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Original Article

Screening of Various Abnormal Hemoglobins in Antenatal Women-A Prospective Study

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ABSTRACT

Background: Hemoglobinopathies including β-thalassemia, sickle cell disease and other abnormal hemoglobin variants are significant causes of maternal anemia and adverse fetal outcomes in India. Early detection through antenatal screening allows for timely genetic counseling and prenatal management.
Objectives: To determine the prevalence and spectrum of abnormal hemoglobins in antenatal women and to assess the severity of anemia in affected individuals.
Methods: A prospective study was conducted on antenatal women attending OPD at a tertiary care hospital. Blood samples were screened for abnormal hemoglobins using High-Performance Liquid Chromatography (HPLC). Hemoglobin levels were measured, and anemia was classified as mild (10–13 g/dL), moderate (7–10 g/dL), or severe (<7 g/dL).
Results: Among 159 women diagnosed with abnormal hemoglobins:
• β-thalassemia trait was most common (87 cases, 54.73%), followed by sickle cell trait (54 cases, 33.97%).
• Less frequent variants included Hb D Punjab (8, 5.03%), Hb E heterozygous (4, 2.52%), HPFH (3, 1.89%), and rare variants Hb Q India, Hb J India, Hb D Iran/Deer Lodge (1 case each, 0.62%).
Conclusion: β-thalassemia and sickle cell traits are the most prevalent hemoglobinopathies in antenatal women with a significant proportion presenting with moderate to severe anemia. Routine antenatal screening using HPLC is recommended to identify carriers, provide genetic counseling and guide prenatal management, reducing the risk of severe hemoglobinopathies in offspring.

Keywords*: Hemoglobinopathies, β-thalassemia, Sickle Cell Trait, Antenatal Screening, HPLC, Anemia.*

INTRODUCTION

Thalassemia and other hemoglobinopathies are among the most common inherited hematological disorders worldwide. According to the World Health Organization(WHO), approximately 7% of the global population are carriers of hemoglobin (Hb) disorders [1,2]. Individuals inheriting two abnormal alleles may present with a spectrum of clinical manifestations, including anemia, acute pain crises, vaso-occlusive events, acute chest syndrome, fatigue, hemolysis, and jaundice**.**

The most effective strategy to reduce the burden of these disorders is the implementation of population-level screening programs**,** accompanied by timely genetic counseling and appropriate interventions for affected individuals [3]. Although these disorders occur globally, their prevalence varies significantly between regions and even within countries, and is gradually increasing due to population migration [1,2].

In India, β-thalassemia is the most common hemoglobinopathy. Other variants show regional predominance, such as

hemoglobin S (HbS) in Odisha, hemoglobin D (HbD) in Punjab, and hemoglobin E (HbE) in the northeastern states [4,5]. This prospective study was conducted at a tertiary care center in Rajkot, Gujarat, including 2087 antenatal women. Complete blood counts were performed, and hemoglobinopathies were detected using High-Performance Liquid Chromatography (HPLC) with the BIORAD Variant II analyzer. Since antenatal women are the population most likely to benefit from early detection of inherited hemoglobin disorders, this study specifically focused on screening pregnant women.

AIM

To assess the prevalence and spectrum of thalassemia and other hemoglobinopathies among antenatal women attending a tertiary care center in the Gujarat region of India by screening during the first trimester of pregnancy.

OBJECTIVES

1. To determine the frequency and types of abnormal hemoglobin variants in antenatal women.
2. To assess the severity of anemia in women diagnosed with hemoglobinopathies.
3. To provide baseline data for regional prevalence, which can guide genetic counseling and prenatal care strategies.
4. To evaluate the utility of HPLC screening for detecting hemoglobinopathies in the antenatal population

MATERIALS AND METHODS

This prospective cross-sectional study was conducted at the tertiary care institute PDU Medical College and Hospital, Gujarat, India, and included 2087 pregnant women attending the obstetrics outpatient department. Women in the first trimester of pregnancy (up to 12 completed weeks of gestation) were included. Participants with a recent history of blood transfusion were excluded.

The purpose and importance of screening for thalassemia and hemoglobinopathies were explained to all participants, and informed written consent was obtained prior to enrollment.

For each participant, 2 milliliters (ml) of venous blood was collected in EDTA vacutainers for laboratory analysis. Complete blood counts (CBC) were performed using the Mindray BC 6200 automated analyzer. Detection and characterization of abnormal hemoglobins were carried out using High-Performance Liquid Chromatography (HPLC) with the BIORAD VARIANT II system. Chromatograms were generated for each sample and carefully analyzed to identify hemoglobin variants.

RESULTS

Among 2087 antenatal women screened in the first trimester (ages 19–40 years, gestation 4–12 weeks**),** 159 women (7.61%) were found to have abnormal hemoglobins**.**

- β-thalassemia trait was the most common (87, 4.16%), followed by sickle cell trait (54, 2.58%).
- Less frequent variants included Hb D Punjab (8, 0.38%), Hb E heterozygous (4, 0.19%), HPFH (3, 0.14%), and rare variants such as Hb D Iran, Hb Q India, and Hb J India (1 case each, 0.05%).

These findings indicate that β-thalassemia and sickle cell traits predominate**,** while rare hemoglobinopathies are also present, emphasizing the need for comprehensive antenatal screening **and** genetic counseling**.**

Table 1: Prevalence of abnormal hemoglobinopathies:

Abnormal hemoglobin variants	Total number of cases with abnormal hemoglobin	Percentage of cases(%)
Beta thalessemia minor(trait)	87	54.73 %
Sickle cell heterozygous	54	33.97 %
HbD Punjab heterozygous	8	5.03 %
HbQ India	1	0.62 %
HbJ India	1	0.62 %
Hb E Heterozygous	4	2.52 %
Hb D Iran/Hb Deer Lodge	1	0.62 %
Hereditary Persistence of fetal hemoglobin(HPFH)	3	1.89 %
Total	159	100 %

Table 2: Distribution of Hemoglobin Variants and Corresponding Anemia Severity among Antenatal Women (n = 159)

Hemoglobin levels(gm/dl)	5-5.9	6-6.9	7-7.9	8-8.9	9-9.9	10-10.9	11-11.9	12-12.9	Total
Hemoglobin Variants									
Beta thalassemia minor(trait)	23	30	10	5	10	5	2	2	87
Sickle cell heterozygous	12	2	18	15	2	1	2	2	54
HbD Punjab heterozygous	0	2	0	2	1	2	1	0	8
HbQ India	0	0	0	0	0	0	0	1	1
HbJ India	0	0	0	0	0	0	1	0	1
Hb E Heterozygous	0	0	0	0	1	1	1	1	4
Hb D Iran/Hb Deer Lodge	0	0	0	0	0	0	1	0	1
Delta beta trait/Hereditary Persistence of fetal hemoglobin(HPFH)	0	0	0	0	0	1	1	1	3
									159

Using a cut-off value of Hb of 13 gm/dl for further analysis. They were divided into three groups, as described above. Of these 159, 16% of the women had mild IDA. 40% of women had moderate IDAThe distribution of cases in the three groups is shown in Table 3.

Table 3: Severity of Anemia among Antenatal Women with Abnormal Hemoglobins (n = 159)

Severity of anemia	Total no. of cases	Percentage
Mild (10-13gm/dL)	26	16.35%
Moderate (7-10gm/dL)	64	40.25%
Severe (<7gm/dL)	69	43.4%
Total	159	100%

DISCUSSION

Hereditary hemoglobin (Hb) disorders are broadly divided into two major categories: hemoglobinopathies and thalassemia syndromes. Hemoglobinopathies arise from the synthesis of structurally abnormal globin chains, whereas thalassemia syndromes result from reduced production of otherwise normal globin chains. In India, inherited Hb disorders represent a significant healthcare challenge, with β-thalassemia, HbS, HbD, and HbE being the most frequently reported variants [6,7].

Hemoglobinopathies are inherited in an autosomal recessive pattern. Thalassemia occurs due to genetic defects that impair the production of specific globin chains—either α or β. In β-thalassemia, diminished synthesis of β-globin chains leads to reduced formation of HbA ($\alpha_2\beta_2$). This imbalance between α and non-α chains is partially compensated by increased production of δ and γ chains, resulting in elevated levels of HbA_2 ($\alpha_2\delta_2$) and HbF [8].

Sickle cell anemia develops due to specific mutations in the β-globin gene, leading to the formation of structurally abnormal HbS. When one parent carries the β-thalassemia trait and the other has normal hemoglobin, each pregnancy carries a 50% chance of producing a child with the trait. If both parents are carriers, the probability of having a child with thalassemia major is 25%, while 50% of offspring are expected to be carriers. These inheritance patterns highlight the importance of partner screening to reduce the risk of homozygous thalassemia major births.

Identification of sickle cell trait during the antenatal period is similarly important, as it enables appropriate counseling regarding preventive options and potential complications in affected offspring [9].

In the present study, β-thalassemia trait was the most prevalent abnormal hemoglobin variant, accounting for 54.73% of cases, followed by sickle cell trait at 33.97%. Hb D Punjab (5.03%) and heterozygous Hb E (2.52%) were observed less frequently. Rare variants—including Hb Q India, Hb J India, Hb D Iran/Deer Lodge, and hereditary persistence of fetal hemoglobin (HPFH)—were detected only occasionally.

Similar observations have been documented in antenatal screening studies conducted in Ahmedabad [10] and Jaipur [13], where β-thalassemia trait predominated among hemoglobinopathies. Research from Surat [11] also reported a considerable proportion of sickle cell trait, comparable to the 33.97% observed in the current study. Conversely, investigations from Kolkata [12] have identified a higher frequency of Hb E trait, likely reflecting regional genetic diversity. Multicenter antenatal screening programs across India have estimated β- thalassemia carrier frequencies between 2–8% in the general population, with the prevalence of sickle cell trait varying according to geographic and ethnic distribution [14].

The detection of uncommon variants such as Hb Q India, Hb J India, and Hb D Iran in this study is consistent with isolated reports in Indian literature and underscores the effectiveness of high- performance liquid chromatography (HPLC) in comprehensive antenatal screening programs [15]. With respect to anemia severity, severe anemia (<7 g/dL) was most frequently observed, affecting 69 cases (43.4%). Moderate anemia (7–10 g/dL) was noted in 64 cases (40.25%), while mild anemia (10–13 g/dL) was present in 26 cases (16.35%).

Comparable findings have been reported from Western India. A study from Ahmedabad [16] noted that most antenatal women with β-thalassemia trait and other hemoglobinopathies exhibited moderate to severe anemia. Similarly, research from Surat [17] found that carriers commonly presented with moderate anemia, with fewer cases classified as mild. In contrast, a study from Jaipur [18] documented that the majority of β-thalassemia carriers had mild anemia, and severe cases were relatively uncommon. The higher proportion of severe anemia in the present study may be attributable to coexisting factors such as iron deficiency, inadequate nutritional status, delayed antenatal care registration, or the presence of compound hemoglobinopathies.

These observations reinforce the necessity for thorough antenatal assessment, including clear differentiation between nutritional anemia and anemia secondary to hemoglobinopathies, as the clinical management, counseling, and preventive strategies differ considerably between these conditions [19,20].

CONCLUSION

The present study demonstrates that β-thalassemia trait and sickle cell trait are the most prevalent hemoglobinopathies among antenatal women in the Gujarat region**,** while other variants such as Hb D Punjab, Hb E, and rare hemoglobinopathies occur at lower frequencies. A substantial proportion of affected women presented with moderate to severe anemia, highlighting the clinical significance of these inherited disorders.

Routine antenatal screening**,** particularly in the first trimester, is essential for early detection of hemoglobinopathies. This facilitates genetic counseling, timely intervention, and informed reproductive planning**,** thereby reducing the risk of severe hemoglobin disorders in offspring. Additionally, comprehensive evaluation can distinguish nutritional anemia from hemoglobinopathy-related anemia**,** enabling appropriate management strategies.

Overall, the findings underscore the importance of population-based antenatal screening programs in regions with a high prevalence of inherited hemoglobin disorders to improve maternal and fetal outcomes.

CONFLICT OF INTEREST

The authors declare **no conflicts of interest** related to this study.

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