Original Article

Prevalence and Spectrum of Hemoglobinopathies Among Antenatal Women in Gujarat: An HPLC-Based Screening Study at a Tertiary Care Center

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Abstract

Background: Hemoglobinopathies pose a significant public health challenge in India. Antenatal screening is vital for early detection and informed reproductive choices. While traditional complete blood count (CBC) methods lack specificity, High-Performance Liquid Chromatography (HPLC) offers a precise diagnostic tool. This study evaluated HPLC's effectiveness in antenatal females at a tertiary care center in Gujarat.

Methods: This retrospective, observational study analyzed data from 484 pregnant women who underwent HPLC screening during their antenatal period at a tertiary civil hospital in Ahmedabad, Gujarat. Inclusion criteria ensured complete demographic, hematological (CBC), and interpretable HPLC results. Data were analyzed using descriptive statistics. Ethical approval with a waiver of informed consent was obtained.

Result: Of 484 antenatal women screened, 86 (17.8%) were diagnosed with a hemoglobinopathy. Beta-thalassemia minor was most common (n=62, 72% of positive cases), followed by sickle cell trait (n=14, 16%). Less common findings included HbD Punjab (n=5, 6%), HbE trait (n=2, 3%), and single cases of HbE homozygous, SCD+BT, and delta-beta thalassemia (1% each). Hematological analysis showed elevated HbA2 and microcytosis in beta-thalassemia minor, distinct variant percentages (e.g., HbS 27.82% in sickle cell trait), and varying red cell indices.

Conclusion: This study revealed a high prevalence of hemoglobinopathies (17.8%) among antenatal women in Ahmedabad, Gujarat, predominantly beta-thalassemia minor and sickle cell trait. HPLC proved crucial for accurate diagnosis of these diverse variants. The significant carrier burden underscores the urgent need for comprehensive antenatal screening, genetic counseling, and prenatal diagnostic options to reduce severe hemoglobinopathy incidence in India.

Keywords: HPLC; Beta-Thalassemia Minor; Sickle Cell Trait; HbD Punjab; HbE Trait

Introduction

Hemoglobinopathies are a major group of inherited single-gene disorders that cause chronic anemia, organ damage, and significant morbidity. Their burden is especially high in countries like India, where genetic diversity and regional consanguinity contribute to a higher prevalence. Beta-thalassemia trait (BTT) affects about 3–4% of the Indian population, while sickle cell trait (SCT) can vary widely, reaching over 40% in some tribal groups. The antenatal period provides an important opportunity for screening these disorders. Identifying carriers during pregnancy helps detect at-risk couples,

who otherwise have a 25% chance in each pregnancy of having an affected child. Early detection enables timely genetic counseling, consideration of prenatal diagnostic options (CVS or amniocentesis), and preparation for appropriate post-natal management. Traditional markers such as low MCV and MCH are nonspecific and can overlap with iron deficiency anemia, making confirmatory tests essential. High-Performance Liquid Chromatography (HPLC) has become the preferred method due to its accuracy in quantifying hemoglobin fractions and detecting variants such as HbA2, HbF, HbS, HbE, and HbD. At a tertiary care center with a diverse referral population, implementing HPLC-based screening for pregnant women can substantially improve early carrier detection, enhance genetic counseling, and reduce the birth of children with severe hemoglobinopathies. This study evaluates the effectiveness of HPLC screening among antenatal females at our center and its role in improving overall management and preventive strategies for hemoglobinopathies.

Methods

Aims

To assess the role and effectiveness of High-Performance Liquid Chromatography (HPLC) as a screening tool for detecting hemoglobinopathies in antenatal females attending a tertiary care center in Gujarat, and to evaluate its contribution to timely genetic counseling and prenatal management.

Objectives

To determine the prevalence of various hemoglobinopathies among antenatal females who underwent HPLC screening at the tertiary care center in Gujarat. To highlight the significance of a systematic HPLC-based antenatal screening program in a tertiary care setting for the prevention and improved management of severe hemoglobinopathies. To analyze the correlation between the types of hemoglobinopathies identified by HPLC and the hematological parameters (such as hemoglobin levels, MCV, and MCH) of the antenatal females. To provide recommendations for optimizing antenatal hemoglobinopathy screening strategies within tertiary care settings based on the study findings.

Methodology

Study Design

Study Type: Retrospective, Observational Study. Study Period: Data collection was done for the mother who had visited between January 2024 to August 2024. Study Setting: Medical records department, Antenatal Clinic (ANC) records, and Hematology Laboratory records of a tertiary care center Civil hospital, Asarwa, Ahmedabad, Gujarat.

Study Population & Data Sources

Data Sources: Electronic Medical Records (EMR) or physical patient files from the Antenatal Clinic. Hematology Laboratory registers/database specifically for HPLC results.

Inclusion Criteria

Records of pregnant women who attended the Antenatal Clinic during the defined study period. Records indicating that an HPLC screening for hemoglobinopathies was performed during their antenatal period. Availability of complete demographic information, hematological parameters (CBC with RBC indices), and interpretable HPLC results in the records.

Exclusion Criteria

Records with incomplete or missing essential data required for the study objectives (e.g., no HPLC result, missing key CBC parameters, illegible entries). Records of pregnant women with documented recent blood transfusions (within 3 months prior to HPLC testing), as this can confound intrinsic hemoglobin patterns. Duplicate records for the same patient.

Sample Size

Total 484 medical record of the antenatal mother visited at the Civil hospital; Ahmedabad was collected in specific time period.

Diagnostic criteria

$HbA_2 \geq 3.5\%$ = Diagnostic for β-thalassemia trait, HbS $>50\%$ with no HbA $\rightarrow$ SCD (SS or Sβ^0), HbS $\sim$35–45% with HbA present $\rightarrow$ Sickle trait (AS).

Data Collection (Retrospective Review)

Age, unique hospital ID (for internal tracking, not for analysis), reported community/ethnicity (if routinely collected and documented), residential area (to infer urban/rural and potentially regional prevalence). Hemoglobin (Hb), Mean Corpuscular Volume (MCV), Mean Corpuscular Hemoglobin (MCH), Mean Corpuscular Hemoglobin Concentration (MCHC), Red Cell Distribution Width (RDW), Red Blood Cell (RBC) count. Documented percentages of HbA, HbA2, HbF, and any identified abnormal hemoglobin variants (e.g., HbS, HbE, HbD, etc.), along with their respective percentages.

Data Management and Statistical Analysis

All extracted data were entered into a secure, password-protected electronic database (e.g., Microsoft Excel, SPSS). All statistical analyses were performed using appropriate statistical software (e.g., SPSS).

Results

Total 484 antenatal mother had been visited and screened retrospectively. Out of 484 antenatal women screened, 86 female ($\approx$17.8%) were diagnosed with some form of hemoglobinopathy as shown in table 1. The most common was B Thal minor, accounting for 71% of positive cases (n=61 women). SCT was identified in 16% (n=14 women).

Less frequent findings included HbD Punjab (6%), HbE trait (3%), HbE homozygous (HbE homo), SCD with B Thal, delta-beta thalassemia, and atypical hemoglobinopathy, each representing 1% of positive cases. If we are considering the percentage of the disease in the screened population that those were 18.81%, 2.89%, 1.03%, 0.41%, 0.20%, 0.20%, 0.20%, respectively.

Table 1: Frequency of hemoglobinopathies. A total of 484 antenatal women were screened.

Hemoglobinopathy Type	n	% of Positive Cases	Prevalence (%)
B Thal Minor	62	72	12.81
SCT	14	16	2.89
HbD Punjab	5	6	1.03
HbE trait	2	3	0.42
HbE Homo	1	1	0.21
SCD with B Thal	1	1	0.21
Delta B Thal	1	1	0.21
Total	86	100%	

Table no (2) shows various hematological and HPLC parameters across different hemoglobinopathies identified in antenatal women. In cases of B Thal minor (n=62), elevated HbA2 levels (mean 5.24%) were observed along with microcytosis (mean MCV 62.66 fL) and increased RDW (22.17%), while HbF was mildly raised (1.13%) and HbA0 was 83.69% on average.

SCT (n=14) cases showed HbA0 at 60.89%, HbA2 at 2.75%, and HbF at 1.37%, with slightly higher MCV (66.46 fL) compared to B Thal minor. HbD Punjab (n=5) was characterized by HbA0 of 43.46%, HbA2 of 2%, and HbF of 0.6%, with a near-normal MCV of 78.77 fL.

In HbE trait (n=2), HbA2 was markedly elevated (34.15%), with HbA0 of 57.55% and minimal HbF (0.35%). The single case of HbE homozygous showed very high HbA2 (71%) and HbF (10%), with HbA0 reduced to 8.2%. The SCD + B Thal (n=1) case demonstrated significant HbF elevation (11.6%) and low HbA0 (1.6%).

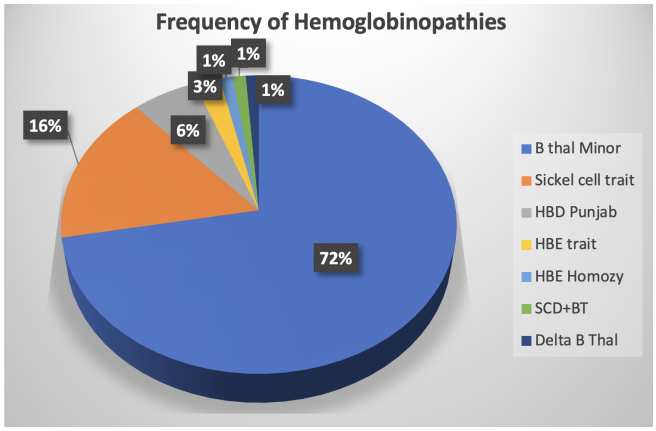

Figure 1: Frequency of hemoglobinopathies.

Lastly, in the rare case of homozygous HbD + B Thal (n=1), HbF was markedly increased (96%) with negligible HbA0 (0.2%). Across all groups, hemoglobin levels were reduced, with the lowest seen in SCD + B Thal (7.9 g/dL), and red cell indices varied according to the specific hemoglobinopathy.

Table 2: Correlation between different hemoglobinopathies and hematological parameter.

	B Thal Minor (n=62)	SCT (n=14)	HbD Punjab (n=5)	HbE Trait (n=2)	HbE Homo (n=1)	SCD with B Thal (n=1)	Delta B Thal (n=1)
HbA2	5.24	2.75	2	34.15	71	7.9	
HbF	1.13	1.37	0.6	0.35	10	11.6	96
HbA0	83.69	60.89	43.46	57.55	8.2	1.6	0.2
Hb	8.34	8.51	8.77	8.7	9.2	7.9	12.7
MCV	62.66	66.46	78.77	64.7	71.7	74.4	67.1
MCHC	31.56	31.57	31.87	33.2	34.6	31.7	32.9
Rbc	4.27	4.03	3.55	4.06	3.69	3.35	5.75
RDWcv	22.17	22.22	19.5	18	18.8	24.4	22.3
HbS	-	27.82	-	-	-	78.23	-

Discussion

The present study aimed to determine the prevalence and spectrum of hemoglobinopathies among antenatal women attending a tertiary care hospital in Ahmedabad, Gujarat. Using high-performance liquid chromatography (HPLC), we identified hemoglobinopathies in 17.8% of screened women (86 out of 484 cases), highlighting a significant carrier burden in this population.

The most common hemoglobinopathy detected was BTT, observed in 61 women, accounting for a prevalence of 12.8% in our study. This prevalence is notably higher than the national average carrier rate of 3–4% reported across India and higher than earlier reports from Western India. For example, Harwani et al. showed the prevalence of B Thal was 3.57% while, Ahuja et al. reported a prevalence of 5.08% in antenatal women in Ahmedabad and Singh et al. documented 6.25% in Pune. Our higher prevalence could reflect regional clustering, referral bias at a tertiary center. As in previous studies, BTT cases consistently showed microcytosis (mean MCV 62.66 fL), hypochromia (mean MCHC 31.56 g/dL), and elevated HbA2 (mean 5.24%), confirming the diagnostic value of combining RBC indices with HPLC for definitive diagnosis [4, 5, 6].

SCT (HbAS) was the second most prevalent hemoglobinopathy in our study, identified in 14 women (2.9% prevalence). This figure is somewhat lower than the 4.03% reported by Ahuja et al. in a similar population and 3.75% by Singh et al., though Gujarat lies within India's sickle cell belt [5, 6]. While Our study results for the prevalence of SCT (2.89%) were consistent with the study done by Harwani et al [4]. The variation could stem from differences in the ethnic composition of the screened women or changing prevalence patterns over time. We also detected one case of SCD in combination with B Thal (SCD + BTT), emphasizing the need for vigilant screening to identify women at risk of bearing children with major hemoglobinopathies.

Less common variants included HbD Punjab trait (5 cases; 1%), which is typically more frequent in Northwestern India but occasionally appears in Gujarat, reflecting the region's genetic diversity. Additionally, rare findings such as HbE trait (2 cases, 0.4%), HbE homozygous (1 case), B Thal (1 case), and homozygous HbD with B Thal (1 case) further underscore the heterogeneous hemoglobinopathy profile encountered at tertiary centers serving diverse populations [6].

Our data support the role of RBC indices—particularly MCV and MCH—as useful initial screening tools, especially for BTT, where consistent microcytosis and elevated RBC count can help differentiate thalassemia trait from iron deficiency anemia (IDA). However, as expected, definitive identification of hemoglobin variants required HPLC, which not only confirmed BTT via HbA2 quantification but also precisely characterized structural variants like HbS, HbE, and HbD. These findings align with prior reports emphasizing that index-based screening, while useful, should be supplemented with HPLC for accurate diagnosis.

The substantial prevalence of hemoglobinopathy carriers (BTT and HbAS together representing over 15% of cases) highlights the public health importance of antenatal screening programs. Early identification of carrier women enables timely partner testing and genetic counseling. This is crucial to identify at-risk couples who face a 25% chance of having a child affected by severe disorders like thalassemia major or sickle cell disease. The study reinforces existing national and international recommendations advocating widespread screening, genetic counseling, and prenatal diagnostic options to reduce the burden of serious hemoglobinopathies in India.

Conclusion

This study demonstrated a high prevalence of hemoglobinopathies among antenatal women at a tertiary care center in Ahmedabad, with 17.8% identified as carriers—predominantly beta-thalassemia minor (72%) and sickle cell trait (16%). The findings highlight the effectiveness of HPLC as a reliable screening tool and emphasize the need for routine antenatal screening, timely genetic counseling, and accessible prenatal diagnostic services to reduce the burden of severe hemoglobinopathies in the community.

Limitations

The study is limited by the quality, completeness, and accuracy of data recorded in routine clinical practice, which was not originally intended for research. Missing or inconsistently recorded information (e.g., detailed genetic counseling content, reasons for declining partner screening/prenatal diagnosis) is an inherent limitation. The study population is drawn from patients who presented to a tertiary care center and underwent HPLC screening, which may not be representative of the entire antenatal population in Gujarat (e.g., sicker patients, those referred for specific reasons, or those from higher socioeconomic strata might be overrepresented).

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Competing Interests: Nil

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