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Demographic Profiling of Haemoglobinopathies-A Rzetrospective Study at Thalassemia and Sickle Cell Society, Telangana

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Abstract

Hemoglobinopathies are common inherited blood disorders, affecting 7% of the global population. In India, β-thalassemia, sickle cell disease, and other variants show varied prevalence across regions and ethnic groups due to genetic diversity and consanguinity. This study analyzed the demographic profile and prevalence of hemoglobinopathies among 4,336 patients registered at the Thalassemia and Sickle Cell Society (TSCS), Hyderabad. Of the 4,336 cases registered from 1998–2025, the highest were in 2023 (9.85%), with minimal contributions from 1998–2002 (each <1.2%). Blood samples were evaluated using CBC and HPLC to assess RBC indices and hemoglobin fractions. Among 4,336 individuals, 57.7% were male and 42.2% female, with mean age of 16.24 years. Most patients were aged 10–20 years (39.58%) and about 39.4% reported consanguineous marriages. Most patients were from Telangana (71.2%) and Andhra Pradesh (23.8%). Hinduism was the predominant religion (76.4%), with Lambadi, Madiga, and Mala being the most represented castes. Beta-thalassemia major was the most prevalent disorder (59.96%), followed by sickle cell disease (22.3%) and sickle beta-thalassemia (9.1%). Other less common hemoglobinopathies included E beta-thalassemia, thalassemia intermedia, delta beta thalassemia and rare variants HbH disease and HPFH. These findings underscore the significant public health burden of hemoglobinopathies in Telangana. The high prevalence of β-thalassemia, highlights the urgent need for targeted genetic screening, counseling, and community-based awareness programs. A coordinated approach involving early detection, multidisciplinary care, and advanced therapies is essential to reduce disease impact. Institutions like TSCS exemplify a successful model of integrated care, combining diagnostics, treatment, and patient support.

Keywords: thalassemia; sickle cell anemia; HPLC; demographic; thalassemia and sickle cell society

1. Introduction

Hemoglobinopathies are the most common monogenic and inherited autosomal recessive disorders of the structure and synthesis of hemoglobin. Around 7% of the global population carries an abnormal hemoglobin gene. Approximately 80% of newborns with these disorders, including thalassemia, are born each year in low-income or middle-income nations. Sub-Saharan Africa, South Asia, and the Middle East bear the highest burden of these disorders, while migration is increasing their prevalence in high-income countries. Due to increased migration patterns, these genetic blood disorders have spread to most parts of Europe, the Americas, Australia, and other countries worldwide. This global movement of people has contributed to the wider distribution of

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haemoglobinopathies, highlighting the need for increased awareness, diagnosis, and management of this condition in diverse populations (Weatherall, 2011; Modell, 2008).

Haemoglobinopathies including β-thalassemia, α-thalassemia, sickle cell disease (SCD), hemoglobin E (HbE) disorder, and other rare hemoglobin variants such as HbD, HbC, and HbS-β thalassemia represent a major public health challenge in India.β-thalassemia is one of the most prevalent haemoglobinopathies in India, with carrier frequencies ranging from 1%–12% across different regions. The highest prevalence was reported to be 3%–9% in Punjab, Haryana, and Delhi (Colah et al., 2015; Balgir, 2006; Verma et al., 2011). According to Patel et al. (2020), carrier frequencies between 2% and 8% have also been observed in communities such as the Sindhi, Gujarati, and Maharashtrian populations, where the disease burden remains notably high. While α-thalassemia is generally less severe in clinical presentation, its carrier rate is estimated at 1% to 3%, with higher concentrations in the northeastern and southern states (Mohanty et al., 2014). α-Thalassemia, though less clinically severe, has a carrier frequency of 1%–3% and is more common in the northeastern and southern states (Mohanty et al., 2014). Sickle cell disease is more commonly seen in central and eastern parts of India, especially among tribal groups like the Gond, Baiga, and Bharia, where the sickle cell trait (SCT) occurs in 10% to 40% of the population (Mohanty et al., 2013). Hemoglobin E (HbE) is another variant that is particularly widespread in northeastern India, with certain ethnic groups in Assam and West Bengal showing prevalence rates of up to 50% (Das et al., 2004). Additionally, rare hemoglobin variants such as HbD Punjab and HbQ India have been identified in northern regions (Sukesh et al., 2024). HbH disease, which results from deletions in the α-thalassemia gene, has also been reported in specific tribal populations and parts of the northeast (Colah et al., 2010).

These disorders result from genetic mutations affecting the production and function of hemoglobin, leading to anemia, haemolysis, and severe complications if left untreated. Due to the country's vast genetic diversity, consanguinity, and endogamous practices, the prevalence of haemoglobinopathies varies across different regions and communities with significantly higher prevalence among specific ethnic and tribal groups (Colah et al., 2015; Verma et al., 2011). Studies have shown that premarital and antenatal screenings are effective in reducing the birth prevalence of affected children (Ahuja et al., 2024). However, these programs remain underutilized, particularly in rural and tribal communities. The geographical diversity in prevalence rates underscores the need for a comprehensive epidemiological assessment of haemoglobinopathies across different Indian regions. Addressing this need requires robust diagnostic capabilities, and High-Performance Liquid Chromatography (HPLC) has emerged as a pivotal tool in the accurate diagnosis of these conditions, facilitating early detection and improved patient management.

In response to the challenges posed by these disorders, the Thalassemia and Sickle Cell Society (TSCS) (Reg No. 5359), a Hyderabad-based NGO was established in 1998 at Hyderabad, Telangana, India. Society aims to promote appropriate treatment and quality life for thalassemia and sickle cell anemia affected patients by providing crucial support, including consultation, blood, blood transfusions, counseling, diagnostic, research and prenatal diagnostic services allowing for early detection and management. The present study aimed to determine the demographic distribution and prevalence of various haemoglobinopathies among 4,336 patients registered at the Thalassemia and Sickle Cell Society (TSCS) in Hyderabad, Telangana.

2. Materials and Methods

2.1. Study Population

This study included 4336 patients registered at the Thalassemia and Sickle Cell Society (TSCS) outpatient clinic in Hyderabad, Telangana India. The data collected includes from 1998 till April 2025. Following the acquisition of informed consent from parents or attendants, demographic data were collected using a standardized proforma including gender, date of birth, age at onset of symptoms, religion, caste.

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2.2. Sample Collection

Blood samples were collected in EDTA vacutainers and analyzed using an automated blood cell counter for complete blood count (CBC) (H560, ERBA, Hematology Analyzer) to determine red blood cell (RBC) indices, including mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), red blood cell count, and red cell distribution width (RDW).

2.3. Sample Analysis

High-performance liquid chromatography (HPLC) (Variant II, Bio-Rad, USA) was employed to quantify hemoglobin fractions, including HbA, HbA2, and HbF.

Samples with normal RBC indices (MCV > 80 fl, MCH > 27 pg, normal RBC count and RDW) and normal HbA2 levels (<3.5%) and HbF levels were classified as normal. Samples with microcytic and hypochromic indices (MCV < 80 fl, MCH < 27 pg, increased RBC count and normal RDW) and elevated HbA2 levels (>3.5%) were identified as potentially having β-thalassemia trait. Samples with HbA2, levels below 3.5% and normal HbF levels and MCV < 80 fl, were diagnosed as alpha thalassemia. Samples with absence of HbA with elevated HbF (>90%) were diagnosed as β-thalassemia major.

The presence of Hb variants (HbS, HbE, HbD, etc.) was determined by specific retention times and peak characteristics in the HPLC chromatogram. Sickle cell trait was determined with HbS 30–40% and normal HbA2 whereas Sickle cell disease with HbS>60% and elevated HbF.

Samples with normal HbA2 (<3.5%) and high HbF (10-25%) with variable RBC indices were noted to be δβ-thalassemia and HPFH (Hereditary Persistence of Fetal Hemoglobin) carriers.

Patients with abnormal hemoglobin patterns underwent molecular testing for further confirmation in the molecular genetics' laboratory at the society. Detailed molecular diagnostic data and treatment outcomes from this study will be presented in a forthcoming journal publication.

2.4. Statistical Analysis

Data analysis was performed using the Statistical Package for the Social Sciences (SPSS) version 20. Descriptive statistics like frequency distributions were used to analyze demographic data.

3. Results

3.1. Year wise Distribution of Patients Registration:

A total of 4,336 cases were registered from the year 1998 to 2025. The highest proportion of cases was registered in 2023, accounting for 9.85%, followed by 2024 with 8.97% and 2022 with 8.12%. The lowest proportions were observed in the earlier years, with 1998–2002 contributing minimally to the overall distribution (each below 1.2%). (Figure 1)

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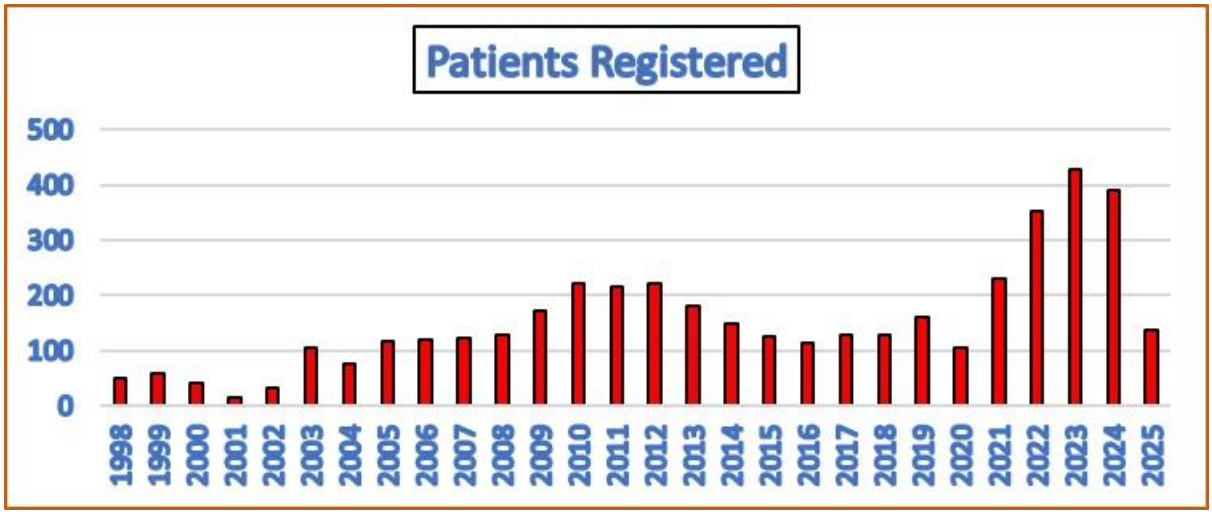

Figure 1. Year wise Distribution of Patients Registration.

Among the 4336 cases registered 50.55% of them are active, while a significant portion were receiving treatment elsewhere or not coming regularly due to travel constraints (40.13%). The death rate among the cases is 6.39%, and 2.93% had successful bone marrow transplants.

3.2. Economic Category Distribution

Economic Category Distribution results show that the majority of registered cases fall under the Below Poverty Line (BPL) category, accounting for 72.92% of the total. Above Poverty Line (APL) patients represent 21.93% while the economic status of 5.14% of cases is unknown (as per Socio-Economic and Caste Census (SECC) 2011).

3.3. State wise Distribution

Figure 2 illustrates State wise distribution of registered patients which revealed that though the majority (71.24%) of the individuals belonged to Telangana (as the society is in Hyderabad), significant representation was also made from Andhra Pradesh (23.82%), and small percentages belonged to other states. Analysis by location revealed that the vast majority of cases were registered in Hyderabad, representing 95.73% of the total. Khammam and Kurnool contributed 2.58% and 1.68% of the cases, respectively.

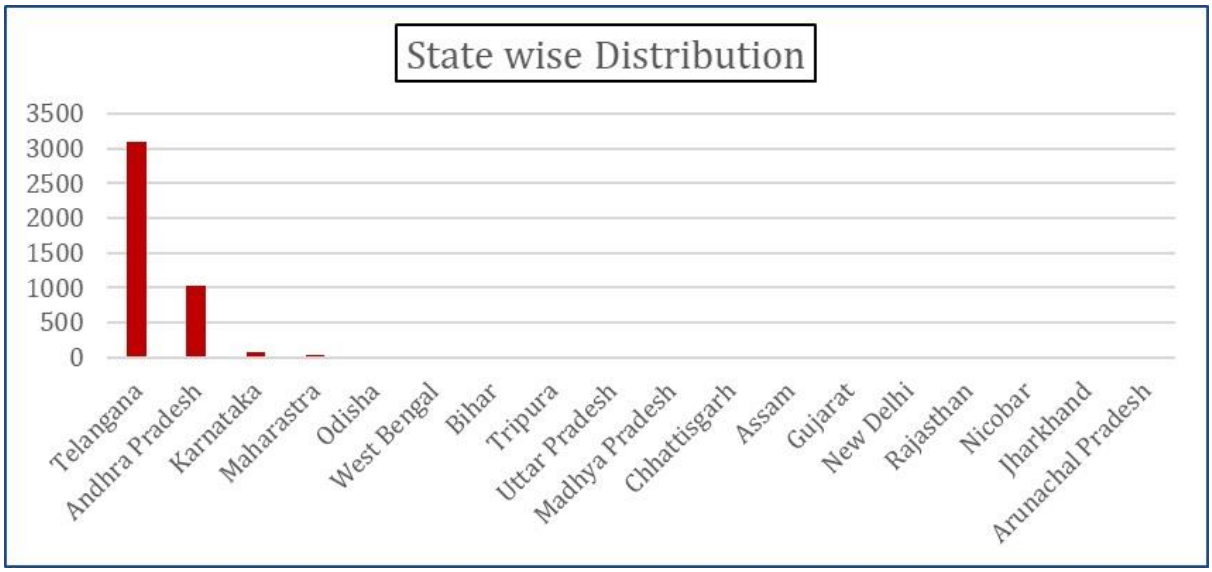

Figure 2. State wise distribution of registered patients.

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3.4. Gender and Age Distribution

Table 1 describes the gender and age distribution of 4,336 individuals.57.7% were males and 42.2% were females. The mean age of the study population was 16.24 years. Age-wise distribution among 4,336 registered patients shows that the majority belonged to the 10–20 years age group, accounting for 39.58% followed by the 20–30 years group at 26.20%. The 0–10 years group also formed a significant proportion, representing 23.18%. In contrast, older age groups contributed relatively fewer cases: 30–40 years accounted for 8.30% while the 40–50 years and above groups each constituted less than 3% of the total, with declining survival rate as the age increases. Only 0.07% were recorded in the 70–80 years age group.

Table 1. Age Group and Gender Distribution.

Age and Gender Distribution					Total	
Age Group	**Females**	**%**	**Males**	**%**	**N**	**%**
0-10	400	39.8	605	60.2	1005	23.18
10-20	750	43.71	966	56.29	1716	39.58
20-30	481	42.34	655	57.66	1136	26.2
30-40	141	39.17	219	60.83	360	8.3
40-50	46	51.11	44	48.89	90	2.08
50-60	13	59.09	9	40.91	22	0.51
60-70	1	25	3	75	4	0.09
70-80	1	33.33	2	66.67	3	0.07
Total	**1833**	**42.27**	**2503**	**57.73**	**4336**	**100**

The gender distribution across age groups shows a consistent male predominance, particularly among younger patients. In the 0–10 and 10–20 years groups, males constituted 60.2% and 56.29% of cases, respectively. Among the 20–30 years group, males constituted 57.66% and 30–40 years 60.83% were males. Female predominance was seen in 40–50 (51.11%) and 50–60 years (59.09%) groups. In the oldest age groups (60–80 years), male proportions remain higher.

3.5. Blood Group Distribution

The distribution of blood groups among the 4,336 cases showed that the most common blood group was O+, observed in 39.41% of patients followed by B+ at 30.28% and A+ at 19.44% which is consistent with the distribution of blood groups in the general population. The AB+ group accounted for 6.48% (n = 281), while negative blood groups were less prevalent, with O-, B-, A-, and AB- together forming only 4.31% of cases. Rare entries included one case of the Bombay blood group (Bom) blood group. (Table 2)

Table 2. Distribution of blood groups among the registered cases.

Blood Group	**n**	**%**

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O+	1710	39.43
B+	1314	30.30
A+	843	19.44
AB+	281	6.48
O-	70	1.61
B-	58	1.34
A-	48	1.11
AB-	11	0.25
Bom	1	0.02
Total	**4336**	**100.00**

3.6. History of Consanguinity

Out of 4,336 cases, **39.44%** (1710) reported a history of consanguinity, while **60.56%** (2626) did not show consanguinity.

3.7. Religion and Caste Wise Distribution

Table 3 illustrates Religion and caste wise distribution of the data which showed that Hinduism is predominant (77.93%), followed by Islam (19.6%) and Christianity (2.3%). Small percentages represented Sikhism, Buddhism, and Jainism also.

Table 3. Religion and caste wise distribution of the registered patients.

Religion	**Frequency**	**Percent**
Hinduism	3379	77.93
Islam	850	19.60
Christianity	99	2.28
Sikhism	5	0.12
Buddhism	2	0.05
Jainism	1	0.02
Total	**4336**	**100.0**
Caste	**Frequency**	**Percent**
Lambadi	566	13.8
Madiga	442	10.8
Mala	411	10

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Shaik	230	5.6
Mohd	219	5.3
Netakani	114	2.8
Kapu	91	2.2
Padma Shali	85	2.1
Syed	78	1.9
Sunni	71	1.7
Boya	54	1.3
Goud	45	1.1
Yadav	43	1
Brahmin	39	1
Mudiraj	39	1
Others	128	3.1
Total	**2655**	**100.0**

Caste could be recorded for 2665 patients only. Among the caste, the Lambadi caste is highly prevalent with 13.8% of the total .10.8% were Madiga caste followed by 10% Mala caste. Shaik (5.6%), Mohd (5.3%), Netakani (2.8%), Kapu (2.2%), Padmashali (2.1%), Syed (1.9%), Sunni (1.7%), Boya (1.3%), Goud (1.1%), Yadav (1%), Brahmin (1%), and Mudiraj (1%) are also observed. The category "Others" constitutes 3.1%.

3.8. Distribution of Hemoglobinopathies

Table 4 describes the distribution of hemoglobinopathies and related conditions. Out of the total 4,336 individuals screened, Thalassemia Major was the most prevalent haemoglobinopathy, observed in 2,600 cases (59.96%). The second most common condition was Sickle Cell Anemia, found in 968 individuals (22.32%), followed by Sickle Beta Thalassemia, representing 396 cases (9.13%). Other haemoglobinopathies included E Beta Thalassemia (2.86%), Thalassemia Intermedia (2.35%), and Sickle D Punjab (0.35%). Several rare variants were also detected, including Delta Beta Thalassemia (0.25%), Alpha Beta Thalassemia (0.28%), Alpha Thalassemia (0.14%), Beta HPFH (0.14%), E Thalassemia (0.09%), Alpha Thalassemia (HbH type) (0.09%), Sickle Alpha Thalassemia and Sickle Delta Thalassemia (0.05% each), Sickle E Thalassemia (0.02%) and Sickle HPFH (0.02% each)

Table 4. Distribution of hemoglobinopathies in the study population.

Haemoglobinopathies	**n**	**%**
Thalassemia Major	2600	59.96
Sickle Cell Anemia	968	22.32
Sickle Beta Thalassemia	396	9.13

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E Beta Thalassemia	124	2.86
Thalassemia Intermedia	102	2.35
Sickle D Punjab	15	0.35
Sickle E Thalassemia	1	0.02
Sickle Alpha Thalassemia	2	0.05
Alpha Thalassemia	6	0.14
Delta Beta Thalassemia	11	0.25
Alpha Beta Thalassemia	12	0.28
E Thalassemia	4	0.09
Sickle HPFH	1	0.02
Sickle Delta Thalassemia	2	0.05
Alpha Thal (HbH)	4	0.09
Beta HPFH	6	0.14
Others	42	0.96
Beta Thalassemia Trait	18	0.42
Sickle Cell Trait	12	0.28
Alpha Thalassemia Trait	2	0.05
Normal	6	0.14
Total	**4336**	**100**

In addition to hemoglobinopathies, other hematological disorders were diagnosed and grouped as others (42 cases) which included Hemolytic Anemia, Fanconi Anemia, Aplastic Anemia, Spherocytosis (Types 1 and 3), Pyruvate Kinase Deficiency, Sideroblastic Anemia, Pure Red Cell Aplasia, and Polycystic Kidney Disease. Additionally, a single case each of vasculitis with autoinflammation was recorded. Additionally, carriers were identified, including Beta Thalassemia Trait (0.42%), Sickle Cell Trait (0.28%), and Alpha Thalassemia Trait (0.05%).

4. Discussion

Thalassemia and sickle cell anemia (SCA) are the most common hemoglobinopathies that are inherited disorders in India with an existence of differences in the types and prevalence due to regional differences. The present study carried out at Thalassemia and Sickle Cell Society (TSCS), Hyderabad is aimed to understand the demographic profile and prevalence of haemoglobinopathies among the registered cases. This study presents retrospective information on 4,336 patients registered from 1998 to 2025, and provides useful information on the epidemiology and clinical profile of hemoglobinopathies and hematologic disorders in our population. Most registrations were relatively recent, with 2023, 2024, and 2022 representing the highest annual caseloads. This increase

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in registration may reflect improved diagnostic capabilities, greater awareness, expanded screening programs, or enhanced referral systems in recent years. In contrast, the early years of the registry (1998–2002) contributed minimally, likely due to lower awareness during that period.

Among the registered patients only 50.55% of them remained active at the time of analysis, 40.13% were taking transfusions elsewhere or not coming regularly due to travel constraints, reflecting challenges in long-term follow-up and continuity of care. The mortality rate of 6.39% highlights the severity of these conditions when not managed appropriately. 2.93% of patients successfully underwent bone marrow transplantation, reflecting progress in curative therapies under the supervision of the TSCS.

Socioeconomic analysis of the cohort revealed that a majority of patients (72.92%) belonged to below Poverty Line (BPL), These findings highlight the need for targeted public health interventions, subsidized treatment programs, and socioeconomic support mechanisms to reduce health disparities and improve outcomes in these groups.

It was observed that 39.58% of the study population is under the age of 10- 20 years, which aligns with the high incidence of inherited conditions like β-thalassemia and sickle cell disease observed in younger populations. Approximately 8,000–10,000 children are born annually with β-thalassemia major in India, a trend that is particularly prevalent in tribal communities and regions like Punjab and Tamil Nadu (Colah et al., 2013; Balgir, 2014; Ghosh et al., 2015).

A male predominance of 57.73% was noted, likely due to greater healthcare-seeking behavior for male children in India, though gender-specific prevalence data for hemoglobinopathies remains inconsistent (Kumar & Gupta, 2017). Additionally, 39.44% of the study population reported consanguineous marriages, which increase the risk of homozygous recessive conditions like β-thalassemia major and sickle cell disease (Vijay et al., 2023; Kumar et al., 2013; 2015; Sukesh, 2024). Consanguinity is prevalent in South India, where awareness and screening are essential to mitigate health risks in these high-risk populations.

The predominance of Rh-positive blood types (O and B) has important implications for blood transfusion support in this high-transfusion-dependent population, especially for conditions like Thalassemia Major. It emphasizes the need for well-equipped blood banks with capacity for rare blood group screening and Rh-negative inventory management which is maintained by the thalassemia society.

The prevalence of hemoglobinopathies is influenced by caste, tribe, and regional factors. In this study, 77.93% of patients were Hindu, with 13.8% from the Lambadi caste followed by Mala (10.8%) and Madiga (10%). Regional studies from Eastern Uttar Pradesh and Maharashtra highlight the prevalence in specific ethnic groups (Sujata et al., 2004; Urade, 2013). Our previous study (VR Rao et al 2020) showed that Sunni (27.2%), Lambadi (20.8%), Madiga (12.5%) Mala (4.5%) and Mudiraj (4.5%) contributed 69.5%, and another 43 groups contributed 30.5%. Endogamous marriage practices within these communities and migration of the communities contribute to higher genetic disorders by limiting genetic diversity and perpetuating the inheritance of harmful alleles (Bittles, 2002; Aggarwal et al., 2017).

β-thalassemia represents one of the most widespread haemoglobinopathies in India, with significant variation in prevalence across different regions and communities. The present findings indicate that beta thalassemia major is the most prevalent condition, accounting for 59.6% of cases which is extremely high compared to other studies as in retrospective study from Southern India, beta-thalassemia major accounted for 10.98% of cases (Kumar et al., 2022). In Jammu and Kashmir, the prevalence of hemoglobinopathies ranges from 3.15% to 36%, with beta-thalassemia major prevalence reported at 1.29% (Bhasin et al., 2006; Shukla et al., 2013). A study from south India reported that beta thalassemia prevalence is 2.3–7.47%, while the β-thalassemia trait ranges from 8.5% to 37.9% (Kulkarni et al., 2014). Studies from Madhya Pradesh, reported a β-thalassemia trait prevalence of 1.4–3.4% (Sharma et al., 2003; Saxena et al., 2012).

The second common condition observed in our study was sickle cell disease, accounting for 968 cases (22.3%). The prevalence of sickle cell disease (SCD) in India varies across regions and

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population groups, with tribal communities showing the highest prevalence rates ranging from 1% to 40% across different communities (Narayanasamy et al., 2024) (Rao et al., 2024). Studies have observed HbS allele was at higher frequency in communities like the Gond, Pradhan, and Kondh (Urade, 2012) (Kaur et al., 1997). There was also increase among scheduled castes and some non-tribal ethnic groups (Jain et al., 2024). A systematic review and meta-analysis reported a pooled prevalence of 1.17% for SCD and 5.9% for sickle cell trait (SCT) in the general population, with higher rates in states like Maharashtra and Gujarat (Rao et al., 2024).

Sickle beta-thalassemia is observed in 9.13% of cases. This disorder poses a considerable health challenge, especially in regions where both sickle cell and beta-thalassemia are found together. In contrast, E beta-thalassemia, seen in around 2.9% of cases, this was more prevalent in the northeastern parts of India (Sharma et al., 2020). Thalassemia intermedia (2.35%) is less common still raises serious concerns because of its unpredictable symptoms and the need for continuous care and long-term management (Kumar et al., 2022).

The presence of rare haemoglobinopathies such as Sickle D Punjab (0.35%), Alpha Beta Thalassemia and Delta Beta Thalassemia (0.25 & 0.28%respectively), and hereditary persistence of fetal hemoglobin (HPFH) with beta-thalassemia mutations (0.1%) indicates vast genetic diversity in Indian populations (Bhasin et al., 2006; Shukla et al., 2013). Additionally, Hemoglobin H (HBH) disease, a rare form of alpha-thalassemia (0.09%), has been reported in a limited number of cases, in tribal and Northeastern populations (Dastidar & Gajra, 2013). Other anemia disorders, including sideroblastic anemia, Fanconi's anemia, and hemolytic anemia observed, emphasize the need for comprehensive genetic screening and management strategies.

In conclusion these findings highlight the substantial public health challenge posed by hemoglobinopathies in Telangana, particularly β-thalassemia major and sickle cell disease. The high prevalence of β-thalassemia major, indicates that there is need for targeted screening, genetic counseling, and community-based awareness initiatives among high-risk groups. Addressing this burden requires a comprehensive, coordinated strategy that includes early detection, multidisciplinary care and innovative therapies. Institutions like the Thalassemia Society play a important role not only in-patient care but also in advanced molecular diagnostics and treatment. Their integrated approach serves as a model for effective management of hemoglobinopathies. Strengthening and replicating such community driven frameworks at regional and national levels will be instrumental in reshaping the future of thalassemia care in India.

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