Research Article

Prevalence of Hemoglobinopathies in a Tertiary Care Hospital

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ABSTRACT

Background: Hemoglobinopathies are among the most common inherited monogenic disorders worldwide, with significant prevalence in India, particularly in tribal and rural populations. Gujarat reports a high burden of both β-thalassemia and sickle cell disorders.

Aim: To evaluate the spectrum and clinico-hematological profile of hemoglobinopathies in a tertiary care hospital.

Methods: A retrospective and prospective descriptive study was conducted over four years (Jan 2021 to Dec 2024) at a tertiary hospital in Gujarat. A total of 100 confirmed cases of hemoglobinopathies were analyzed. Complete blood count and peripheral smear were performed in a NABL accredited laboratory, and data were analyzed.

Results: The most common hemoglobinopathy was β-thalassemia trait (44%), followed by sickle cell disorders (27%) and sickle cell trait (24%). Rare variants included HbE+S (1%) and HbD-Punjab trait (1 %). Two cases of homozygous β-thalassemia were identified in children under 10 years. Clinical features included pallor, fever, and jaundice, particularly in sickle disorders and thalassemia major. Most cases belonged to OBC, SC, and ST communities.

Conclusion: Hemoglobinopathies remain a significant public health challenge in Gujarat, with β-thalassemia trait being the most prevalent. Hemoglobin electrophoresis is a cost-effective screening tool that should be integrated into antenatal, premarital, and adolescent health programs. Genetic counselling and molecular follow-up are essential for long-term disease reduction.

Keywords: Hemoglobinopathies, Thalassemia, Sickle cell syndrome, Electrophoresis

1. INTRODUCTION

Hemoglobinopathies are the most prevalent single-gene disorders worldwide and represent a major cause of chronic anemia and related morbidity in many regions, including India. Globally, several million carriers and thousands of affected newborns are identified each year, imposing significant clinical and public health burdens. [1] In India, the combined burden of β-thalassemia and sickle-cell disorders is substantial, with large regional and community-specific variation driven by endogamy, consanguinity and historical population structure. [2-5]

Gujarat is recognized as an important regional focus for hemoglobinopathies, where distinct community and tribal pockets show high carrier frequencies of β-thalassemia, HbS and other variants such as HbD-Punjab and HbE.[6-9] These local patterns produce a heterogeneous clinical spectrum that ranges from asymptomatic carriers to transfusion-dependent thalassemia major and severe sickle-cell disease, contributing to repeated hospitalizations, iron-overload complications and impaired quality of life in affected families.[10-13]

The present study idea originated from routine clinical observations at our tertiary care centre: a considerable number of patients particularly young adults and antenatal women presented with persistent microcytic anemia despite adequate iron indices, yet were not being systematically evaluated for hemoglobinopathies. This diagnostic gap,

together with limited availability of confirmatory testing at peripheral centres, suggested under-recognition of carriers and affected individuals and motivated a structured institutional audit of the local hemoglobinopathy spectrum. [14-17]

Recent diagnostic and programmatic evidence supports the need for such institution-level data. Large laboratory surveys and regional screening programs have shown that capillary electrophoresis and HPLC markedly improve variant detection and that targeted community screening (school, antenatal and premarital) coupled with counselling reduces the number of affected births when effectively implemented.[18-23] At the same time, practical analyses from semi-urban and rural settings emphasize that hemoglobin electrophoresis remains a pragmatic, cost-effective first-line test where molecular methods are unavailable.[24-27]

Justification for aims and objectives : Accurate knowledge of the local prevalence and clinico-hematological profile of hemoglobinopathies is essential to: (a) identify asymptomatic carriers who would benefit from counselling; (b) detect affected individuals early to optimize management and reduce complications; (c) support targeted antenatal and premarital screening programs in high-risk communities; and (d) provide an evidence base for scaling up molecular diagnostics and health policy planning in the region.

Purpose of the study : To determine the prevalence, demographic distribution and clinico-hematological spectrum of hemoglobinopathies among patients attending a tertiary care hospital using hemoglobin electrophoresis, and to evaluate how these data can be used to strengthen genetic counselling and targeted preventive strategies.

2. MATERIALS & METHODS

2.1 Materials

Method :

Study design : Hospital based observational study with both retrospective and prospective study to evaluate the clinico-hematological spectrum of hemoglobinopathies. Dual design was chosen because retrospective cases helped to obtain adequate number of cases and prospective cases to allow better clinical laboratory correlation. No methodological differences in two phases. This study involved only confirmed cases of hemoglobinopathies and did not involve population level screening. Therefore, a denominator of total screened patients was not applicable.

Study setting and duration : The study was carried out in the Shree Krishna Hospital, Karamsad, a tertiary care hospital over a period of four years of which two years retrospective (Jan 2021 to Dec 2022) and two years prospective (Jan 2023 to Dec 2024). Hemoglobin analyses were performed using Hemoglobin Capillary Electrophoresis or High Performance Liquid Chromatography at a referral laboratory. Quality control procedures as per laboratory standards were followed. Reference ranges provided by the reporting laboratory were used.

Inclusion criteria: Patients of any age and sex with clinical features of hemolytic anemia from various clinical departments of Shree Krishna Hospital as well as referred patient of hemoglobinopathies (including trait and disease) and confirmed by Hemoglobin Capillary electrophoresis or High-PerformanceLiquid Chromatography.

Exclusion criteria : Cases with incomplete clinical or laboratory data and cases other than hemoglobinopathies.

Sampling method and sample size : Consecutive sampling was employed. All patients fulfilling the inclusion criteria were included. A total of 100 cases were included until the desired sample size achieved.

Data collection : For retrospective cases, data were retrieved from Laboratory Information System, Hospital based records and patient files. For prospective cases, data were collected through clinical history, physical examination and laboratory investigations.

Hemoglobin variants were confirmed using a single validated diagnostic modality. Repeat testing or molecular studies were not routinely performed due to resource constraints.

Statistical analysis : Data were entered into a master chart and analyzed using descriptive statistics. Continuous variables were expressed as mean and standard deviation.

Table 1 Age, Sex and Prevalence of Various Hemoglobinopathies Distribution

Age Group	Sex	Beta Thal Trait	Sickle Trait	Sickle Cell Syndrome	Homozygous β-Thal	HbD Punjab	HbE+Sickle	Total
0–10	Female	1	1	2	1	0	0	5
	Male	3	0	1	0	1	0	5
11–20	Female	3	3	5	0	0	0	11
	Male	0	1	4	0	0	0	5
21–30	Female	11	8	6	0	0	1	26
	Male	6	3	6	0	1	0	16
31–40	Female	3	5	1	1	0	0	10
	Male	3	1	2	0	0	0	6
41–50	Female	2	1	0	0	0	0	3
	Male	1	0	0	0	0	0	1
51–60	Female	2	0	0	0	0	0	2
	Male	3	1	0	0	0	0	4
61–70	Female	3	0	0	0	0	0	3
	Male	3	0	0	0	0	0	3

Table : 2 Hb electrophoresis mean Findings of Various Hemoglobinopathies

Diagnosis	HbA (%)	HbA2 (%)	HbS (%)	HbF (%)	HbE (%)	HbD (%)
Beta Thalassemia Trait	94.77 ± 0.78	6.05 ± 7.26	0.0 ± 0.0	0.95 ± 0.63	–	–
Double heterozygous (HbE+Sickle)	–	4.5 ± NA	64.9 ± NA	3.9 ± NA	26.7 ± NA	–
HbD Punjab Trait	52.4 ± 4.67	3.05 ± 0.07	–	9.8 ± NA	–	39.65 ± 2.19
Homozygous β-Thalassemia	8.55 ± 3.18	2.65 ± 0.78	–	89.05 ± 3.61	–	–
Sickle Cell Syndrome	20.25 ± 17.80	2.83 ± 1.15	68.7 ± 14.86	19.59 ± 7.74	–	–
Sickle Cell Trait	66.95 ± 6.62	2.90 ± 0.44	31.3 ± 9.75	2.33 ± 1.18	26.7 ± NA	–

Table : 3 Clinical symptoms vs Various Hemoglobinopathies

Symptom	Beta Thal Trait	HbE+ Sickle	HbD Punjab	Homo β-Thal	Sickle Cell Syndrome	Sickle Trait
Fever	1	0	0	0	7	2
Joint Pain	0	1	0	0	11	0
Jaundice	0	1	0	0	6	1
Weakness	0	0	1	1	9	0
Pallor	0	0	1	1	9	1
Splenomegaly	0	0	0	1	0	0

3. RESULTS & DISCUSSION

Results

Out of 100 cases, β-thalassemia trait was most common (44%), followed by sickle cell syndrome (27%) and sickle cell trait (24%). Rare variants included HbE+ HbS (1%) and HbD-Punjab trait (2%). Two cases of homozygous β-thalassemia occurred in children below 10 years. Most patients belonged to OBC, SC, and ST groups. Clinically, pallor, fever, and jaundice were the common features in sickling disorders and thalassemia major.

Discussion

The present study demonstrates that β-thalassemia trait is the most prevalent hemoglobinopathy in Gujarat, with prevalence figures comparable to earlier studies from the region, thereby reaffirming its significant regional burden. Minor variations are likely due to differences in study population, sample size, and screening strategies. Overall, the findings confirm the high burden of hemoglobinopathies in Gujarat, consistent with existing literature. High prevalence among OBC, SC, and ST communities aligns with community-specific carrier rates described in Indian literature. The study reinforces the role of hemoglobin electrophoresis as a cost-effective diagnostic tool and emphasizes the importance of community-based screening, genetic counselling, and prevention strategies to reduce disease burden.

4. CONCLUSION

β-thalassemia trait and sickle cell disorders are highly prevalent in Gujarat. Early detection using hemoglobin electrophoresis, combined with genetic counselling and public health programs, is essential for effective control and prevention of hemoglobinopathies.

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Conflict of Interest

We wish to confirm that there are no known conflicts of interest associated with this publication and there has been no significant financial support for this work that could have influenced its outcome.

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Ethical Information

Approved by Institutional Ethics Committee letter no IEC/BU/147/Faculty/27/271/2023, conducted in accordance with ethical standards with maintained patient confidentiality.

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