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Research Article

STATUS OF THALASSEMIA AND SICKLE CELL DISEASE IN NANDURBAR DISTRICT: AN OBSERVATIONAL STUDY ON DISEASE BURDEN, CLINICAL PATTERNS, AND MANAGEMENT

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Abstract

Thalassemia and sickle cell disease (SCD) are among the most prevalent inherited hemoglobin disorders globally and represent major public health concerns, particularly in low- and middle-income countries. India represents one of the major global burden zones for hemoglobinopathies, with particularly high prevalence among tribal and socioeconomically vulnerable populations. Maharashtra, especially tribal-dominated districts such as Nandurbar, carries a disproportionately high burden of sickle cell disease and thalassemia due to genetic, environmental, and socio-cultural factors. The present observational study aimed to assess the status, disease burden, clinical manifestations, diagnostic approaches, and treatment patterns of thalassemia and sickle cell disease in Nandurbar district. Data were collected from documented patient observations involving 28 cases. Parameters analyzed included demographic distribution, symptoms, blood group patterns, treatment duration, diagnostic testing, treatment modalities, and medication utilization. The study findings indicated a higher disease occurrence among individuals aged 18–40 years, with male predominance. Common clinical manifestations included jaundice, fatigue, weakness, brittle bones, and dark urine. High-performance liquid chromatography (HPLC) and blood investigations were commonly used diagnostic tools, while major treatment modalities included blood transfusion, chelation therapy, and emerging gene therapy approaches. The findings highlight the substantial burden of inherited hemoglobin disorders in Nandurbar and emphasize the urgent need for strengthened screening, genetic counselling, early diagnosis, and comprehensive disease management programs.

Keywords: Thalassemia, sickle cell disease, hemoglobinopathy, Nandurbar district, Maharashtra, HPLC, blood transfusion, genetic disorders

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Introduction

Hemoglobinopathies constitute a major class of inherited blood disorders characterized by abnormalities in the structure, synthesis, or function of hemoglobin. Hemoglobin is the oxygen-carrying metalloprotein found within red blood cells and is essential for systemic oxygen transport and cellular metabolism. Genetic abnormalities affecting hemoglobin synthesis result in chronic anemia, tissue hypoxia, haemolysis, organ dysfunction, and reduced quality of life (1).

Among hemoglobinopathies, thalassemia and sickle cell disease are the most clinically significant inherited disorders worldwide. Although both diseases involve defective hemoglobin biology and chronic hematological complications, they differ substantially in their genetic mechanisms, pathophysiology, clinical progression, and treatment requirements.

Thalassemia is a quantitative hemoglobin disorder caused by defective synthesis of alpha or beta globin chains, leading to ineffective erythropoiesis, chronic haemolysis, and varying degrees of anemia. Depending on the mutation severity, thalassemia may manifest as mild carrier states or severe transfusion-dependent disease. Beta-thalassemia major represents one of the most severe forms, requiring lifelong transfusion support and iron chelation therapy (2).

Sickle cell disease is a structural hemoglobin disorder caused by a point mutation in the beta-globin gene resulting in the production of hemoglobin S (HbS). Under deoxygenated conditions, HbS polymerizes, causing erythrocyte deformation into rigid sickle-shaped cells. These abnormal cells obstruct microvasculature, trigger haemolysis, induce inflammatory injury, and contribute to recurrent Vaso-occlusive crises and progressive organ damage (3).

Globally, sickle cell disease affects hundreds of thousands of newborns annually, with the highest disease burden concentrated in sub-Saharan Africa, the Middle East, Mediterranean countries, and South Asia. The persistence of these genetic disorders in malaria-endemic regions has been attributed to balanced polymorphism, wherein carrier states provide partial protection against severe malaria infection (4).

India represents one of the most significant global burden zones for hemoglobinopathies, especially among tribal and rural populations. Carrier frequencies vary widely depending on ethnicity, geography, and community practices. Maharashtra has emerged as one of the most affected Indian states, particularly in tribal-dominated districts such as Nandurbar, Palghar, and Thane.

Nandurbar district is particularly important because of its substantial tribal population, socioeconomic challenges, limited healthcare infrastructure, and increased prevalence of inherited hemoglobin disorders. State registry data indicate a significant concentration of sickle cell disease cases in this district, emphasizing its public health relevance.

Given this epidemiological burden, the present study was undertaken to assess the local status of thalassemia and sickle cell disease in Nandurbar district, with emphasis on demographic characteristics, disease presentation, diagnosis, and management.

Materials and Methods

Study Design

The present study was designed as an observational descriptive study aimed at evaluating the disease burden and management status of inherited hemoglobin disorders, specifically thalassemia and sickle cell disease, in Nandurbar district, Maharashtra.

Study Area

The study was conducted in Nandurbar district, Maharashtra, India, a predominantly tribal region recognized as a high-prevalence zone for sickle cell disease and related hemoglobinopathies.

Study Population

Data were collected from documented patient observations involving individuals diagnosed with thalassemia and related inherited hematological disorders. A total of **28 patient cases** were included in the observational analysis.

Data Collection Parameters

Clinical and observational data were analyzed based on the following parameters:

- age distribution
- gender distribution
- symptom profile
- diagnostic investigations
- treatment duration
- treatment modalities
- medication usage
- blood group distribution

The objective was to identify local disease trends and management practices.

Diagnostic Assessment

Diagnostic evaluation was based on routinely employed hematological investigations.

Blood testing served as a primary assessment tool for identifying anemia and hematological abnormalities. High-performance liquid chromatography (HPLC) was used as a confirmatory diagnostic technique for hemoglobinopathy identification due to its sensitivity in detecting abnormal hemoglobin variants.

Rapid solubility screening methods were also referenced as preliminary detection approaches for sickle hemoglobin screening.

Evaluated Clinical Variables

Clinical observations included commonly reported manifestations associated with inherited hemoglobin disorders, including:

- jaundice
- fatigue
- weakness
- brittle bones
- dark-coloured urine

These symptoms were assessed as indicators of disease burden and progression.

Treatment Variables

Treatment patterns were evaluated by documenting therapeutic interventions received by patients, including:

- blood transfusion therapy
- iron chelation therapy
- supportive drug treatment
- advanced gene therapy approaches

Drug utilization observations included deferiprone as a major chelation medication.

Statistical Analysis

Data were analyzed descriptively using frequency-based observational summaries. Numerical counts were used to characterize distributions of symptoms, treatments, diagnostic investigations, and demographic variables.

No inferential statistical analysis was performed due to the observational descriptive nature of the study.

Results

The observational assessment of inherited hemoglobin disorders in Nandurbar district revealed important demographic, clinical, diagnostic, and therapeutic trends. A total of **28 documented patient cases** were analyzed as part of the study.

Demographic analysis showed a higher prevalence among males (**16 cases**) compared to females (**12 cases**). Age distribution indicated that the highest disease burden was observed in the **18–40-year age group**, suggesting that young adults represent a significantly affected population segment. Paediatric cases were also substantial, with **12 children identified**, emphasizing the hereditary and early-onset nature of these disorders.

Symptom analysis demonstrated that **jaundice** was the most frequently observed clinical manifestation, reported in five patients. Other documented symptoms included fatigue and weakness, brittle bones, pale or yellowish skin, and dark-coloured urine, all consistent with chronic haemolysis and anemia-related complications.

Diagnostic evaluation revealed that **high-performance liquid chromatography (HPLC)** was the most frequently utilized diagnostic investigation, performed in nine cases, followed by conventional blood investigations in four cases. This finding reflects the importance of HPLC as a reliable diagnostic modality for identifying hemoglobin variants and confirming hemoglobinopathies.

Treatment analysis demonstrated that disease management was largely dependent on conventional supportive therapies. **Gene therapy was documented in eight cases**, chelation therapy in five cases, and blood transfusion therapy in four cases. Among pharmacological interventions, **deferiprone** was the most documented medication, recorded in seven patients, highlighting the importance of iron chelation in transfusion-dependent disease management.

Blood group distribution indicated the presence of affected individuals across multiple blood groups, with type O showing relatively greater representation compared to other groups in the observed dataset.

Discussion

The present observational study provides insight into the burden and management patterns of inherited hemoglobin disorders in Nandurbar district, a region recognized for its high prevalence of sickle cell disease and related hemoglobinopathies. The findings reinforce the significant public health burden posed by these genetic disorders, particularly in tribal and socioeconomically vulnerable populations.

Hemoglobinopathies remain among the most common inherited genetic disorders worldwide, with sickle cell disease and thalassemia accounting for substantial morbidity and mortality in developing countries (1). India, particularly central and western tribal regions, carries a disproportionate share of this burden due to genetic clustering, endogamous marriage practices, inadequate screening, and limited healthcare accessibility (2).

The predominance of cases in the 18–40-year age group observed in this study may reflect improved survival among affected individuals due to advances in supportive care, alongside delayed diagnosis in certain patients. The substantial paediatric representation further confirms the hereditary nature of these disorders and highlights the importance of newborn screening and early intervention programs.

Male predominance in the present dataset may reflect healthcare access patterns, sociocultural treatment-seeking behavior, or sampling variability rather than a true biological predisposition, since both thalassemia and sickle cell disease follow autosomal recessive inheritance patterns affecting both sexes equally (3).

Jaundice emerged as the most common clinical manifestation in the observed cohort, consistent with chronic haemolytic processes characteristic of hemoglobinopathies. Ongoing red blood cell destruction results in increased bilirubin production, contributing to jaundice, dark urine, and hepatobiliary complications (4). Fatigue and weakness observed in patients are classical manifestations of chronic anemia resulting from impaired oxygen transport and reduced hemoglobin functionality.

The high utilization of HPLC observed in this study is consistent with current diagnostic practice. HPLC remains one of the most reliable methods for hemoglobinopathy detection, particularly for differentiating hemoglobin variants and confirming carrier or disease states (5). Early diagnostic confirmation is essential not only for treatment initiation but also for family screening and genetic counselling.

The predominance of supportive treatment modalities such as blood transfusion and iron chelation reflects current standard management practices. Regular blood transfusions remain essential for severe thalassemia management, whereas iron chelation therapy is necessary to prevent transfusion-related hemosiderosis and organ damage (6). The documented use of deferiprone aligns with established therapeutic practice for iron overload management.

Gene therapy documentation in the dataset reflects awareness of evolving therapeutic strategies. Recent advances in gene editing and hematopoietic stem cell–based interventions offer promising curative potential for inherited hemoglobin disorders, although accessibility remains limited in most low-resource settings (7).

The high burden of hemoglobinopathies in Nandurbar is likely influenced by multiple interacting factors including genetic clustering, tribal population density, healthcare access limitations, inadequate public awareness, delayed diagnosis, and historical malaria-related gene persistence. These findings underscore the urgent need for integrated regional public health strategies.

Screening programs, premarital counselling, antenatal testing, newborn screening, community education, and improved access to specialist haematology services remain critical interventions for reducing disease burden and improving patient outcomes (8).

Limitations of the Study

Despite the useful observational insights generated by the present study, certain limitations should be acknowledged. The sample size was relatively small, involving only 28 documented cases, which may limit the generalizability of the findings to the broader Nandurbar population. The study employed a descriptive observational design without inferential statistical analysis, limiting the ability to establish associations or causal relationships between demographic and clinical variables. Detailed laboratory parameters such as hemoglobin concentration, ferritin levels, reticulocyte counts, and molecular genetic confirmation were not consistently available for all patients. The dataset also lacked standardized longitudinal follow-up information, making assessment of long-term outcomes, treatment effectiveness, and disease progression difficult. Furthermore, differentiation between thalassemia and sickle cell disease cases within certain treatment summaries was not always explicitly defined, which may affect interpretation. Future studies should incorporate larger sample populations, standardized diagnostic classification, molecular confirmation, and longitudinal clinical monitoring to strengthen epidemiological and therapeutic conclusions.

Conclusion

The present study highlights the significant burden of inherited hemoglobin disorders in Nandurbar district, particularly among tribal and vulnerable populations. The findings demonstrate that thalassemia and sickle cell disease continue to pose major public health

challenges due to chronic morbidity, diagnostic complexity, and long-term treatment requirements.

Young adults represented the most affected age group, while common clinical manifestations included jaundice, fatigue, weakness, and anemia-related complications. HPLC emerged as a key diagnostic tool, while treatment largely depended on blood transfusion, iron chelation, and supportive management.

These findings emphasize the urgent need for stronger preventive and management strategies, including population screening, early diagnosis, genetic counselling, premarital carrier detection, newborn screening, and improved healthcare accessibility in high-burden districts such as Nandurbar.

With continued advancements in diagnostics, therapeutics, and public health interventions, improved survival and quality of life for affected individuals can be achieved.

References

1. Piel FB, Steinberg MH, Rees DC. Sickle cell disease. N Engl J Med. 2017;376(16):1561–1573.
2. Colah R, Gorakshakar A, Nadkarni A. Global burden and distribution of hemoglobinopathies in India. Indian J Med Res. 2010;132:507–521.
3. Weatherall DJ, Clegg JB. The Thalassaemia Syndromes. 4th ed. Blackwell Science; 2001.
4. Hoffbrand AV, Moss PAH. Essential Haematology. 7th ed. Wiley Blackwell; 2016.
5. Bain BJ. Haemoglobinopathy diagnosis by HPLC and electrophoretic methods. Blood Rev. 2011;25(5):205–213.
6. Cappellini MD, Porter JB, Viprakasit V, Taher AT. Guidelines for thalassemia management. Thalassemia Int Fed. 2021.
7. Frangoul H, Altshuler D, Cappellini MD, et al. CRISPR-based gene therapy for sickle cell disease and beta-thalassemia. N Engl J Med. 2021;384:252–260.
8. Ware RE, de Montalembert M, Tshilolo L, Abboud MR. Sickle cell disease. Lancet. 2017;390:311–323.