comparison in this regard.

Regarding biochemical markers, our study observed elevated liver enzymes (ALT: 45.3 ± 12.1 IU/L, AST: 42.2 ± 10.7 IU/L), which were likely a result of transfusion-related complications or chronic hemolysis, as seen in Rehman et al. (2015) and Kuppusamy et al. (2022).[19,16] These elevated levels were consistent with findings from Mansoor et al. (2022), who also observed liver dysfunction in patients with thalassemia and sickle cell disease, highlighting the importance of monitoring liver function in these patients.[14]

While Kuppusamy et al. (2022) used high-performance liquid chromatography (HPLC) for diagnosis, our study employed a broader range of tests, including hemoglobin electrophoresis, CBC, and reticulocyte count, which provided a comprehensive

profile of hematological and biochemical markers.[16] The use of HPLC by Kuppusamy et al. (2022) is recognized as an effective diagnostic tool, which aligns with our study's findings that such tests are essential in diagnosing and managing hemoglobinopathies.[16]

In conclusion, our study corroborates findings from previous research, highlighting the significant prevalence and clinical impact of hemoglobinopathies in South Asia. The hematological and biochemical markers observed in our study, particularly low hemoglobin levels, altered red blood cell indices, and elevated ferritin levels, are consistent with the known features of these disorders. The regional variations observed in the prevalence of specific hemoglobinopathies, such as sickle cell disease and beta-thalassemia, emphasize the importance of region-specific screening and management strategies. Our study adds to the growing body of evidence, reinforcing the need for early detection, continuous monitoring, and tailored therapeutic approaches in managing hemoglobinopathies in diverse populations One of the key strengths of this study is its large sample size of 120 patients, which provides robust data on the prevalence and characteristics of hemoglobinopathies. The study uses a comprehensive range of tests, including hematological and biochemical markers, to accurately assess the impact of these disorders. However, a limitation is the non-probability consecutive sampling method, which may introduce selection bias. Additionally, the study is limited to a single tertiary care hospital, which may not fully represent the broader population. The reliance on retrospective data from patient records could also result in incomplete or missing information. Finally, the study does not include genetic testing, which would enhance diagnostic accuracy.

CONCLUSION

This study provides valuable insights into the prevalence of hemoglobinopathies and their associated hematological and biochemical markers. The findings underscore the importance of early detection and comprehensive profiling for effective management. Future research should incorporate genetic testing for more precise diagnoses and explore broader populations to enhance generalizability.

REFERENCES

1. Mansoor N, Meraj F, Shaikh A, Jabbar N. Spectrum of hemoglobinopathies with hematological and biochemical profile: A five year experience from a tertiary care hospital. Pakistan Journal of Medical Sciences. 2022 Nov;38(8):2143.
2. Ghosh K, Ghosh K, Agrawal R, Nadkarni AH. Recent advances in screening and diagnosis of hemoglobinopathy. Expert review of hematology. 2020 Jan 2;13(1):13-21.
3. Aleluia MM, Fonseca TC, Souza RQ, Neves FI, da Guarda CC, Santiago RP, Cunha BL, Figueiredo CV, Santana SS, da Paz SS, Ferreira JR. Comparative study of sickle cell anemia and hemoglobin SC disease: clinical characterization, laboratory biomarkers and genetic profiles. BMC hematology. 2017 Dec;17:1-0.
4. Giambona A, Passarello C, Renda D, Maggio A. The significance of the hemoglobin A2 value in screening for hemoglobinopathies. Clinical biochemistry. 2009 Dec 1;42(18):1786-96.
5. Khaled SA, Ahmed HA, Elbadry MI, NasrEldin E, Hassany SM, Ahmed SA. Hematological, biochemical properties, and clinical correlates of hemoglobin S variant disorder: a new insight into sickle cell trait. Journal of Hematology. 2022 Jun 27;11(3):92.
6. Maier-Redelsperger M, Lévy P, Lionnet F, Stankovic K, Haymann JP, Lefèvre G, Avellino V, Perol JP, Girot R, Elion J. Strong association between a new marker of hemolysis and glomerulopathy in sickle cell anemia. Blood Cells, Molecules, and Diseases. 2010 Dec 15;45(4):289-92.
7. Yerlikaya E, Karagecili H, Kaya MO. Comparison of biochemical parameters of prevalent hemoglobinopathies with healthy individuals. Medicine. 2020;9:708-13.
8. Quinn CT, Smith EP, Arbabi S, Khera PK, Lindsell CJ, Niss O, Joiner CH, Franco RS, Cohen RM. Biochemical surrogate markers of hemolysis do not correlate with directly measured erythrocyte survival in sickle cell anemia. American journal of hematology. 2016 Dec;91(12):1195-201.

9. Begum F. A Study of Biochemical Abnormalities in Thalassemia and Hemoglobinopathies Prevalent in North-East India. Sch J App Med Sci. 2021 Jul;7:1241-7.
10. Chen Z, Shao L, Jiang M, Ba X, Ma B, Zhou T. Interpretation of HbA1c lies at the intersection of analytical methodology, clinical biochemistry and hematology. Experimental and Therapeutic Medicine. 2022 Oct 4;24(6):707.
11. Yahouédéhou SC, da Guarda CC, Figueiredo CV, Santiago RP, Carvalho SP, Fiuza LM, Ndidi US, Oliveira RM, Carvalho MO, Nascimento VM, Rocha LC. Hydroxyurea alters hematological, biochemical and inflammatory biomarkers in Brazilian children with SCA: Investigating associations with βS haplotype and α-thalassemia. PloS one. 2019 Jul 15;14(7):e0218040.
12. Yahouédéhou SC, da Guarda CC, Figueiredo CV, Santiago RP, Carvalho SP, Fiuza LM, Ndidi US, Oliveira RM, Carvalho MO, Nascimento VM, Rocha LC. Hydroxyurea alters hematological, biochemical and inflammatory biomarkers in Brazilian children with SCA: Investigating associations with βS haplotype and α-thalassemia. PloS one. 2019 Jul 15;14(7):e0218040.
13. Harteveld CL, Achour A, Arkesteijn SJ, Ter Huurne J, Verschuren M, Bhagwandien-Bisoen S, Schaap R, Vijfhuizen L, El Idrissi H, Koopmann TT. The hemoglobinopathies, molecular disease mechanisms and diagnostics. International Journal of Laboratory Hematology. 2022 Sep;44:28-36.
14. Jeenduang N, Horpet D, Plyduang T, Nuinoon M. Association of thalassemia, hemoglobinopathies, and vitamin D levels with lipid profile in adults: Community-based research in southern Thai population. Heliyon. 2024 May 30;10(10).
15. Mansoor N, Meraj F, Shaikh A, Jabbar N. Spectrum of hemoglobinopathies with hematological and biochemical profile: A five year experience from a tertiary care hospital. Pakistan Journal of Medical Sciences. 2022 Nov;38(8):2143.
16. Kuppusamy D, Vinod KV, Kar R. Screening for hemoglobin disorders and investigating their hematological and demographic profile among patients attending a tertiary-care hospital in southern India—a descriptive study. Porto Biomedical Journal. 2024 Sep 1;9(5):271.
17. Balgir RS. Hematological profile of hemoglobinopathies in maternal health and reproductive outcome in pregnant mothers at a tertiary hospital in central India. J Hematol Mult Myeloma. 2018;3:1012.
18. Rakib NM, Rishad MM, Prince SK, Jannat M, Khan TR, Zinan TI, Jeshan T, Kabir MA. Types of Inherited Hemoglobin Disorders Among the Patients Attending Hematology Outdoor of a Tertiary Care Hospital in Bangladesh. Journal of Medicine. 2023 Jul 2;24(2):96-105.
19. Rehman A, Ahmed S, Ansari U. Clinical and haematological characteristics of haemoglobin H. Pak J Med Health Sci. 2015;9(3):1096-8.
20. Subramanian T, Parvathi G, Kumar CK, Lakshmi CV. A study on the hematological profiles of hemoglobinopathies in pregnant women in a tertiary health care center. Int J Acad Med Pharm. 2021;5(2):45-51.