Section: Pathology

Original Research Article

A STUDY ON THE HEMATOLOGICAL PROFILES OF HEMOGLOBINOPATHIES IN PREGNANT WOMEN IN A TERTIARY HEALTH CARE CENTER

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Received : 07/11/2023
Received in revised form : 29/12/2023
Accepted : 14/01/2024

Keywords:
Hemoglobinopathies, Pregnancy, Hematological Profiles, Sickle Cell Anemia, β-Thalassemia, Hemoglobin Levels, RBC Count, Tertiary Health Care.

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DOI: 10.47009/jamp.2024.6.1.71

Source of Support: Nil,
Conflict of Interest: None declared

Int J Acad Med Pharm
2024; 6 (1); 369-373

Abstract

Background: Hemoglobinopathies, a group of genetic disorders affecting hemoglobin structure or synthesis, pose significant health risks, particularly in pregnant women. This study aims to elucidate the hematological profiles of various hemoglobinopathies in pregnant women at a tertiary health care centre. **Material & Methods:** A total of 826 pregnant women underwent comprehensive screening, including sickling tests, peripheral smears, complete blood counts, and High-Performance Liquid Chromatography (HPLC) for those indicative of Beta Thalassemia. **Results:** Among the participants, 150 (18.16%) were diagnosed with hemoglobinopathies. The age group 21-25 years had the highest prevalence (60.66%). The study identified Sickle cell trait (10.8% of screened, 59% of diagnosed), Sickle cell anemia (3.02%, 17%), β-Thalassemia trait (1.4%, 8%), Sickle-β thalassemia double heterozygous trait (2.6%, 15%), and HbE-β Thalassemia (0.2%, 1%). Hematological analysis showed varying degrees of anemia, with mean hemoglobin levels and RBC counts differing significantly across the hemoglobinopathies. Sickle cell anemia cases often presented with severe anemia, whereas other conditions showed milder forms. **Conclusion:** This study highlights the significant prevalence and diverse hematological impacts of hemoglobinopathies in pregnant women. Early diagnosis and tailored management are crucial for maternal and fetal health. The findings underscore the need for routine screening for hemoglobinopathies in pregnant women.

INTRODUCTION

Hemoglobinopathies, characterized by genetic alterations affecting the structure or synthesis of hemoglobin, are among the most common genetic disorders worldwide.[1] They pose significant health challenges, particularly in the context of pregnancy, where they can impact both maternal and fetal outcomes.[2,3] This study focuses on the hematological profiles of hemoglobinopathies in pregnant women, with an aim to provide insights into their prevalence and clinical implications in a tertiary health care setting.

The importance of understanding hemoglobinopathies in pregnancy cannot be overstated. Conditions such as Sickle Cell Disease (SCD), Sickle Cell Trait (SCT), and various forms of thalassemia have been associated with increased risks of maternal anemia, miscarriages, preterm births, and low birth weight infants.[4,5,6] Moreover, the physiological changes in pregnancy can exacerbate the symptoms and complications of these disorders, posing additional risks to maternal health.

Despite these challenges, the diagnosis of hemoglobinopathies during pregnancy remains underemphasized in many regions. The lack of awareness and limited access to screening and diagnostic facilities contribute to this gap.[7] Furthermore, the wide spectrum of hematological manifestations of these disorders—from mild anemia to severe complications—makes their management in pregnancy a complex clinical endeavor.

This study aims to bridge this knowledge gap by providing a comprehensive analysis of the hematological profiles of various hemoglobinopathies in pregnant women. By

ISSN (O): 2687-5365; ISSN (P): 2753-6556

assessing parameters such as hemoglobin levels, red blood cell counts, and the severity of anemia, this research seeks to underscore the significance of routine screening and personalized care in managing these conditions during pregnancy. The findings are expected to contribute to better clinical guidelines and improved outcomes for both mothers and their babies.

In summary, this research not only highlights the prevalence of hemoglobinopathies among pregnant women but also emphasizes the critical need for their early detection and management in prenatal care. The results are anticipated to be a valuable addition to the existing literature and a guide for healthcare providers in optimizing maternal and fetal health in the context of hemoglobinopathies.

MATERIALS AND METHODS

Study Setting and Design: This hospital-based observational study was conducted at the Department of Obstetrics and Gynecology OPD and inpatient wards of King George Hospital, a tertiary health care center. The study spanned 22 months, from January 2021 to October 2022.

Study Population and Sample Size: The study population comprised pregnant women visiting the Department of Obstetrics and Gynecology. A total of 826 pregnant women were initially screened, out of which 150 met the inclusion criteria and were included in the study.

Inclusion Criteria

Pregnant women who provided informed consent.

Pregnant women with hemoglobin (Hb) levels less than 11 g/dL, mean corpuscular volume (MCV) less than 80 femtoliters (fl), mean corpuscular hemoglobin (MCH) less than 27 picograms (pg), or positive sickling test.

Pregnant women with a history of multiple blood transfusions, pain crises, recurrent infections, splenomegaly, or a family history of hemoglobinopathy.

Exclusion Criteria

Pregnant women who had received a blood transfusion within the last three months.

Ethical Considerations: The study was approved by the Institutional Ethics Committee (IEC) of Andhra Medical College/King George Hospital. Informed consent was obtained from all participating women.

Sample Collection and Handling

Venous blood (3ml) was drawn from each participant under aseptic conditions into an EDTA (Ethylene Diamine Tetra Acetic Acid) vacutainer.

Blood samples were stored at 2 to 8 degrees Celsius until analysis.

Laboratory Analysis

Complete Blood Count (CBC): Performed using a Sysmex five-component hematology analyzer.

Peripheral Smear Preparation and Leishman Staining: Blood films were prepared from EDTA-anticoagulated blood, air-dried, stained with Leishman stain, and examined under a microscope.

Reticulocyte Count: Conducted using New Methylene Blue stain and examined under a microscope with a 100X oil-immersion objective.

Sickling Test: Performed using a freshly made 2% sodium metabisulphite solution mixed with a blood drop, covered with a waxed cover slip, and examined at various intervals.

High-Performance Liquid Chromatography (HPLC): Samples were analyzed using a BIORAD variant Hb typing system TOSHO G8 Analyzer - Variant β-Thal Short program. Haemoglobin variants were identified based on their elution in specific windows and distinguished by concentration, smear findings, and clinical correlation.

Data Analysis

Results were documented and analyzed using Microsoft Office Excel for statistical significance. Comparisons were made across different types of hemoglobinopathies, focusing on hemoglobin levels, RBC count, and other relevant hematological parameters.

Controls: Control samples were run with every batch of tests to ensure accuracy and reliability of the results.

Ethical Considerations

This study protocol was approved by the Institutional Ethics Committee, Andhra Medical College, Visakhapatnam and addressing the specific needs of pregnant women as a vulnerable population. These measures ensured the study's integrity and compliance with ethical standards.

RESULTS

Study Population and Demographics

A comprehensive screening for hemoglobinopathies was conducted on 826 pregnant women at a tertiary health care center. This included sickling tests, peripheral smears, and complete blood counts. Women showing signs indicative of Beta Thalassemia on these tests were further evaluated with High-Performance Liquid Chromatography (HPLC).

Of the 826 women screened, 150 (18.16%) were diagnosed with various hemoglobinopathies. The majority of these women were in the 21-25 age group, constituting 60.66% of the diagnosed cases.

Types and Prevalence of Hemoglobinopathies

The study found several types of hemoglobinopathies among the pregnant women:

Sickle cell trait was identified in 89 cases (10.8% of the screened population, 59% of diagnosed cases).

Sickle cell anemia was found in 25 cases (3.02% of the screened population, 17% of diagnosed cases).

β-Thalassemia trait was present in 12 cases (1.4% of the screened population, 8% of diagnosed cases).

Sickle-β thalassemia double heterozygous trait occurred in 22 cases (2.6% of the screened population, 15% of diagnosed cases).

ISSN (O): 2687-5365; ISSN (P): 2753-6556

HbE-β Thalassemia was diagnosed in 2 cases (0.2% of the screened population, 1% of diagnosed cases).

Hematological Profiles in Different Hemoglobinopathies

Sickle Cell Anemia

In the 25 cases of sickle cell anemia, anemia was universally present, with 20% classified as mild, 44% as moderate, and 36% as severe. The average hemoglobin level was 7.82 g/dl, and the mean RBC count was 2.93 million/mm3. The most common symptoms were fever, fatigue, paleness, and headache.

Sickle Cell Trait

Among the 89 cases with sickle cell trait, 88.8% had anemia. The severity distribution was 35.9% mild, 40.44% moderate, and 12.3% severe. The average hemoglobin level in these cases was 8.8 g/dl, and the mean RBC count was 3.5 million/mm3.

Sickle-β Thalassemia Double Heterozygous Trait

In the 22 cases of Sickle-β Thalassemia double heterozygous trait, 81.8% had anemia, predominantly of moderate severity (54.54%). The average hemoglobin level was 9.15 g/dl, with a mean RBC count of 3.77 million/mm3.

β Thalassemia Trait

All 12 cases of β Thalassemia trait showed moderate anemia. The mean hemoglobin level was 8.5 g/dl, and the average RBC count was 4.14 million/mm3.

HbE-β Thalassemia Double Heterozygous Trait

Both cases diagnosed with HbE-β Thalassemia double heterozygous trait had moderate anemia. The mean hemoglobin level was 8.2 g/dl, and the average RBC count was 3.95 million/mm3.

Comparative Analysis of Hematological Indices

A comparative analysis revealed significant variations in mean hematological indices across the different hemoglobinopathies, highlighting the diverse impacts of these conditions on hematological parameters in pregnant women.

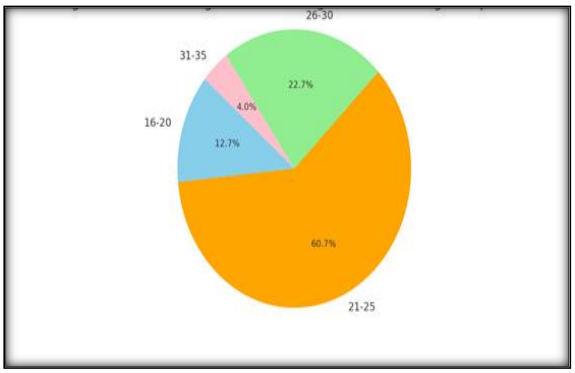

Figure 1A: Age Distribution of Pregnant Women Diagnosed with Hemoglobinopathies

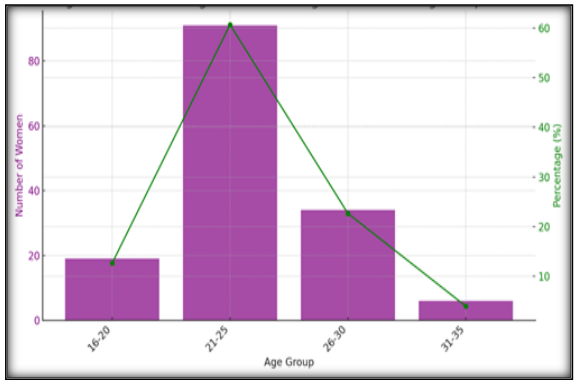

Figure 1 B: Age Distribution of Pregnant Women Diagnosed with Hemoglobinopathies

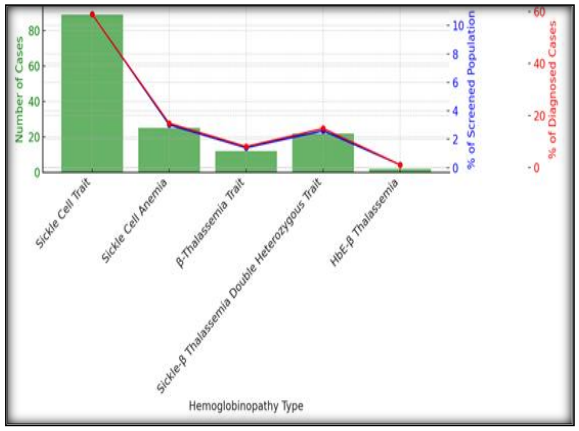

Figure 2: Prevalence of Hemoglobinopathies

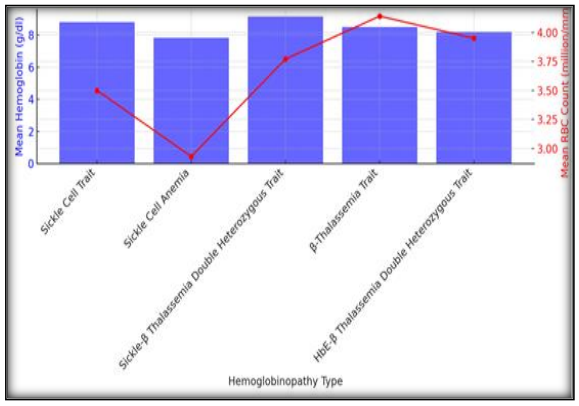

Figure 3: Comparative Analysis of Hematological Indices

Table 1: Age Distribution of Pregnant Women Diagnosed with Hemoglobinopathies

Age Group	Number of Women	Percentage (%)
16-20	19	12.66
21-25	91	60.66
26-30	34	22.66
31-35	6	4
Total	150	100

ISSN (O): 2687-5365; ISSN (P): 2753-6556

Table 2: Prevalence of Hemoglobinopathies

Hemoglobinopathy Type	Number of Cases	% of Screened Population	% of Diagnosed Cases
Sickle Cell Trait	89	10.8%	59%
Sickle Cell Anemia	25	3.02%	17%
β-Thalassemia Trait	12	1.4%	8%
Sickle-β Thalassemia Double Heterozygous Trait	22	2.6%	15%
HbE-β Thalassemia	2	0.2%	1%

Table 3: Hematological Profiles - Sickle Cell Anemia

Parameter	Mean Value	Severity of Anemia (%)
Hemoglobin (g/dl)	7.82	Mild: 20%, Moderate: 44%, Severe: 36%
RBC Count (million/mm3)	2.93	-

Table 4: Hematological Profiles - Sickle Cell Trait

Parameter	Mean Value	Severity of Anemia (%)
Hemoglobin (g/dl)	8.8	Mild: 35.9%, Moderate: 40.44%, Severe: 12.3%
RBC Count (million/mm3)	3.5	-

Table 5: Hematological Profiles - Sickle-β Thalassemia Double Heterozygous Trait

Parameter	Mean Value	Severity of Anemia (%)
Hemoglobin (g/dl)	9.15	Moderate: 54.54%
RBC Count (million/mm3)	3.77	-

Table 6: Hematological Profiles - β Thalassemia Trait

Parameter	Mean Value	Severity of Anemia (%)
Hemoglobin (g/dl)	8.5	Moderate: 100%
RBC Count (million/mm3)	4.14	-

Table 7: Hematological Profiles - HbE-β Thalassemia Double Heterozygous Trait

Parameter	Mean Value	Severity of Anemia (%)
Hemoglobin (g/dl)	8.2	Moderate: 100%
RBC Count (million/mm3)	3.95	-

Table 8: Comparative Analysis of Hematological Indices

Hemoglobinopathy Type	Mean Hemoglobin (g/dl)	Mean RBC Count (million/mm3)
Sickle Cell Trait	8.8	3.5
Sickle Cell Anemia	7.82	2.93
Sickle-β Thalassemia Double Heterozygous Trait	9.15	3.77
β-Thalassemia Trait	8.5	4.14
HbE-β Thalassemia Double Heterozygous Trait	8.2	3.95

DISCUSSION

This study, conducted at a tertiary health care center, provides significant insights into the hematological profiles of various hemoglobinopathies in pregnant women. The findings indicate a notable prevalence of hemoglobinopathies, with 18.16% of the screened pregnant women being diagnosed with one or more forms of the condition. This prevalence emphasizes the importance of routine screening for these genetic disorders in pregnant populations.[8]

Prevalence and Demographics: The most common hemoglobinopathy identified was the Sickle cell trait, followed by Sickle cell anemia, β-Thalassemia trait, Sickle-β thalassemia double heterozygous trait, and HbE-β Thalassemia. The highest prevalence was observed in the age group of 21-25 years. This demographic distribution aligns with the reproductive age group, underscoring the need for targeted screening and counseling in this population.[9,10]

Hematological Variations: The study revealed significant differences in mean hemoglobin levels and RBC counts among different hemoglobinopathies.[11] Sickle cell anemia cases often presented with severe anemia, highlighting the critical need for specialized care in this subgroup.[12] In contrast, conditions like Sickle cell trait and β-Thalassemia trait showed milder forms of anemia, which could lead to misdiagnosis or underestimation of the condition's impact on maternal and fetal health.[13]

Clinical Implications: These findings have important implications for the management of pregnant women with hemoglobinopathies. Early diagnosis and tailored management strategies are crucial. The variability in hematological impact among different hemoglobinopathies necessitates a personalized approach to care, considering the specific needs and risks associated with each condition.[14]

Screening and Counseling: The significant prevalence found in this study advocates for routine

ISSN (O): 2687-5365; ISSN (P): 2753-6556

screening for hemoglobinopathies in pregnant women. Early detection can facilitate better management of the condition, potentially improving maternal and fetal outcomes. Moreover, genetic counseling should be offered to affected individuals and their families to inform them about the risks and implications of these genetic disorders.

Limitations and Future Research: While this study provides valuable information, it is limited by its hospital-based design and may not represent the general population. Further community-based studies are needed to understand the broader prevalence and impact of hemoglobinopathies. Additionally, research focusing on the long-term outcomes for both mothers and their offspring could provide deeper insights into the management and prognosis of these conditions.

CONCLUSION

The present study reveals a notable increase in the prevalence of hemoglobinopathies among pregnant women, emphasizing the necessity for routine screening. Such screening is crucial not only for the immediate management of the current pregnancy but also for informed planning of future pregnancies. The study underscores the need for increased awareness about hemoglobinopathies, particularly among pregnant women and individuals in the reproductive age group. The findings highlight the diverse hematological impacts these genetic disorders have on the obstetric population and the importance of implementing tailored management strategies to effectively address the specific challenges they present.

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ISSN (O): 2687-5365; ISSN (P): 2753-6556