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RESEARCH ARTICLE

Haemoglobin Structure, Sickle Cell Anemia, Thalassemia, and Public Health Challenges in Kenya: A Comprehensive Review

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ABSTRACT

Hemoglobinopathies such as sickle cell disease (SCD) and thalassemia are among the most common inherited disorders worldwide. They impose heavy health and socioeconomic burdens, especially in sub-Saharan Africa where malaria endemicity drives their distribution. In Kenya, prevalence is highest in Western and coastal regions, but screening is limited, health infrastructure is weak, and cultural myths persist. This review synthesized evidence on hemoglobin structure and genetics, distribution of hemoglobinopathies in Kenya particularly diagnostic approaches, management challenges, and strategic interventions in Western Kenya. A narrative review of peer-reviewed and gray literature (2015–2025) was conducted using major databases and institutional repositories. Search terms included "sickle cell anemia," "thalassemia," "hemoglobinopathies," "Kenya," and "sub-Saharan Africa." Hemoglobinopathies cluster in malaria-endemic regions, particularly western Kenya, a region predominated by the Luo, Luhya, Teso, and Abagusii communities. Sickle cell disease is most common in these populations, while thalassemia remains less studied but increasingly reported. Diagnostic methods are largely limited to electrophoresis and high-performance liquid chromatography (HPLC) in a few centers. Key challenges include underfunding, limited political commitment, a shortage of hematologists, and cultural misconceptions that delay care. Hemoglobinopathies represent a serious but under-addressed public health threat in Kenya. Interventions such as newborn screening, genetic counseling, community education, specialist training, and investment in advanced diagnostics and therapies including gene therapy are urgently needed to reduce morbidity and mortality.

Keywords: *Hemoglobinopathies, Sickle Cell Disease, Diagnostics, Kenya*

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INTRODUCTION

Hemoglobinopathies pose a major global health burden, affecting millions worldwide through conditions such as sickle cell disease (SCD) and thalassemia. About 7% of the world's population carries a hemoglobin disorder trait, with over 300,000 infants born annually with severe disease, mostly SCD and thalassemia (World Health Organization [WHO], 2021). These disorders are both medical and socioeconomic problems, causing recurrent illness, growth delays, early mortality, and financial strain on families and health systems (Makani et al, 2018). Outcomes vary greatly between high-income and low and middle-income countries. In developed countries, newborn screening, prophylaxis, hydroxyurea therapy, and curative approaches like bone marrow transplantation have dramatically improved survival, with life expectancy exceeding 50 years in some cases (Hankins et al., 2014). However, over two-thirds of global cases occur in sub-Saharan Africa, and 50–80% of children with SCD die before age five due to delayed diagnosis, limited services, and lack of national programs (Makani et al., 2017; Arishi et al., 2021).

The high prevalence of hemoglobinopathies in Africa is closely linked to malaria. The sickle cell trait (HbAS) confers partial protection against severe Plasmodium falciparum malaria, explaining its persistence in endemic regions (Fofana et al., 2023). Thalassemias, though less documented, are frequently underdiagnosed and misclassified as nutritional anemia, especially alpha and beta thalassemia traits that contribute to microcytic anemia (Macharia et al., 2023). Across sub-Saharan Africa, hemoglobinopathies significantly contribute to pediatric admissions and mortality. In Tanzania, SCD is a major cause of death in children under five (Makani, 2018). Similar patterns are observed in Nigeria, Ghana, and Uganda (Ndeezi et al., 2016). In Kenya, their burden remains under-prioritized compared to diseases like HIV, TB, and malaria. National data is fragmented, with most studies concentrated in Kisumu and the Lake Victoria basin (Uyoga et al., 2019; Kosiyo et al., 2020). Moreover, in Kenya, hemoglobinopathies are concentrated in malaria-endemic regions, especially around Lake Victoria and the coast. The Luo and Luhya communities show the highest sickle trait frequencies, while data from groups like the Kisii and Kuria remain limited (Mutua et al., 2022). Health system gaps such as lack of newborn screening, limited laboratory capacity outside urban centers, and a severe shortage of hematologists worsen the problem (Makani et al.,2017). There are also aspects of sociocultural beliefs associating SCD with curses, infidelity or delayed medical intervention, as families often consult traditional healers first.

Despite their impact, hemoglobinopathies remain marginalized in Kenya's health agenda. There are no dedicated government programs or funding streams, and research investment is minimal. This is alarming given projections that by 2050, sub-Saharan Africa will account for over 80% of global sickle cell births. Without action, the growing burden risks overwhelming fragile health systems. (Global Burden of Disease Study 2021, 2023). This review consolidated evidence on hemoglobin structure, SCD, and thalassemia, emphasizing their distribution in Western Kenya, diagnostic approaches, and challenges in management. It proposes strategic, evidence-based interventions to guide clinicians, researchers, and policymakers in reducing their long-term health and socioeconomic impact.

METHODS

Study Design

A narrative literature review design was used to provide a comprehensive overview of hemoglobinopathies in Kenya, focusing on Western Kenya where these disorders are most prevalent. Unlike systematic reviews that use rigid criteria, the narrative approach allowed integration of diverse sources including epidemiological studies, molecular research, sociocultural analyses, and health system reports. This was essential because Kenyan research on hemoglobinopathies is fragmented and often includes unpublished hospital reports, theses, and policy documents. The design also facilitated synthesis across different timeframes and regions, highlighting knowledge gaps and contextual healthcare challenges

Data Sources and Databases

Literature was searched in PubMed, Scopus, Web of Science, and Google Scholar, complemented by manual searches in Kenyan university repositories, Ministry of Health reports, and WHO documents. Search terms included: "sickle cell anemia," "thalassemia," "hemoglobinopathies," "Kenya," "Western Kenya," "sub-Saharan Africa," and "hemoglobin variants." Both controlled vocabulary and free-text keywords were used. Backward and forward citation tracking helped identify additional relevant studies. This multi-source strategy minimized publication bias and ensured coverage of both peer-reviewed and gray literature.

Inclusion and Exclusion Criteria

The inclusion criteria for this review encompassed studies focusing on hemoglobin disorders such as SCD, thalassemia, and other

structural hemoglobin variants. Particular emphasis was placed on research involving African populations, with priority given to data from Kenya and regional comparative analyses. Eligible studies were those published between January 2015 and March 2025 and included peer-reviewed journal articles, academic theses, official government reports, and documents produced by reputable non-governmental or international agencies. Conversely, the exclusion criteria ruled out case reports or small case series involving fewer than five patients, studies published in languages other than English without available translations, and papers lacking primary or secondary data relevant to Kenya. These criteria ensured that the review remained comprehensive while maintaining a clear focus on the most pertinent and contextually relevant evidence.

Study Selection Process

The search yielded 47 records. After removing duplicates, 25 were screened by title and abstract by two reviewers. Seventeen full-texts were then assessed using the inclusion/exclusion criteria, focusing on prevalence data, diagnostic methods, and clinical outcomes. Using multiple literature types enabled triangulation of findings, enhancing reliability.

Data Extraction

Data extraction was conducted using a structured template designed to organize information into key thematic domains, including hemoglobin structure and genetics, prevalence and distribution in Kenya, diagnostic methods, clinical burden and mortality, health system and sociocultural challenges, and recommended interventions. Two reviewers independently extracted the data to ensure accuracy and consistency, followed by a thorough cross-checking process to resolve any discrepancies. This systematic approach ensured that the extracted information was comprehensive, reliable, and aligned with the study objectives.

Data Analysis

Prevalence and diagnostic data were synthesized both thematically and narratively, highlighting disparities across regions and healthcare facilities. Policy documents were examined alongside empirical findings to identify critical gaps, such as the absence of a nationwide newborn screening program despite WHO recommendations. Evidence synthesis principles were applied to maintain transparency, rigor, and clarity in the analysis.

Ethical Considerations

This review used only secondary data and required data and required no ethical approval. Ethical scholarship was upheld through proper attribution (APA 7th edition), transparent documentation of methods, and acknowledgment of open-access sources. The ethical implications of disparities, stigmatization, and policy neglect were recognized in the discussion.

RESULTS

Hemoglobin Structure

Normal adult hemoglobin consists of three major types: HbA1 ($\alpha_2\beta_2$), comprising 95–98% of total hemoglobin; HbA2 ($\alpha_2\delta_2$), 1.5–3.5%; and fetal hemoglobin (HbF, $\alpha_2\gamma_2$), which persists below 2% in adults. Mutations or deletions in globin genes lead to structural variants (e.g., HbS, HbC, HbE, HbD) or quantitative abnormalities (thalassemia), both of which impair oxygen transport and cause clinical disease (Patel & Sharma, 2024).

Sickle Cell Disease

Sickle cell disease results from a β-globin gene point mutation (GAG→GTG), substituting valine for glutamic acid at position 6. This alters hemoglobin behavior, promoting polymerization under low oxygen and deforming red cells into rigid “sickle” shapes. This leads to hemolysis, vaso-occlusive crises, and chronic complications (Dufu et al., 2016, Patel & Sharma, 2024). Homozygotes (HbSS) present with severe disease; heterozygotes (HbAS) are usually asymptomatic but epidemiologically significant.

Thalassemia

Thalassemias are inherited blood disorders characterized by reduced or absent synthesis of one or more globin chains, which form part of the hemoglobin molecule. The imbalance in globin chain production leads to defective hemoglobin formation, resulting in varying degrees of anemia and other related complications (Macharia, 2023). Alpha thalassemia arises from deletions or mutations affecting the α-globin genes located on chromosome 16. The clinical severity depends on the number of affected genes, ranging from silent carriers with minimal clinical impact to severe forms such as hydrops fetalis, which is typically incompatible with life (Baird et al., 2022). Beta thalassemia, on the other hand, results from mutations in the β-globin gene on chromosome 11. Its severity varies from the mild, asymptomatic β-thalassemia trait to the severe, transfusion-dependent β-thalassemia major (Fibach & Rachmilewitz, 2017). Both alpha and beta thalassemias lead to ineffective erythropoiesis, increased red blood cell destruction (hemolysis), and chronic anemia, often necessitating long-term medical management and supportive care (Baird et al., 2022).

Diagnostic Approaches in Kenya

Diagnosis of hemoglobinopathies in most African settings is primarily limited to hemoglobin electrophoresis, which detects major variants such as HbS and HbF. High-performance liquid chromatography (HPLC) is available only in a few tertiary or research centers, where it provides more accurate quantification of hemoglobin fractions. Molecular diagnostic methods, such as polymerase chain reaction (PCR), remain largely confined to research laboratories due to high costs, limited technical expertise, and inadequate infrastructure. As a result, many individuals with hemoglobin disorders remain undiagnosed or are identified at advanced stages of the disease (Makani et al., 2020; Mutua et al., 2022; WHO, 2021).

Geographical Distribution and Environmental Influences

The geographical distribution of hemoglobinopathies in Kenya closely corresponds to areas of high malaria endemicity. The highest prevalence rates are found in Western Kenya particularly in the Lake Victoria basin, encompassing the former Nyanza and Western provinces, as well as Trans-Nzoia County and along the coastal region, all of which are malaria-endemic zones (Mutua, 2022; Kosiyo et al., 2020). Western Kenya is characterized by significant ecological diversity, consisting of four major zones: the highlands, lowlands, upper midlands, and the upper Rift Valley (Matsushita et al., 2019). These zones differ in altitude, rainfall, and temperature; factors that influence malaria transmission intensity and, consequently, the prevalence of hemoglobinopathies.

The highlands, including areas such as Kakamega and Bungoma, experience moderate malaria transmission, which has been further altered by environmental modifications such as swamp drainage (Omukunda et al., 2012). In contrast, the lowlands surrounding Lake Victoria, such as Kisumu and Homa Bay, exhibit persistently high malaria transmission due to their hot, humid climate and abundant swampy environments (Ojowi et al., 2001). The upper midlands, including Kisii, Nyamira, and Migori counties, show intermediate prevalence rates influenced by ecological variation and mixed ethnic composition (Alwy & Schech, 2004). Trans-Nzoia County serves as a transitional zone with moderate malaria transmission and a cosmopolitan population structure. Overall, geography exerts a strong influence on malaria heterogeneity, which in turn shapes the distribution of sickle cell anemia and related hemoglobinopathies. The lowland regions consistently record the highest frequencies of sickle cell traits, reflecting the evolutionary relationship between malaria exposure and the protective advantage conferred by the heterozygous HbAS genotype (Matsushita et al., 2019).

Socio-Demographic Characteristics

Sickle cell anemia prevalence is highest among Luo and Luhya populations (14–22%), with lower but significant levels among the Abagusii (Uyoga et al., 2019; Njoroge et al., 2020). Gender prevalence is similar, but the disease burden falls disproportionately on children under five, who experience high mortality from severe anemia, infection, and stroke.

Challenges in Addressing Hemoglobinopathies in Western Kenya

Addressing hemoglobinopathies in Western Kenya faces numerous challenges spanning health, social, and policy dimensions. One of the major barriers is inadequate funding, as few programs specifically target SCD despite its increasing burden. High program implementation costs and the prioritization of other health concerns, particularly infectious diseases, limit the availability of grants and constrain both research and intervention efforts.

Without significant investment, the projected burden by 2050 could overwhelm the region's already strained health systems. Compounding this is a lack of political commitment; national policies on premarital screening, newborn screening, and community sensitization remain underdeveloped, leaving SCD largely neglected in public health planning (Mutua, et al., 2022). A critical shortage of human resources further worsens the situation, with Kenya having fewer than two dozen hematologists to serve the entire population. In contrast, countries like Tanzania have demonstrated improved outcomes through dedicated specialist training programs (Makani et al., 2018). Access to advanced therapies such as gene therapy remains extremely limited due to prohibitive costs, inadequate infrastructure, and a lack of technical expertise. At the community level, myths and misconceptions about SCD such as beliefs that it is a curse, punishment, or consequence of infidelity delay appropriate health-seeking behavior, often leading families to seek help from traditional healers before turning to formal healthcare systems.

Social practices also contribute to the persistence of SCD. Intra-ethnic marriages, which are common in high-prevalence regions, increase the likelihood of sickle cell trait (SCT) couples producing affected children.Additionally, cultural practices such as widow inheritance can perpetuate consanguinity and elevate genetic risks (Sawaimul et al., 2018; Mutua, et al., 2022). The neglect of hemoglobinopathies, compounded by

outdated cultural norms and minimal government engagement, has resulted in a lack of sustainable, long-term strategies for disease control and prevention in Western Kenya.

DISCUSSION

This review highlights that hemoglobinopathies particularly SCD and thalassemia represent a growing but under-recognized public health burden in Kenya, especially in malaria-endemic regions such as Western Kenya and the coast. The geographic overlap between high malaria transmission and sickle trait prevalence illustrates classic natural selection dynamics: the HbAS genotype offers partial protection against severe Plasmodium falciparum malaria, sustaining high gene frequencies over generations (Kosiyo et al., 2020).

While prevalence patterns are well described, diagnostic and treatment gaps remain profound. Most Kenyan facilities rely on hemoglobin electrophoresis, and few offer HPLC or molecular testing, limiting early detection. Many affected individuals are diagnosed late, often after recurrent crises or severe anemia. The lack of national newborn screening contrasts sharply with countries like the USA, UK, and Ghana, where early diagnosis has transformed outcomes (Makani et al., 2018; Arishi et al., 2021). Health system constraints including limited funding, lack of hematologists, and absence of national strategies perpetuate morbidity and mortality. With fewer than two dozen hematologists nationally, most patients rely on general clinicians with minimal training in hemoglobinopathy management. This mirrors findings in other African countries before specialist training programs were scaled up (Makani et al., 2018).

Sociocultural factors compound these structural gaps. Myths associating SCD with curses or infidelity discourage formal health-seeking. Marriage practices, such as intra-ethnic unions and widow inheritance, maintain high-risk pairings (Sawaimul et al., 2018; Mutua et al.,2022). Addressing these requires culturally sensitive community engagement, genetic counseling, and integration of religious and community leaders into public health strategies.

The policy neglect of hemoglobinopathies in Kenya is striking given their projected growth. Global estimates suggest that by 2050, sub-Saharan Africa will account for 80% of sickle cell births, yet Kenyan policies remain fragmented (Shamira et al., 2025). Lessons from Tanzania show that targeted training, community screening, and political commitment can improve outcomes. Tanzania's program, which began with pilot newborn screening, expanded through partnerships and now provides hydroxyurea and specialist care (Makani et al., 2018). Kenya lacks comparable initiatives. Public health opportunities exist. Premarital and antenatal screening can identify carriers early, while newborn screening enables timely prophylaxis (e.g., penicillin), vaccination, and hydroxyurea initiation. Integration with existing maternal and child health platforms, including HIV PMTCT and immunization programs, could provide cost-effective entry points (Dilli et al., 2024). Additionally, research investment in molecular diagnostics and gene therapy capacity is needed to align with global advances.

The socioeconomic impact is also substantial. Families face frequent hospitalizations, lost income, and stigma, while health systems bear recurrent crisis management costs. Without early diagnosis and preventive strategies, the cumulative burden will rise. Integrating hemoglobinopathies into Kenya's Universal Health Coverage and NHIF packages would alleviate financial strain and improve access to essential services. Overall, Kenya stands at a crossroads. Without deliberate policy shifts, the growing hemoglobinopathy burden could overwhelm the health system. With evidence-based interventions screening, early diagnosis, specialist training, and culturally sensitive community engagement Kenya could follow successful models from other countries and avert preventable deaths.

Conclusion

Hemoglobinopathies, particularly sickle cell disease and thalassemias, remain a pressing yet insufficiently addressed public health challenge in Kenya. Concentrated in malaria-endemic areas such as Western Kenya, these conditions contribute significantly to childhood morbidity, hospitalizations, and mortality. Despite global advances in diagnosis and treatment, Kenya continues to face critical constraints in diagnostic capacity, access to curative and preventive interventions, and health system preparedness. The country's limited pool of hematology specialists, combined with inadequate funding and poor policy prioritization, hampers the establishment of effective programs. Compounding these barriers are persistent cultural myths that stigmatize affected families and delay health-seeking.

This review emphasizes the urgent need for Kenya to adopt comprehensive strategies tailored to hemoglobinopathies. Interventions should include structured newborn screening programs, community sensitization to dispel

harmful myths, integration of genetic counseling into reproductive health services, and investment in modern diagnostic and therapeutic technologies. Addressing these issues will require coordinated collaboration among government agencies, academic institutions, civil society organizations, and international partners. If hemoglobinopathies continue to be neglected, Kenya risks facing an overwhelming increase in preventable childhood deaths and long-term socioeconomic strain as the projected burden grows toward 2050.

Recommendations

- Premarital Screening for Sickle Cell Trait in Kenya:The Kenyan government should formulate and implement a premarital screening policy to reinforce early detection of sickle cell trait (SCT) through hemoglobin electrophoresis or high-performance liquid chromatography (HPLC). This policy would mandate voluntary screening for couples before marriage to identify carriers of SCT. In cases where both partners are identified as carriers, structured genetic counseling should be provided to ensure that couples are well informed of the potential risk of having children with sickle cell disease (SCD).Such an approach would enable couples to make marriage decisions with full awareness of the genetic implications for their future children, rather than facing the unexpected realization of having offspring with SCD.
- Newborn and Population Screening: Establish nationwide newborn screening programs for hemoglobinopathies, beginning with county referral hospitals in high-prevalence regions and gradually expanding to cover all maternity units. Such programs would enable early diagnosis and timely interventions.
- Community Education: Implement culturally sensitive awareness campaigns at the community level. These should be designed to challenge harmful myths, reduce stigma, and encourage families to seek medical care early.
- Capacity Building: Expand training opportunities for hematologists, pediatricians, and laboratory scientists. Establish regional centers of excellence for sickle cell care, equipped with advanced diagnostics such as HPLC and molecular genotyping.
- Research and Data Systems: Develop a national patient registry for hemoglobinopathies to monitor trends, guide resource allocation, and evaluate program outcomes. Support further research into cost-effective interventions and long-term outcomes.
- Therapeutic Access: Ensure equitable access to hydroxyurea by subsidizing its cost and improving supply chain reliability. Strengthen blood transfusion safety and prepare to participate in gene therapy trials as global technologies advance.
- Policy Integration: Incorporate hemoglobinopathies into Kenya's broader non-communicable disease strategies. Dedicated funding streams should be established to support screening, treatment, and research initiatives.

Conflict of Interest

The authors declare no conflict of interest.

Author Contributions

MNM conceptualized the review and performed literature search drafted the manuscript.
BM also wrote the original manuscript in addition to critical reviews. Both authors proofread and approved the final version.

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