

Prevalence of Thalassemia in Children in Africa: A Systematic Review and Meta-Analysis

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Abstract

Thalassemia is a blood disorder caused by numerous inherited mutations in the globin gene. This meta-analysis aimed to provide an update on the prevalence of thalassemia in Africa between 2007 and 2025. A systematic literature search was conducted across the PubMed, AJOL, and Google Scholar databases to identify studies published between 2007 and

2025, using specific inclusion and exclusion criteria. Quality assessment was performed using the Mixed Methods Appraisal Tool (MMAT) for prevalence studies. The heterogeneity of the included studies was assessed using the I^2 and Q statistics. Funnel plots and Egger tests were performed to determine publication bias in this meta-analysis. The pooled 95% confidence interval (95% CI) prevalence of thalassemia across studies was determined using a generic random-effects inverse-variance method. Eleven studies involving 26025 subjects were included, and the prevalence rates were pooled using random-effects models due to high observed heterogeneity ($I^2 > 75\%$, p -value < 0.05). The overall prevalence of thalassemia was 13.4% (95% CI 8.5-19.1%, $I^2 = 99.1\%$). Subgroup analyses showed that the pooled prevalence of thalassemia was 14% (95% CI, 0-33%; $I^2 = 98.5\%$) in neonates, 22% (95% CI, 11-33%; $I^2 = 98.8\%$) in patients ≤ 5 years of age and 6% (95% CI, 3-9%; $I^2 = 82.6\%$) in patients over 5 years of age. The prevalence of alpha-thalassemia carriers was 18% (95% CI, 7-28%; $I^2 = 99.2\%$) and beta-thalassemia 9% (95% CI, 6-12%; $I^2 = 98.6\%$). This meta-analysis suggests a high prevalence of thalassemia in certain African countries. These data help design thalassemia screening programs and improve disease management.

Keywords: Prevalence, Thalassemia, Systematic review, Meta-analysis, Africa

Introduction

Thalassemia is an inherited disorder characterized by a deficit in haemoglobin synthesis due to mutations in the α -, β -, and δ -globin genes, leading to inefficient erythropoiesis. The World Health Organization (WHO) has classified thalassemia as a significant global health problem. The most common forms are α - and β -thalassemia (Tuo et al., 2024).

Worldwide, thalassemia affects around 4.4 in 10,000 live births. Transmission is autosomal recessive, regardless of sex (Smith Yolanda, 2022). Approximately 5 to 7% of the world's population carries a thalassemia gene, meaning that more than 400 million people are carriers of some form of thalassemia (mostly minor), and around 60,000 children are born each year with a significant form of thalassemia that requires treatment (UNFPA, 2022).

The prevalence of thalassemia is highest in India, the Mediterranean basin, the Middle East, Southeast Asia, and Africa. Alpha-thalassemia is the most common form of beta-thalassemia worldwide. Approximately 1-3% of the African population is believed to carry a thalassemia gene, with 18.2% of these individuals having clinically significant hemoglobinopathies (Baird et al., 2022).

In sub-Saharan Africa, the prevalence of thalassemia, particularly beta-thalassemia, is high due to genetic predisposition and historical factors such as the endemicity of malaria (Obeagu, 2025). Beta-thalassemia is the most common form in Africa. Alpha-thalassemia is also prevalent, notably in East Africa and specific populations in central Africa (WHO, 2025).

Thalassemia is suspected in patients with microcytic anaemia and normal or elevated ferritin levels. Clinical manifestations vary from asymptomatic to transfusion-dependent severe forms. Haemoglobin electrophoresis may reveal features standard to the different thalassemia subtypes, but genetic testing is required to confirm the diagnosis (Baird et al., 2022). Regular blood transfusions and iron chelation therapy improve the prognosis of homozygous β -thalassemia. Life expectancy for sufferers has increased considerably, from a mere decade to around 30 years (Isaiah et al., 2024).

Increasing global migration has introduced thalassemias into many regions where they were not originally endemic (Kattamis et al., 2020). In Africa, particularly, there are few studies on thalassemia, and it is often confused with sickle cell disease due to limited diagnostic resources (Origa, 2017). Very few countries keep patient registers, and in many others, children die from the most serious transfusion-related syndromes before they are even diagnosed (Angastiniotis & Lobitz, 2019).

Hence, this systematic review aims to determine the prevalence of thalassemia in Africa, to improve knowledge about this disease.

Materials and Methods

Search strategy

This review was carried out in accordance with the PRISMA (Preferred Reporting Items for Systematic reviews and Meta-Analyses) recommendations for conducting and reporting the results of this meta-analysis.

Eligibility criteria

To be included, articles had to be available in full text, written in English or French, and report original studies presenting quantitative data on the prevalence of thalassemia in children in Africa. Studies with an analytical component were also included.

Study selection strategy

Pubmed, Google Scholar, and African Journal Online (AJOL) databases were searched in January 2025, with a lower limit of 2007, using keywords related to prevalence, thalassemia, Africa, and children to identify articles published in English and French.

A systematic literature review was conducted to identify articles reporting the prevalence of thalassemia among children in Africa. Several searches were conducted in PubMed, Google Scholar, and the African Journal Online (AJOL), and 11 studies were included in the final analysis. Our search strategy used various relevant keywords: “Prevalence”, “Thalassemia”, AND “Africa”.

Two authors (IAH and TO) independently reviewed the studies and made the selection decision. The third author reviewed the discrepancies (BM). No discrepancies were reported.

Quality assessment and risk-of-bias analysis

Independent reviewers (IAH, TO) critically appraised the included studies to ensure reliability and consistency of results. The Mixed Methods Assessment Tool (MMAT) (Hong et al., 2018), a critical appraisal tool, was used to assess the quality of the included studies. The MMAT assesses different study designs, including qualitative studies, randomized controlled trials, non-randomized studies, quantitative descriptive studies, and mixed-methods studies. Briefly, three steps must be followed: (i) answer two optional selection questions to determine whether the article is an empirical study, (ii) choose the appropriate study categories to be evaluated according to the study category (qualitative, quantitative or mixed methods) and (iii) evaluate the criteria of the chosen category using “Yes” (satisfied), ‘No’ (not satisfied) or “Can't say” (insufficient information). The criteria for assessing study quality are detailed in Supplementary Table 1.

Data extraction

Appropriate data from each eligible study were independently extracted by two reviewers (IAH and TO), with disagreements resolved through discussion. A third researcher (MDA) confirmed the eligibility of included studies before they were included in the analysis. The following data were extracted: name of first author, year of publication, country, sample size, and prevalence of thalassemia.

Statistical analysis

Data were analyzed using RStudio version 4.4.1. The prevalence and distribution of thalassemias were grouped by country, age group, and thalassemia type. A narrative summary of the results was produced, highlighting trends, geographical variations, and factors associated with thalassemia prevalence. [12] and metafor [13] are used. For the meta-analysis, meta (Balduzzi et al., 2019) and metafor (Viechtbauer, 2010) software were used to calculate the pooled percentage and 95% confidence interval (CI) using a random-effects model. Subgroup meta-analyses were

performed by age group and thalassemia type, according to the prevalence of thalassemia.

Statistical heterogeneity between studies was assessed using the heterogeneity statistic and the inconsistency index ($I^2 > 75\%$). A p-value of < 0.05 was considered substantial heterogeneity. A funnel plot and Egger's weighted regression test were used to detect publication bias in the meta-analysis. The funnel plot asymmetry, combined with a significant Egger's test (p-value < 0.05), indicates publication bias.

Results

The literature search yielded 1165 unique studies after title and abstract screening; 30 articles were eligible for full-text review and were reviewed in detail. Finally, 11 articles (El-Beshlawy et al., 2007; El-Shanshory et al., 2023; Barker et al., 2017; Brito et al., 2022; Engle-Stone et al., 2017; Gahutu et al., 2012; Laghmich et al., 2019; Macharia et al., 2020; Osman et al., 2020; Suchdev et al., 2014; Siala et al., 2008) were included in our systematic review. The selection process is illustrated in Figure 1.

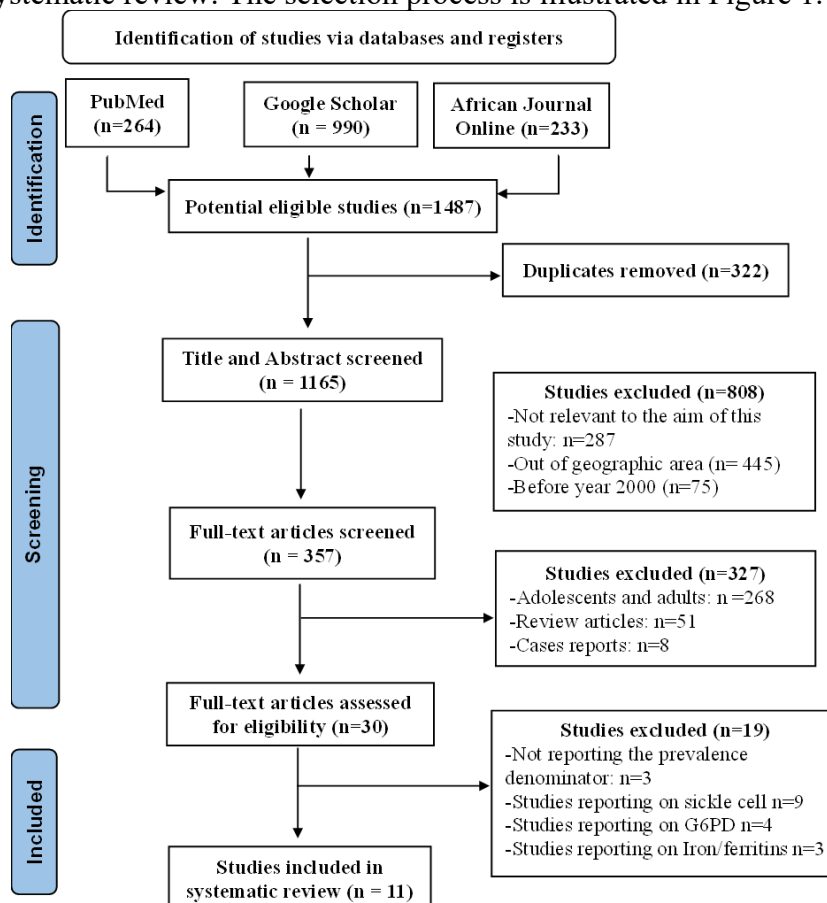


Figure 1: Diagram of literature search and study selection

Characteristics of included studies

This review covered 11 studies, including 10 cross-sectional and 1 cohort studies (Macharia et al., 2020). The 11 studies were published between 2007 and 2023 in 9 African countries (Figure 2): Egypt (n =2), Democratic Republic of Congo (n =1), Mozambique (n =1), Cameroon (n =1), Rwanda (n =1), Morocco (n =1), Kenya (n =2), Sudan (n =1) and Tunisia (n =1). Table 1 summarizes the basic characteristics of the populations studied.

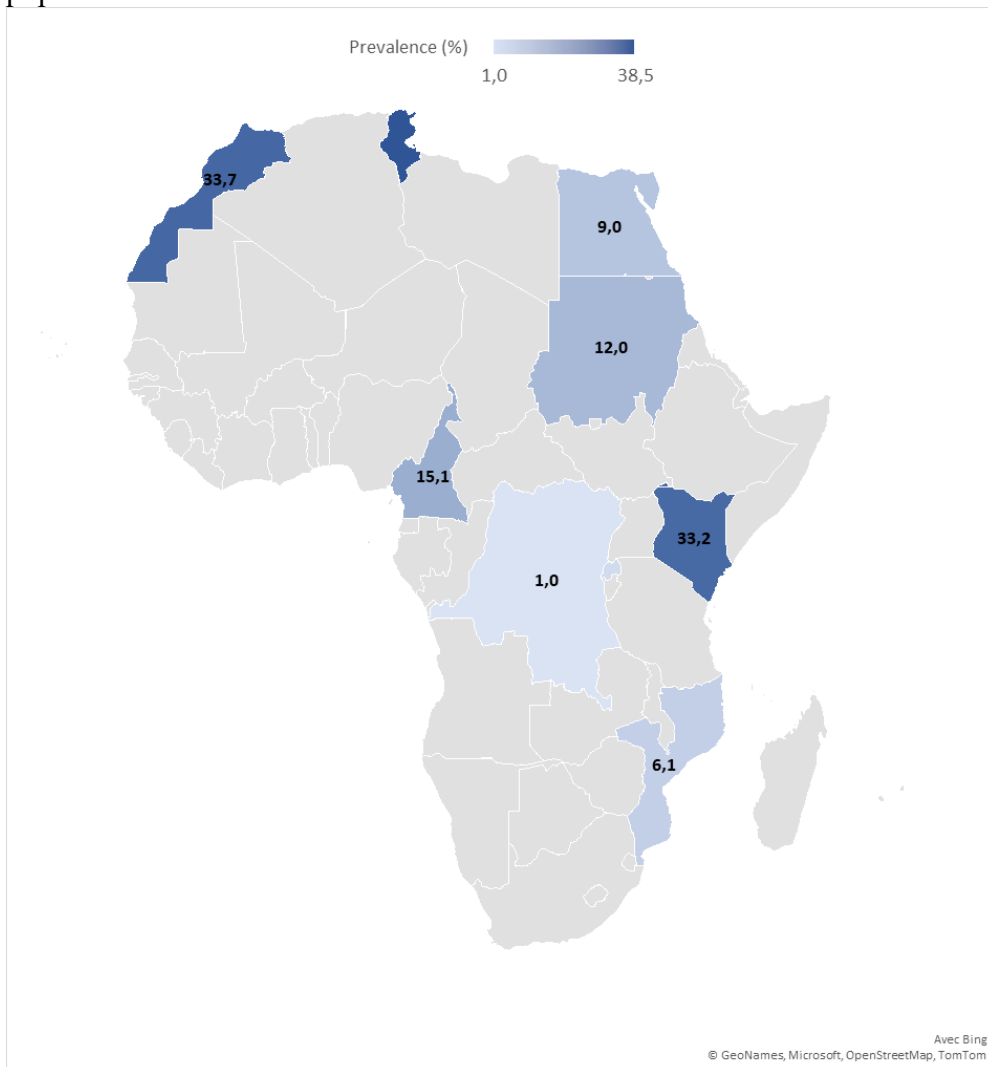


Figure 2: The prevalence of thalassemia disease in Africa

Table 1: Characteristics of the included studies

Authors, Year of publication	Country	Study design	Sample seize	Study population	Gender Male n (%)	Type of thalassemia	Proportion of thalassemia (%)	Technique used	Mutations
El-Beshlawy et al., 2007 (El-Beshlawy et al., 2007)	Egypt	Cross- sectional study	1000	Randomly selected children 5-16 years	NA	β - thalassemia	9.0	NA	NA
El-Shanshory et al. 2023 (El-Shanshory et al., 2023)	Egypt	Cross- sectional study	4320	Randomly selected students 14- 18 years	2172 (50.0)	β - thalassemia	6.1	High- performance liquid chromatography	NA
Barker et al., 2017 (Barker et al., 2017)	Democratic Republic of Congo	Cross- sectional study	744	Randomly selected children 6-59 months	372 (50.0)	α - thalassemia	12.0	PCR	$\alpha 3,7$
Brito et al., 2022 (Brito et al., 2022)	Mozambique	Cross- sectional study	232	Non- probability sampling of newborns	115 (49.0)	α - thalassemia	33.2	PCR Sequencing	$\alpha 3.7/4.2/-\alpha 3.7/4.2$ (9,9%)
Engle-Stone et al., 2017 (Engle-Stone et al., 2017)	Cameroon	Cross- sectional study	291	Randomly selected children 12- 59 months	154 (53.0)	α - thalassemia	33.7	PCR	homozygous heterozygous – $\alpha 3.7$ deletion
Gahutu et al., 2012 (Gahutu et al., 2012)	Rwanda	Cross- sectional study	749	Randomly selected children 0-59 months	NA	α - thalassemia	15.1	PCR	α +-thalassaemia
Laghmich et al., 2019	Morocco	Cross- sectional	1658	Randomly selected	933 (56.0)	α - thalassemia	1.0	PCR	$\alpha 3.7$ deletion (0,33%) $\alpha 4.2$ deletion (0,12%)

(Laghmich et al., 2019)		study		newborn					$\alpha\alpha\alpha$ anti 3.7 deletion (0,06%) HS-40 deletion (0,03%) --Med I deletion (0,03%)
Macharia et al., 2020 (Macharia et al., 2020)	Kenya	Cohort Study	15577	Randomly selected children 3-12 months	NA	β -thalassemia	11.4	PCR Sequencing	β 0-CD22 (GAA→TAA) β 0-Initiation codon (ATG→ACG) β 0-IVS1-3' end del 25 bp β +-IVS-I-110 (G→A)
Osman et al., 2020 (Osman et al., 2020)	Sudan	Cross-sectional study	67	Randomly selected patients 12-17 years	28 (42.0)	α -thalassemia	3.0	PCR	α 3.7 deletion (2,99%)
Suchdev et al., 2014 (Suchdev et al., 2014)	Kenya	Cross-sectional study	858	Randomly selected children 6-35 months	429 (50.0)	α -thalassemia	38.5	PCR	NA
Siala et al., 2008 (Siala et al., 2008)	Tunisia	Cross-sectional study	529	Randomly selected newborn	NA	α -thalassemia	7.4	PCR Sequencing	α 2 -globin gene (acd23-GAG→Stop), α 1 -globin gene (acd119-CCT→TCT),

Proportion of thalassemia in Africa

A total of 26025 participants were included in this systematic review. We determined the prevalence of thalassemia as the proportion of a given population affected by thalassemia. The children studied, in whom thalassemia (type α or β) had been diagnosed, belonged to varied age groups: children under five, those between five and ten, and those over ten (Table 1). The overall combined prevalence of thalassemia was 13.4% (95% CI, 8.5-19.1%, $I^2 = 99.1\%$) (Figure 3).

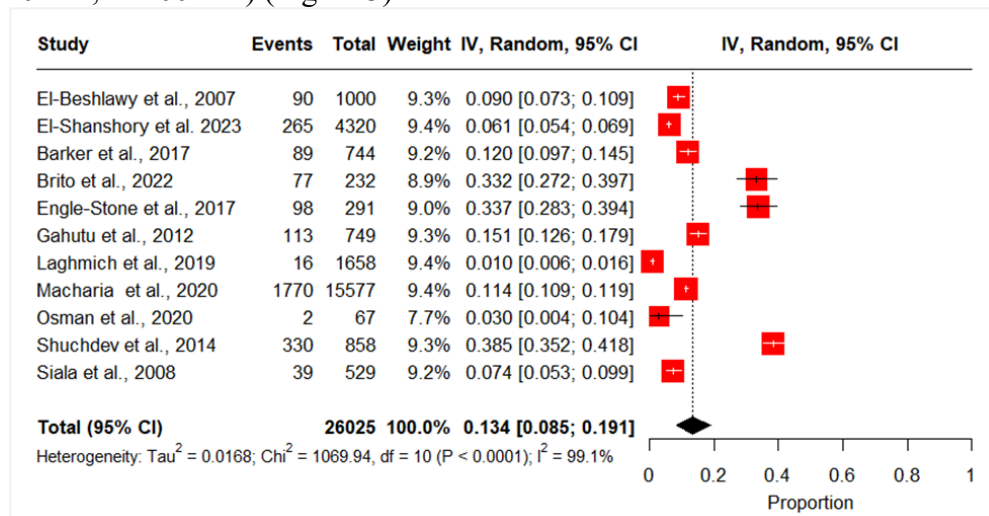


Figure 3: Prevalence of thalassemia in Africa

Subgroup analysis

Figures 3 and 4 show the subgroup analysis of thalassemia prevalence by age category and thalassemia type. Subgroup analysis by age category revealed that the newborn, under-5, and over-5 age groups, respectively, had 14% (95% CI: 0-33%), 22% (95% CI: 11-33%), and 6% (95% CI: 3-9%) prevalence of thalassemia (Figure 4).

Subgroup analysis by thalassemia type revealed that the prevalence of alpha-thalassemia was 18% (95% CI: 7-28%) and beta-thalassemia 9% (95% CI: 6-12%) (Figure 5).

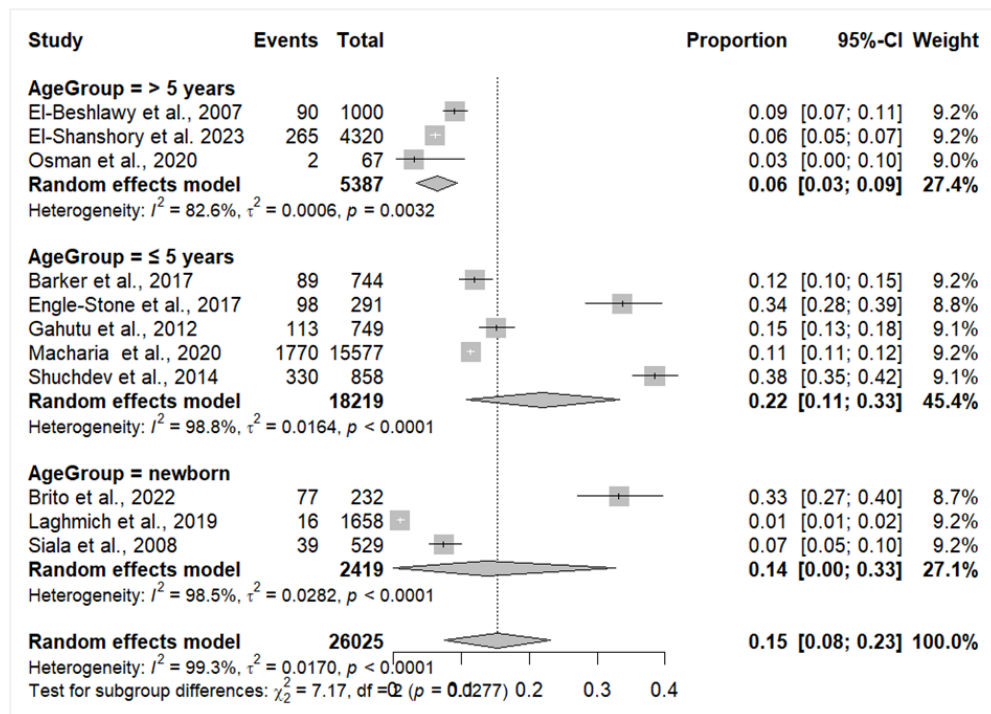


Figure 4: Analysis by patient age subgroup

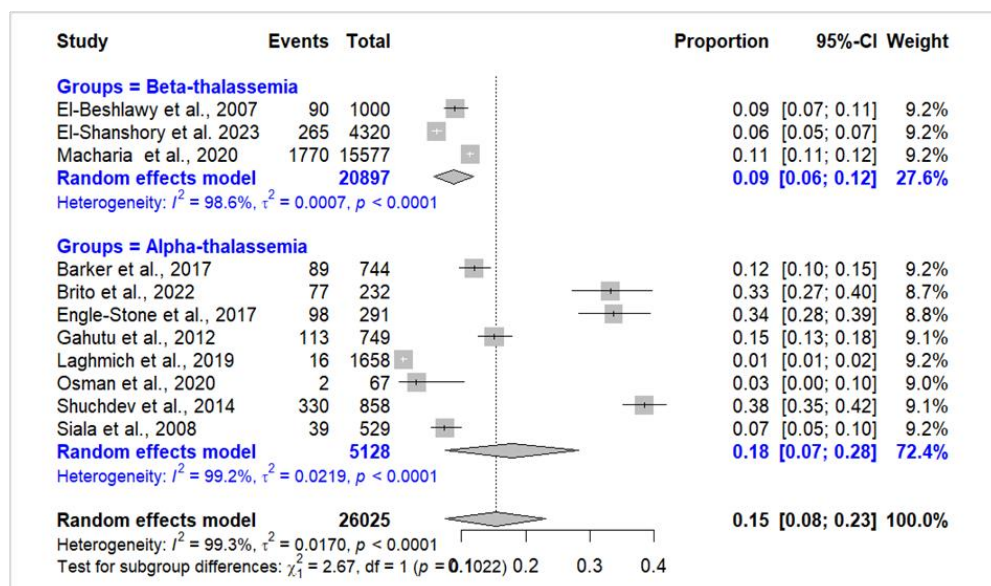


Figure 5: Analysis by thalassemia type subgroup

Study quality and assessment of risk of bias

The included studies were visually assessed for publication bias using a funnel plot. The funnel plot asymmetry indicated publication bias (Figure 6). Similarly, Egger's test revealed significant publication bias ($p < 0.0001$).

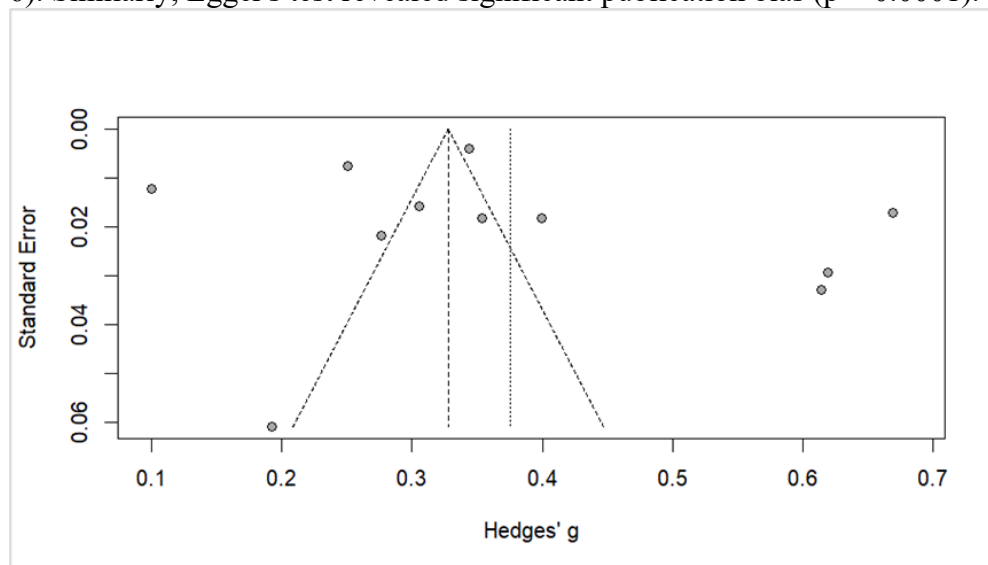


Figure 6: Funnel plot for publication bias of pooled prevalence of thalassemia in Africa from 2007 to 2025

Discussion

This systematic review aimed to determine the prevalence of thalassemia in Africa. This systematic review included 11 observational studies conducted in Africa. These studies included 2,889 cases of thalassemia among 25026 children. The meta-analysis, which included studies conducted between 2007 and 2023, estimated the prevalence of alpha-thalassemia at 18% (95% CI, 7-28%; $I^2 = 99.2\%$) and beta-thalassemia at 9% (95% CI, 6-12%; $I^2 = 98.6\%$). The present study contributes to the existing literature by providing a valuable database for accessing thalassemia data.

This research is motivated by the need to reinforce current knowledge and to estimate the prevalence of thalassemia in Africa. Although thalassemia is widely studied in other parts of the world, there is a paucity of research specific to Africa. This meta-analysis aims to fill this gap by providing a contextualized understanding of childhood thalassemia in the African setting.

The overall combined prevalence of thalassemia was 13.4% (95% CI, 8.5-19.1%). The prevalence of thalassemia (carrier or affected) varied across countries, from 1% in the DRC to 38.5% in Tunisia. This result is

comparable to that of Rao et al., who reported that thalassemia prevalence can range from 10% to 20% in sub-Saharan Africa (Rao et al., 2024).

Newborns, children under 5 years, and children over 5 years had respectively 14% (95% CI: 0-33%), 22% (95% CI: 11-33%), and 6% (95% CI: 3-9%) prevalence of thalassemia.

Similarly, a higher prevalence of α -thalassemia (carriers or patients) ranging from 36% to 49% was reported for children under five in Saudi Arabia (Abu-Shaheen et al., 2020).

Sub-Saharan Africa, which has one of the highest malaria burdens in the world, also has a high rate of thalassemia gene mutations, particularly in populations living in areas where malaria is endemic (Egesa et al., 2022; Li et al., 2024).

In addition, sub-Saharan Africa faces various systemic barriers to healthcare infrastructure, including limited access to diagnostic centres, blood transfusion services, and specialized medical care, which hinder effective thalassemia management (Obeagu, 2025).

The prevalence of alpha-thalassemia was 18% (95% CI: 7-28%), and that of beta-thalassemia was 9% (95% CI: 6-12%). Similar to some research, 5-10% of residents in Africa may carry the thalassemia gene. The prevalence rate may vary by country or region. For example, in countries such as Nigeria, Kenya, and Ghana, the proportion of beta-thalassemia carriers ranges from 5 to 10% (Obeagu, 2025). The importance of thalassemia in sub-Saharan Africa is particularly pronounced in countries located in the equatorial belt, where malaria transmission is high (Obeagu, 2025). Because they interact with malaria resistance and thalassemias, especially α -thalassemia, these conditions are common. In Kenya and Tanzania, approximately 40–50% of the population has gene deletions that cause α -thalassemia (Mugisha Emmanuel, 2025). Our findings revealed that β -thalassemia is rare but has been reported in Egypt and Kenya. This result is similar to that reported by Twum et al., who found that β -thalassemia is more prevalent in northern Africa, with Algeria and Morocco showing the highest prevalence (Twum et al., 2023). Increasing these carrier rates increases the risk of children inheriting the disease, particularly beta-thalassemia major, which can lead to severe anaemia and requires ongoing medical care. The epidemiology of β -thalassemia is evolving due to several factors. Migration, the introduction of β -thalassemia prevention initiatives, and increased survival rates are among these causes (Kattamis et al., 2020).

Furthermore, the extent of thalassemia in sub-Saharan Africa is frequently underestimated, as screening for this condition is not systematic. Many individuals with mild forms of the disease may remain undiagnosed or be misdiagnosed with other forms of anaemia (Isaiah et al., 2024; Vichinsky, 2016).

This study provides crucial epidemiological information on paediatric thalassemia in Africa. To prevent and control paediatric thalassemia worldwide, these data are essential for allocating resources and developing public health strategies. It is crucial to set up national programs for compulsory screening for thalassemia through premarital and neonatal screening. It is also essential to target young adults with information about thalassemia and its consequences, to reduce intermarriage among thalassemia carriers and lower the incidence of the disease.

We make practical policy recommendations based on the findings of this study, such as encouraging thalassemia screening programs and public awareness campaigns in Africa, strengthening international cooperation and aid, increasing investment in healthcare resources and infrastructure, training healthcare professionals, implementing economic support policies, and promoting genetic counseling services. By raising public awareness of thalassemia and increasing early detection rates, these actions aim to reduce the impact of the disease in Africa significantly (Li et al., 2024).

This systematic review has several strengths. The search strategy was comprehensive, utilizing numerous electronic databases, and a substantial number of high-quality studies were included in this review. Only two of the 11 studies analyzed were of poor quality.

However, the main limitation of this systematic review is the lack of data from other countries on the continent, as well as on mortality rates and risk factors. The quality of the prevalence studies was limited by methodological problems, including non-randomized sampling, heterogeneity in the populations examined, and the lack of standardized diagnostic tests. High heterogeneity is a significant feature of these results, evident in all groups ($I^2 = 99.1\%$). This means that the studies included vary significantly in terms of methodology, geographical location, types of population examined, and methods of thalassemia diagnosis. This heterogeneity limits the generalizability of prevalence estimates and highlights the need for greater standardization of surveillance methods.

Conclusion

This study on the prevalence of thalassemia in Africa could contribute to the development of health policies for screening large populations and to the implementation of better genetic counseling programs.

Authors' Contributions

IAH, TO, and MB participated in the conceptual design and development of the current study. HO, TO, and AZ assisted in the design and analysis of the search strategies. IAH conducted searches, screened, and reviewed the literature, producing data extraction and spreadsheets with

assistance from MDA and TO, who reviewed and screened studies, performed data extraction, and provided additional support. When a consensus could not be reached, the articles were reviewed by BM. IAH, NTN, and TO contributed to the data analysis. NTN constructed geographic mapping, and TO created the R code used to analyze the extracted data, leading to the development of analytic strategies and methods. All the authors participated in the initial quality assessment of the included studies. IAH drafted the manuscript, which was reviewed and revised by TO, AZ, HO, and OA. BM coordinated and directed the research and reviewed and revised the manuscript. All authors read and approved the final manuscript.

Conflict of Interest: The authors reported no conflict of interest.

Data Availability: All data are included in the content of the paper.

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Appendix

Table 1: Quality of the included papers, Mixed Methods Appraisal Tool (MMAT)

Category	Criteria	El-Beshlawy et al., 2007	El-Shanshory et al. 2023	Barker et al., 2017	Brito et al., 2022.	Engle- Stone et al., 2017	Gahutu et al., 2012	Laghmich et al., 2019	Macharia et al., 2020	Osman et al., 2020	Suchdev et al., 2014	Siala et al., 2008
Screening questions	S1. Are there clear research questions?											
	S2. Do the collected data allow to address the research questions?											
4. Quantitative descriptive studies	4.1. Is the sampling strategy relevant to address the research question?											
	4.2. Is the sample representative of the target population?											
	4.3. Are the measurements appropriate?											
	4.4. Is the risk of nonresponse bias low?											
	4.5. Is the statistical analysis appropriate to answer the research question?											
	Total score	12	14	14	14	14	12	14	14	11	12	10
Legend		Yes = 2	Partially = 1	No = 0								