Section: Pathology

Original Research Article

A STUDY OF HEMOGLOBINOPATHIES USING HPLC AS A DIAGNOSTIC TOOL IN ANTENATAL WOMEN AT A TERTIARY CARE HOSPITAL

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ABSTRACT

Background: Hemoglobinopathies are inherited blood disorders that constitute a major portion of genetic diseases in India. Early detection and categorization are essential for guiding clinical management and counseling at-risk families. **Objective:** To evaluate the role of High Performance Liquid Chromatography (HPLC) as a diagnostic tool in detecting hemoglobinopathies among antenatal women attending a tertiary care hospital.

Materials and Methods: A total of 238 blood samples from antenatal women were analyzed using HPLC over a ten-month period at Government Erode Medical College and Hospital, Perundurai. Red blood cell indices were also assessed.

Results: 13% of the participants were diagnosed with hemoglobinopathies. Of the 31 cases detected, 58% were Beta Thalassemia Trait and 42% were Sickle Cell Trait. The majority of affected individuals were between 20–30 years of age.

Conclusion: HPLC is an effective tool for early diagnosis of hemoglobinopathies in antenatal women, enabling timely counseling and intervention to reduce hematological complications.

Keywords: Hemoglobinopathy, Beta Thalassemia, Sickle Cell Trait, HPLC, Antenatal Screening, Genetic Disorders.

INTRODUCTION

Hemoglobinopathies are a significant global health concern, with approximately 7% of the world population being carriers of abnormal hemoglobin genes.[1] Each year, around 0.3 million infants are born with major hemoglobin disorders.[2] India alone has about 42 million carriers of Beta Thalassemia trait, the highest in the world.[3] In various regions of India, carrier frequencies vary:

- 6.5% in Punjab
- 8.4% in Tamil Nadu
- 4.3% in South India
- 3.5% in Bengal

Highest prevalence reported in Uttar Pradesh.[4] In Central India and states like Gujarat, Maharashtra, and Kerala, the frequency of hemoglobinopathies ranges from 11% to 35%.[3] Early identification through antenatal screening helps prevent severe complications by enabling genetic counseling and prenatal diagnosis.[5]

MATERIALS AND METHODS

2.1 Study Design and Population

This was a cross-sectional study conducted over ten months (September 2024 – June 2025) at the Departments of Pathology and Biochemistry, Government Erode Medical College and Hospital, Perundurai.

2.2 Sample Collection

Blood samples from 238 antenatal women were collected in EDTA vacutainers. Clinical history and relevant investigations were conducted for all participants.

2.3 Hematological Analysis

- Samples were analyzed using an automated hematology analyzer (SYSMEX XP-300).

Parameters assessed included: Hemoglobin (g/dL)

- Mean Corpuscular Volume (MCV)
- Mean Corpuscular Hemoglobin (MCH)
- Mean Corpuscular Hemoglobin Concentration (MCHC)
- Red Cell Distribution Width – Coefficient of Variation (RDW-CV)
- Peripheral smear was also examined.

2.4 HPLC Analysis

All samples were subjected to HPLC using the BIORAD Variant-D10 system. Chromatograms and reports were obtained for every case to detect and differentiate hemoglobin variants.[6]

RESULTS

Out of 238 antenatal women screened,

- 31 (13%) were found to have hemoglobinopathies.
- 18 (58%) were diagnosed with Beta Thalassemia Trait
- 13 (42%) were found to have Sickle Cell Trait

Table 1: Distribution of all cases (238) in the study

Hb Pattern	No of cases	Percentage
Normal Hb pattern	207	87%
Beta thalassemia trait	18	7.6%
Sickle cell trait	13	5.4%

As the study proceeded, a total of 238 antenatal females (mean age 20 to 30 years) were screened. HPLC was found abnormal in 31 cases. The major chunk (n = 18; 58%) was beta thalassemia trait affected women, following it was sickle cell trait (with n = 13 ; 42%), as figured in table 1.

Table 2: Hematological parameters of hemoglobinopathies in this study

Parameter	Beta Thalassemia Trait	Sickle Cell Trait
Hb (g/dL)	**Mean: 9.25 ± 1.73**	**Mean: 10.36 ± 1.70**
MCV (fL)	**Mean: 66.2 ± 6.37**	**Mean: 74.3 ± 7.55**
MCH (pg)	**Mean: 20.52 ± 2.42**	**Mean: 24.6 ± 2.24**
MCHC (g/dL)	**Mean: 30.5 ± 1.06**	**Mean: 32.6 ± 1.35**
RDW-CV (%)	**Mean: 18.64 ± 4.42**	**Mean: 16.57 ± 2.09**

Table-2 illustrates the haematological data expressed as mean ± standard deviation of hemoglobinopathies studied in this study. The mean hemoglobin values were low in women with Beta Thalassemia Trait, whereas the MCV and MCH is higher in Sickle Cell Trait than Beta Thalassemia Trait.

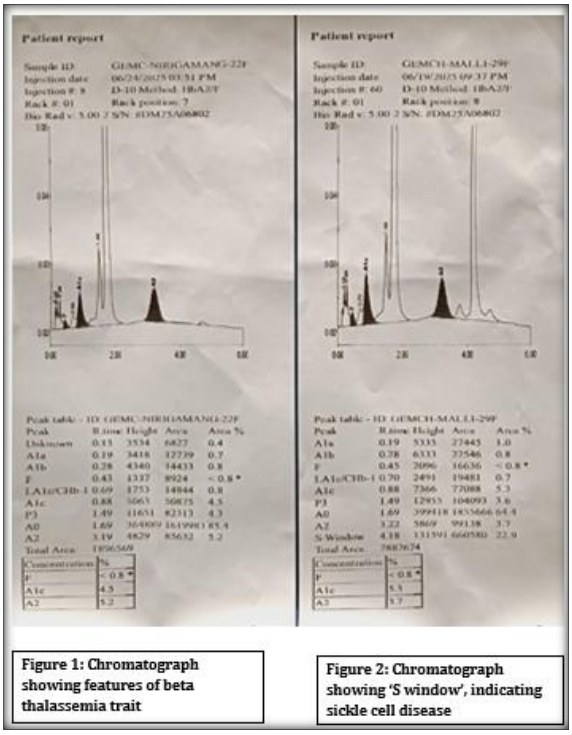

Figure 1: Chromatograph showing features of beta thalassemia trait

Figure 2: Chromatograph showing 'S window', indicating sickle cell disease

The most commonly affected age group was 20–30 years. The findings reinforce the significance of early and routine antenatal screening for inherited hemoglobin disorders.

DISCUSSION

Hemoglobinopathies namely Thalassemia and Sickle Cell Anemia are the most severe forms and need great importance to be dealt with as a public health point of view in India. Thalassemia is classified according to the globin chain deficit. Two major forms include impaired production and stability of either α or β peptide chains causing α-thalassemia or β-thalassemia. There is also an overlap between these groups called compound heterozygous state.[8] β-thalassemia minor has no specific therapy. The screening can be done in prenatal stage, but it may not be possible in all communities and ethnic groups, so the ideal candidate for this would be pregnant women.

Routine hematological parameters like RBC indices along with HPLC can detect carrier state of different hemoglobinopathies which are clinically silent.[9]

In our study the most prevalent hemoglobinopathy is Thalassemia (7.6%). Yet, only a few studies are available providing data related to various hemoglobinopathies in terms of incidence,

prevalence, morbidity and mortality. Next was Sickle Cell Trait (5.4%). Comparable study by Narang et al (Punjab, North India) showed similar results as our present study.

During this present analysis, it was observed that Beta Thalassemia Trait patients had severe anemia, low MCV and MCH as compared to Sickle Cell Trait. Microcytic hypochromic anemia was noted. Compared with Khera et al, Srinivas et al, and Dhawle et al, the same results were shown.[10-12] Peripheral smear study along with routine hemogram will help in diagnosing hemoglobinopathies.Proper screening and early diagnosis of hemoglobinopathies plays a vital role in their management and treatment. HPLC is emerging as one of the best methods for screening and detection of various hemoglobinopathies. Our study provides an additional data about hemoglobinopathies in the reproductive age group/female population.

This study aligns with national data on the high prevalence of Beta Thalassemia and Sickle Cell traits in India.[3,4] The detection rate of 13% among antenatal women highlights the urgent need for screening programs at the primary healthcare level. The integration of RBC indices with HPLC findings significantly improves diagnostic accuracy.[6] Early diagnosis enables timely counseling, reducing the burden of severe transfusion-dependent anemia in offspring.[5]

CONCLUSION

This approach will help in appropriate screening and detection of women who are carriers. Spouse testing and couple counselling at risk will reduce the morbidity and mortality from a potential homozygous offspring. However, it is essential to consider clinical history, hematological parameters and family studies for a comprehensive diagnosis and to address potential challenges posed by iron deficiency and genetic interactions. In conjunction with other diagnostic methods, HPLC plays a vital role in thalassemia screening and management programs.

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