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Research Article

Spectrum of Non-Thalassemic Hemoglobin Variants and Their Hematological Profile in Antenatal Women: An HPLC Study

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ABSTRACT

Background: Hemoglobinopathies are a significant public health concern in India, with varying regional distribution. Antenatal screening using high-performance liquid chromatography (HPLC) enables early detection of hemoglobin variants and helps in preventing adverse genetic outcomes. Evaluation of red cell indices further aids in understanding the hematological profile of these cases.
Aim: To determine the prevalence of non-thalassemic hemoglobin variants in antenatal women and to correlate them with red cell indices.
Materials and Methods: This cross-sectional study included 3000 antenatal women who underwent HPLC testing as part of routine antenatal screening program under Telangana Diagnostics in Vikarabad District. Recent history of Blood transfusion and Cases suggestive of β-thalassemia trait (elevated HbA_2 levels) were excluded. Hemoglobin variants were identified based on retention time and peak characteristics. Red cell indices, including mean corpuscular volume (MCV) and mean corpuscular hemoglobin (MCH), were analyzed in abnormal cases. Data were expressed using descriptive statistics.
Results: Out of 3000 cases, 14 (0.46%) showed abnormal hemoglobin variants, while 2986 (99.6%) were normal. The variants detected included HbS, HbD, HbE, hereditary persistence of fetal hemoglobin (HPFH) and Delta- beta thalassemia trait. The mean MCV and MCH among abnormal cases were [70.07] fL and [22.8] pg, respectively.
Conclusion: The prevalence of non-thalassemic hemoglobin variants in the studied antenatal population was low. However, HPLC remains an effective screening tool, and correlation with red cell indices provides additional hematological insight. Routine antenatal screening is essential for early detection and genetic counseling.

Keywords: *Hemoglobinopathies, High performance liquid chromatography, Hb S, Hb D, Hb E, Hereditary persistence of fetal hemoglobin*

INTRODUCTION

Hemoglobinopathies are a group of inherited disorders triggered by defects in globin genes which lead to abnormal hemoglobin (Hb) level and structure with reduced oxygen-carrying capacity. The abnormality may be quantitative or qualitative. Qualitative defects can occur due to genetic mutations that involve globin protein chains. These defects include either amino acid deletions or substitutions, which cause structural variations of the globin chain that manifest in the form of HbS, HbD, HbE, etc. [1] Adult Hb profiles are normally composed of 95-98% of HbA, 2-3.5% HbA2 and 0.8-2% HbF. The latter type i.e. HbF is higher in newborns (50-80%) and drops to 1-2% six months after birth with rise in HbA.Hemoglobinopathies can be diagnosed by examining different Hb variants i.e. HbA, HbA2, HbF, HbS, HbE, HbC and HbD) and their concentrations are measured to predict the disease severity[2]. High-performance liquid chromatography (HPLC) is a reliable and widely used technique due to its accuracy and reproducibility [3,4].

Antenatal screening is an important strategy for early identification of hemoglobinopathies and prevention of severe disease in offspring [5]. Previous recommendations for hemoglobinopathy testing have used a race/ethnicity-based

strategy. However, race and self-identified ethnicity are poor proxies for genetics since self-identification with a specific race/ethnicity may be incompatible with genetic ancestry.

World literature states that heterozygous carriers of hereditary disorders of Hb are >270 million. Among them, at least 3,00,000 affected homozygotes or compound heterozygotes are born each year [6].Mortality and morbidity associated with these hemoglobinopathies increase a huge burden on health sector facilities and therefore country's economy. We can reduce the incidence of these hemoglobinopathies by carrier screening in whole population in areas where prevalence is high/at least testing the antenatal women. Once couples at risk of inheriting hemoglobinopathies are identified, still we can avoid the birth of an affected fetus by invasive Prenatal diagnosis like amniocentesis and chorionic villi sampling.It also adds to social and psychological problems and poor quality of life [7].

Red cell indices such as MCV and MCH provide useful hematological insights and may aid in differentiating various hemoglobinopathies [4].

MATERIALS AND METHODS

This cross-sectional study was conducted in the Department of Pathology at Telangana Diagnostics Hub at Vikarabad District.

The study included antenatal women whose samples were received for hemoglobin variant analysis as part of the routine screening program under Telangana Diagnostics.

A total of 3000 samples were analyzed.

Inclusion Criteria:
All antenatal samples referred for HPLC screening

Exclusion Criteria:
Cases suggestive of β-thalassemia trait

Recent transfusion cases

METHODOLOGY

EDTA blood samples collected

HPLC performed using - BIORAD D10.

Variants identified based on retention time and peak characteristics

Statistical Analysis:Descriptive statistics were used.

RESULTS

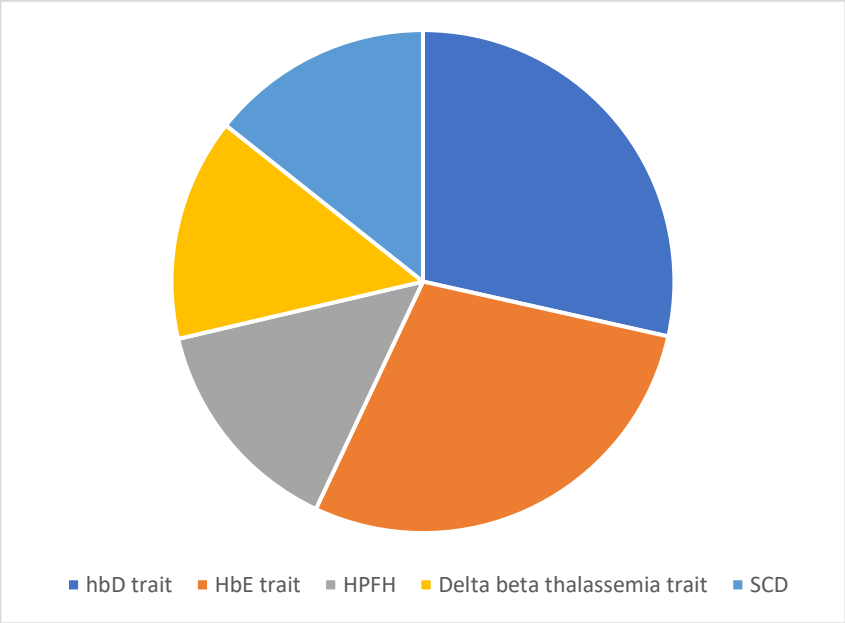
Out of 3000 cases, 14 (0.46%) showed abnormal hemoglobin variants. The variants detected included HbS trait (7),Sickle cell disease (1), HbD trait (2), HbE trait(2),HPFH (1) and Delta beta thalassemia trait (1). The mean MCV and MCH among abnormal cases were [70.07] fL and [22.8] pg, respectively.

Additionally spouse testing by HPLC was done after appropriate genetic counselling to assess carrier status and risk of hemoglobinopathies in offspring. All tested spouses showed normal HPLC findings.

Table: 1

VARIANT	NUMBER	PERCENTAGE
Sickle cell trait	7	50
Sickle cell anaemia	1	7.14
HbD	2	14.2
HbE	2	14.2
HPFH	1	7.14
Delta beta Thalassemia	1	7.14

Figure: 1



DISCUSSION

Hemoglobinopathies represent a major genetic health burden in India with significant regional variation [8].Beyond the clinical burden, these disorders impose substantial emotional and economic strain on affected individuals, their families, and society at large. Consequently, several screening strategies have been developed to identify carriers and reduce disease incidence.

The primary objective of screening programs is to prevent the birth of affected individuals. Different approaches include population-based screening, targeted screening of high-risk groups, family (cascade) screening, premarital screening, and antenatal screening. However, universal population screening is often impractical in India due to logistical and resource constraints.

Cascade screening, particularly among extended family members of affected individuals, has proven to be an efficient strategy. It enables identification of a larger number of at-risk individuals while screening only a limited segment of the population. Additionally, relatives of children with thalassemia major are more likely to participate due to their prior exposure to the disease burden, thereby improving compliance with carrier detection programs.

Premarital screening is considered an effective preventive strategy, as it allows individuals to make informed decisions regarding partner selection and to understand the implications of carrier status. It also facilitates timely prenatal diagnosis and early reporting of pregnancy. However, its implementation in India faces sociocultural challenges, including the prevalence of arranged marriages and the social stigma associated with carrier status.Screening of pregnant women attending antenatal clinics is a practical and effective approach, particularly in urban and semi-urban regions of India where routine antenatal visits are common. Women identified as carriers of thalassemia should receive appropriate genetic counseling, and their spouses should also undergo screening to assess the risk to the foetus [9].

Limited awareness and inadequate education about the disease contribute significantly to reluctance among spouses of carriers to undergo screening, a finding that has also been documented in earlier studies [10].

In this study, prevalence was 0.4%, lower compared to other Indian studies [5]. This may be due to regional variation and exclusion of β-thalassemia trait.

In present study, Sickle cell trait was the most common hemoglobinopathy -7 cases (50%), followed by HbD trait- 2 cases and HbE Heterozygous-2 cases, with (14.2%) each. Other cases detected were Sickle cell disease -1 (7.14%), Delta-Beta thalassemia trait-1 case (7.14%), Hereditary persistence of Fetal hemoglobin-1 case (7.14%). Partner screening was also done with HPLC

Variants such as HbS, HbD, HbE, and HPFH observed in this study are commonly reported in Indian populations [5] and HPLC proved to be an effective diagnostic tool.

Red cell indices such as MCV and MCH provided additional hematological insight.However detection of hemoglobinopathic carriers cannot be reliably performed by complete blood count alone as it may fail to detect few carriers of HbS, HbC, HbD, or HbE who may have a normal MCV or MCH[11].

CONCLUSION

The prevalence of non-thalassemic hemoglobin variants was low. However, HPLC remains an effective screening tool, and correlation with red cell indices provides additional hematological insight.

In conclusion, our findings highlight the need for increased public awareness regarding thalassemia and other hemoglobinopathies through coordinated efforts by governmental and non-governmental organizations, which may contribute to more effective screening and prevention strategies

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