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A Bibliometric Analysis of Gene Mutations in Thalassemia: Research Trends, Knowledge Structure, and Emerging Themes

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ABSTRACT

Introduction: Thalassemia is an inherited blood disorder caused by impaired globin-chain production, with gene mutations playing a central role in its molecular basis and phenotypic diversity. The rapid expansion of genetic and genomic research has generated a broad and increasingly interconnected body of literature, making it important to characterize the development and structure of this research field.
Objective: This study aimed to map research trends, knowledge structure, thematic patterns, and international collaboration in the field of gene mutations in thalassemia.
Method: A bibliometric analysis was conducted using publications retrieved from PubMed on 20 August 2024. The search strategy was "gene mutation AND thalassemia" and was limited to publications from 2019 to 2024. Bibliographic data were analyzed using RStudio with the Biblioshiny application. Analyses included annual scientific production, relevant sources, Bradford's Law, thematic mapping, keyword co-occurrence, and international collaboration.
Result: The analysis demonstrated sustained research activity during 2019–2024, with the highest publication output in 2020 (213 publications). Hemoglobin was the most productive source, with 131 publications, followed by Annals of Hematology (30) and Hematology (23). Thematic and keyword analyses identified mutation, genotype, alleles, sequence analysis, α-thalassemia, β-thalassemia, genetic testing, prenatal diagnosis, fetal hemoglobin, and gene editing as major interconnected research areas. The collaboration analysis demonstrated broad international research networks involving countries across multiple regions.
Conclusion: Research on gene mutations in thalassemia has developed from fundamental mutation and genotype characterization toward genetic diagnosis, prenatal applications, and emerging genomic therapeutic approaches. These findings highlight the importance of continued molecular research and international collaboration to support the translation of genomic knowledge into thalassemia prevention, diagnosis, and personalized care.

Keywords: thalassemia, gene mutation, genotype, genetic testing, bibliometric analysis

Introduction

Thalassemia is one of the most important inherited blood disorders worldwide and represents a substantial public health burden, particularly in regions where hemoglobinopathies are highly prevalent (Shafique et al., 2023). The disease is characterized by impaired synthesis of one or more globin chains, resulting in ineffective erythropoiesis, chronic anemia, and, in severe forms, lifelong transfusion dependence (Gluba-Brzózka et al., 2021). Beyond its clinical consequences, thalassemia imposes considerable social and economic burdens on affected individuals, families, and health systems (Yousuf et al., 2022). The heterogeneous clinical manifestations of thalassemia are closely related to its underlying genetic architecture, making molecular characterization an essential component of understanding disease phenotype, diagnosis, prognosis, and management (Harteveld et al., 2022).

Gene mutations are fundamental to the pathogenesis and phenotypic diversity of thalassemia. Mutations affecting the α-globin and β-globin genes can alter globin-chain synthesis through diverse molecular mechanisms, including point mutations, deletions, insertions, splicing abnormalities, and regulatory alterations (Tesio & Bauer, 2023). The resulting genotypes may range from clinically silent carrier states to severe transfusion-dependent disease. Importantly, the relationship between genotype and phenotype is not always straightforward, as clinical expression may also be influenced by co-inherited genetic variants, fetal hemoglobin regulation, environmental factors, and other biological modifiers (Paschoudi et al., 2023). Consequently, research on thalassemia-associated gene mutations has expanded beyond the identification of pathogenic variants toward genotype, phenotype correlations, molecular diagnosis, population-specific mutation spectra, and increasingly sophisticated genomic approaches (Jaing et al., 2021).

Advances in molecular and genomic technologies have further transformed the landscape of thalassemia research (Ali et al., 2021). Conventional molecular techniques have progressively been complemented by high-throughput sequencing and other genomic approaches, enabling more comprehensive identification and characterization of disease-associated variants (Smirnov et al., 2023). These developments have supported applications ranging from carrier detection and prenatal diagnosis to individualized genetic counseling and molecular classification. At the same time, emerging approaches such as gene editing and strategies targeting fetal hemoglobin and erythropoiesis have introduced new possibilities for therapeutic intervention (Quintana-Bustamante et al., 2022). The increasing diversity of research questions, technologies, populations, and molecular targets has therefore generated a broad and rapidly evolving body of literature on gene mutations in thalassemia.

Despite this expanding evidence base, the research landscape of gene mutations in thalassemia remains dispersed across multiple disciplines and publication sources. Research has addressed distinct aspects of mutation biology, including sequence analysis, genotype characterization, allelic variation, gene expression, genetic testing, prenatal diagnosis, and emerging genomic technologies (Claussnitzer et al., 2020). The bibliometric evidence from the present dataset similarly demonstrates a broad knowledge structure connecting mutation and thalassemia with genotype, alleles, sequence analysis, α-thalassemia, β-thalassemia, genetic testing, prenatal diagnosis, gene expression, fetal hemoglobin, and gene editing (Achour et al., 2021). This diversity reflects the multidimensional nature of the field but also makes it difficult to identify the dominant research directions, influential publication sources, major thematic

clusters, and emerging areas of investigation through conventional narrative assessment alone (Wortmann et al., 2022).

Bibliometric analysis provides a systematic approach for examining the development and structure of a scientific field through quantitative analysis of publications, sources, authors, keywords, citations, and collaboration patterns (Manoj Kumar L et al., 2023). In the context of gene mutations in thalassemia, such an approach can reveal how scientific attention has evolved over time, which journals have served as major knowledge hubs, how research themes are interconnected, and how international collaboration has shaped the field. The present dataset includes 999 documents distributed across 352 sources and involving 5,254 authors, indicating a substantial and multidisciplinary research landscape. The analysis further incorporates source concentration through Bradford's Law, thematic mapping, keyword co-occurrence, and country-level collaboration, allowing the intellectual and collaborative structure of the field to be examined from complementary perspectives.

However, a comprehensive mapping specifically focused on the evolution and knowledge structure of gene mutation research in thalassemia is needed to clarify how this field has developed and where it is heading (Orkin, 2021). Identifying dominant themes and emerging research directions may help researchers recognize knowledge gaps, facilitate interdisciplinary collaboration, and inform future genomic and translational research in thalassemia.

Objective

This study aimed to conduct a bibliometric analysis of the scientific literature on gene mutations in thalassemia, with particular emphasis on research trends, the structure of the knowledge base, major publication sources, thematic patterns, keyword relationships, and international collaboration. By mapping these dimensions, this study seeks to provide a comprehensive overview of the current research landscape and identify emerging directions that may shape future investigations into the genetic basis and genomic management of thalassemia.

Methods

This study employed a bibliometric analysis to systematically evaluate the development, knowledge structure, thematic patterns, and international collaboration of research on gene mutations in thalassemia. Bibliometric analysis was used to quantitatively characterize the scientific literature and identify major research trends, relevant publication sources, conceptual relationships, and emerging research themes within the field.

Bibliographic records were retrieved from PubMed, a comprehensive biomedical literature database covering research in medicine, health sciences, genetics, molecular biology, and related disciplines. PubMed was selected because of its relevance to the biomedical and genetic focus of research on gene mutations in thalassemia. The literature search was conducted on 20 August 2024. The search strategy used the following query: "gene mutation AND thalassemia". The search was limited to publications published between 2019 and 2024.

Publications were considered eligible when they were indexed in PubMed, published within the 2019–2024 study period, addressed thalassemia, and examined gene mutations or related genetic characteristics of thalassemia. Records that were unrelated to the research

topic or lacked sufficient bibliographic information for bibliometric analysis were excluded. The retrieved records were exported from PubMed for subsequent bibliometric analysis.

Prior to analysis, the bibliographic dataset was examined and prepared to ensure consistency of the available bibliographic information. Variations in terminology, keywords, author names, and other metadata were standardized where necessary. Particular attention was given to keywords and indexing terms because genetic and molecular concepts may be represented using different terms across publications. This preprocessing was performed to minimize artificial fragmentation of conceptually related research topics in subsequent network and thematic analyses.

The bibliometric analysis included descriptive publication analysis, annual scientific production, identification of the most relevant sources, Bradford's Law analysis, thematic mapping, keyword co-occurrence analysis, and international collaboration analysis. Annual scientific production was assessed by calculating the number of publications for each year between 2019 and 2024 to characterize temporal changes in research activity. The most relevant sources were identified based on the number of publications related to gene mutations in thalassemia, allowing the principal publication venues contributing to the field to be determined.

Bradford's Law was applied to examine the concentration and distribution of publications across scientific journals and to identify the core sources contributing substantially to the literature. Thematic mapping was performed to examine the conceptual structure of the research field according to the centrality and density of research themes. The resulting thematic structure was used to distinguish established and interconnected research areas from more specialized or emerging topics.

Keyword co-occurrence analysis was conducted to identify relationships among frequently occurring terms within the retrieved publications. A co-occurrence network was generated to visualize the conceptual relationships and clusters within the literature. The network was examined to identify major research areas and relationships among genetic, molecular, diagnostic, and clinical concepts associated with thalassemia.

International collaboration was assessed using country-level co-authorship analysis. Countries were identified from author affiliation information, and a collaboration network was generated to visualize relationships among countries contributing to research on gene mutations in thalassemia. The network was used to characterize the geographical distribution and international connectivity of scientific research in this field.

Bibliometric analysis and visualization were performed using RStudio with the Biblioshiny application. Biblioshiny was used to generate descriptive bibliometric statistics and graphical representations, including annual scientific production, relevant sources, Bradford's Law, thematic mapping, keyword co-occurrence networks, and country collaboration networks. The bibliometric findings were interpreted descriptively, with publication trends used to characterize the temporal development of the field, source analysis used to identify major publication venues, thematic and co-occurrence analyses used to identify dominant and emerging research areas, and country collaboration analysis used to characterize international research connectivity.

This study used bibliographic records obtained from the PubMed database and did not involve human participants, patient-level data, biological specimens, or identifiable personal information. Therefore, ethical approval and informed consent were not required.

Results

Annual Scientific Production

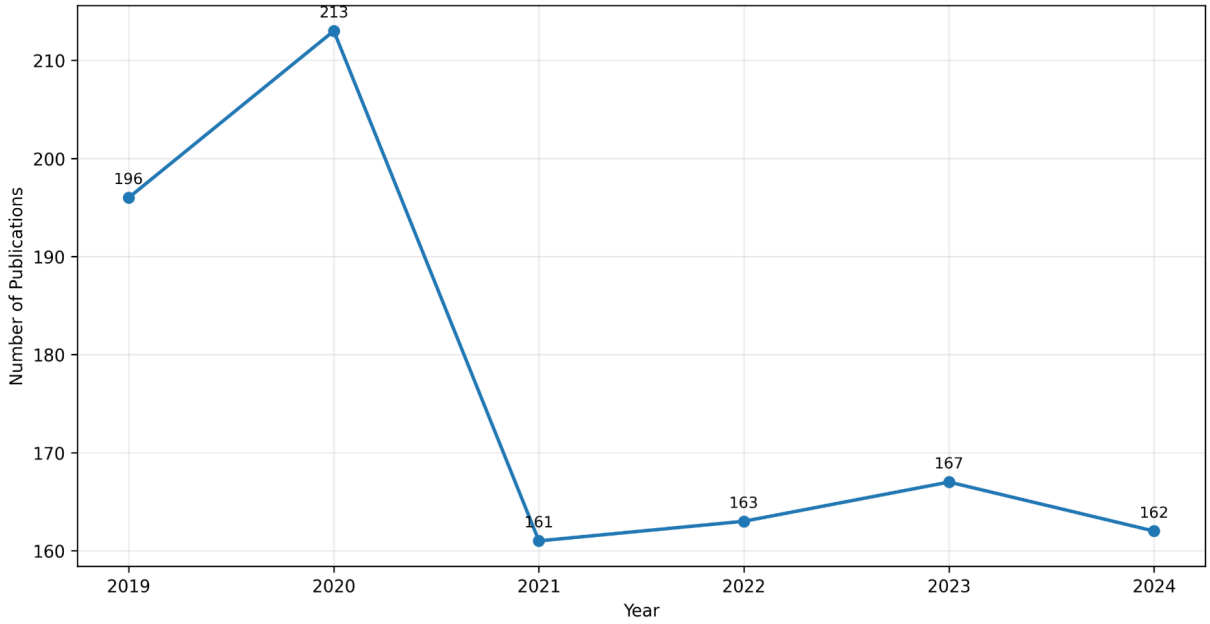

Figure 1. Annual scientific production of research on gene mutations in thalassemia

The annual scientific production of research on gene mutations in thalassemia demonstrated a fluctuating publication pattern during the 2019–2024 study period (Figure 1). A total of 196 publications were recorded in 2019, followed by an increase to 213 publications in 2020, representing the highest annual publication output during the study period. Publication production subsequently declined substantially to 161 publications in 2021. The number of publications then showed a modest recovery, increasing to 163 publications in 2022 and 167 publications in 2023, before slightly declining to 162 publications in 2024.

Overall, the publication trend indicates sustained scientific interest in gene mutations in thalassemia throughout the study period, despite year-to-year fluctuations. The highest level of scientific productivity was observed in 2020, whereas the lowest output occurred in 2021. Following the decline in 2021, publication activity remained relatively stable between 2022 and 2024, suggesting continued research attention to the genetic and molecular aspects of thalassemia.

Most Relevant Sources

The publications were distributed across several scientific journals, with Hemoglobin being the most productive source, contributing 131 publications (Figure 2). It was followed by Annals of Hematology (30), Hematology (23), Frontiers in Genetics (21), and Scientific Reports (20). Other relevant sources contributed between 13 and 19 publications.

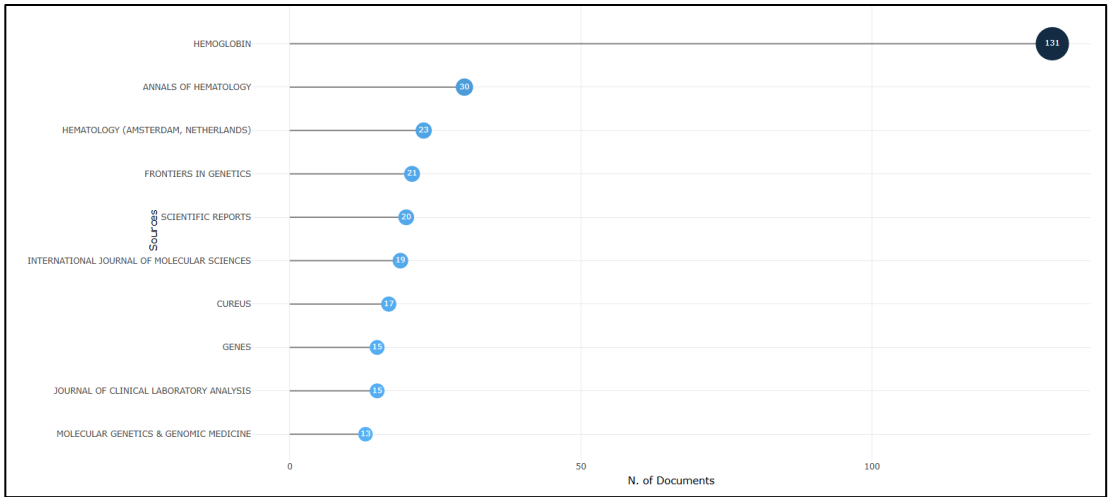

Figure 2. Most relevant sources of research on gene mutations in thalassemia

Bradford's Law

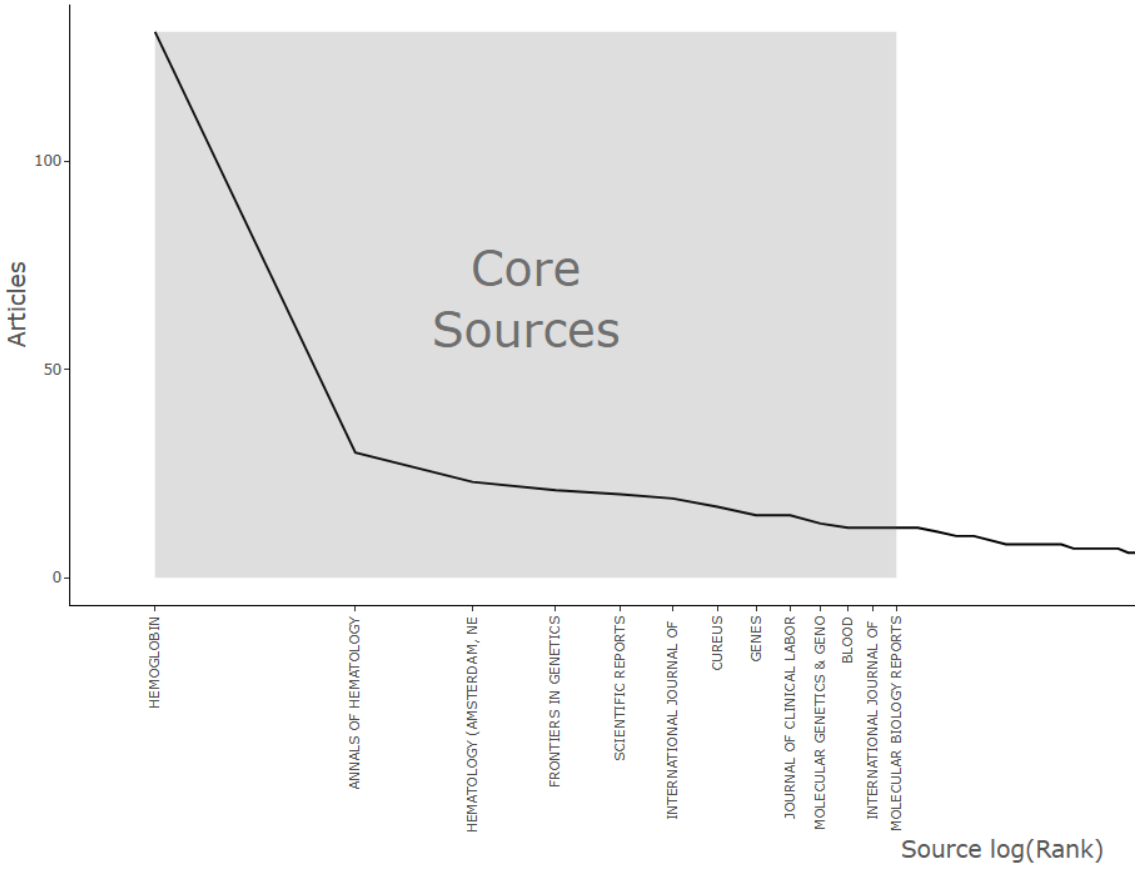

Figure 3. Bradford's Law of research on gene mutations in thalassemia

Bradford's Law showed a strong concentration of publications among a small number of core sources (Figure 3). Hemoglobin was the most prominent source with 131 publications, followed by Annals of Hematology (30) and Hematology (23). The remaining sources contributed fewer than 22 publications each, indicating a marked decline in publication frequency across source ranks.

Thematic Map

The thematic map identified four major thematic clusters in gene mutation research on thalassemia (Figure 4). The themes centered on human mutation and β-thalassemia, genetic and molecular characterization of α-thalassemia, demographic characteristics, and genetic testing with prenatal diagnosis. These clusters indicate that research spans molecular mechanisms, genetic characterization, clinical populations, and diagnostic applications.

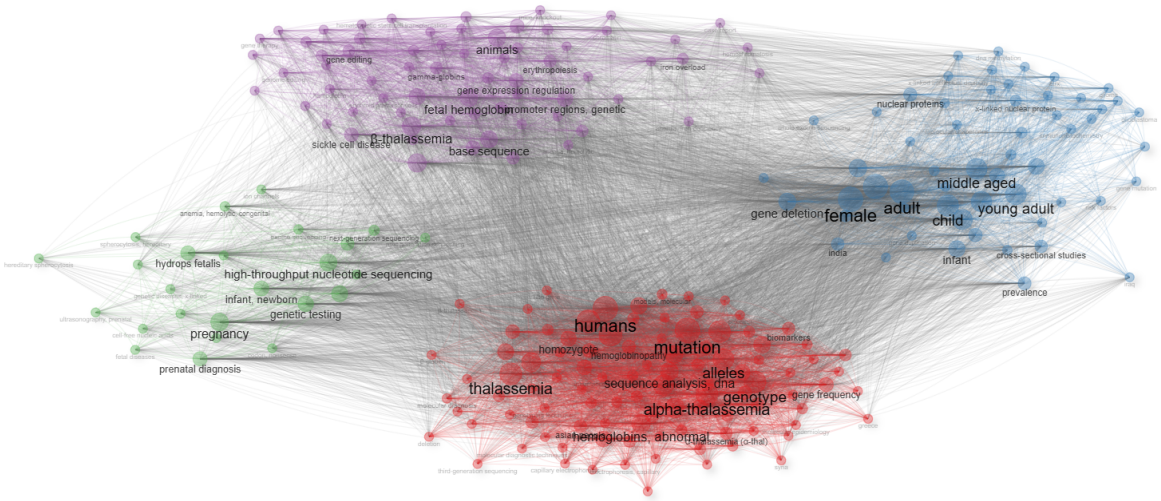

Figure 4. Thematic structure of research on gene mutations in thalassemia

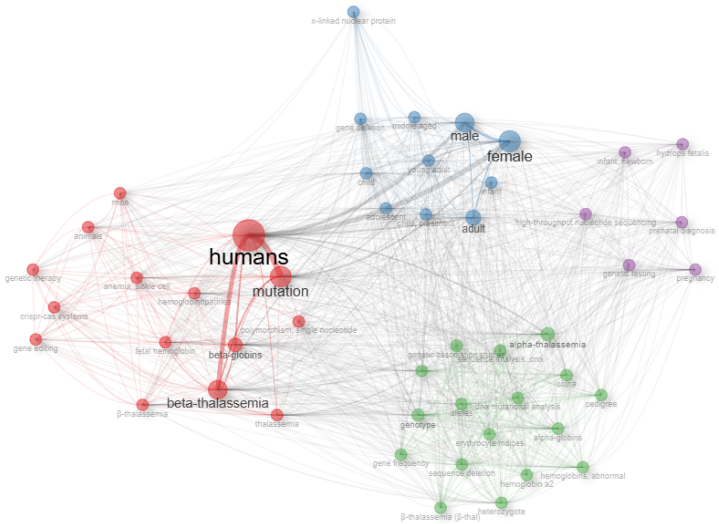

Figure 5. Keyword co-occurrence network of research on gene mutations in thalassemia

Keyword Co-occurrence Network

The keyword co-occurrence network revealed several interconnected research clusters in gene mutation research on thalassemia (Figure 5). The central network was dominated by “humans,” “mutation,” “beta-thalassemia,” and “thalassemia.” Other clusters focused on alpha-thalassemia, genotype, alleles, sequence analysis, genetic testing, prenatal diagnosis, and gene editing. The network demonstrates strong conceptual connections between molecular genetics, genetic diagnosis, and clinical characteristics of thalassemia.

World-Countries Collaboration

The country collaboration map demonstrated extensive international collaboration in gene mutation research on thalassemia (Figure 6). The United States, China, and several European and Asian countries showed prominent collaborative connections. The network also indicated collaboration extending to Australia, Africa, and South America, reflecting the geographically diverse nature of research in this field.

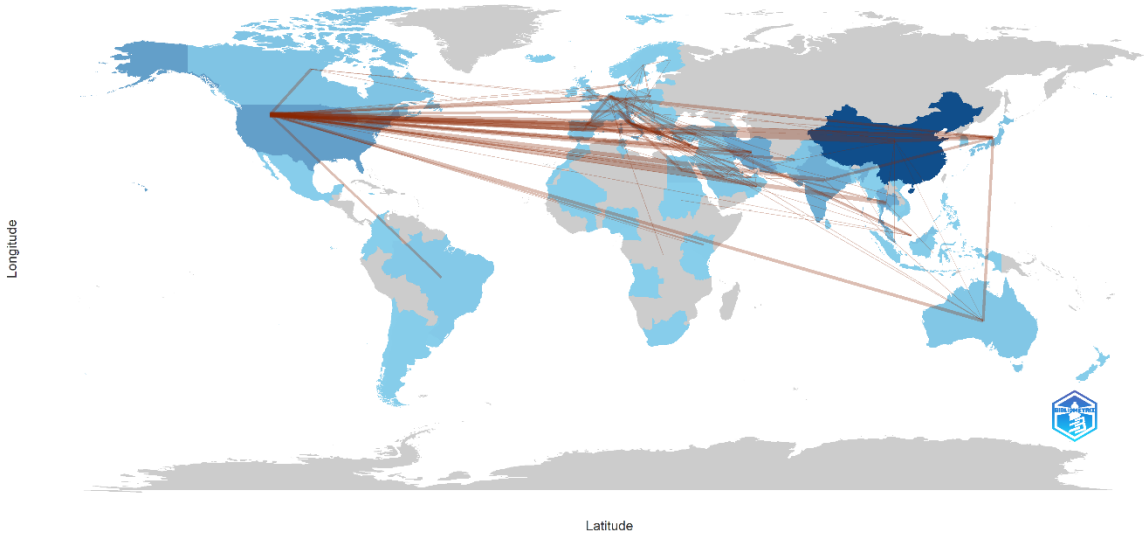

Figure 6. World-countries collaboration in research on gene mutations in thalassemia

Discussion

This bibliometric analysis provides an overview of the research landscape on gene mutations in thalassemia during 2019–2024. The findings demonstrate sustained scientific productivity, a strong concentration of publications in a limited number of hematology and genetics journals, and a research structure centered on mutation, genotype, alleles, sequence analysis, and α- and β-thalassemia. The thematic and co-occurrence analyses further showed that research has expanded toward genetic testing, prenatal diagnosis, gene expression, fetal hemoglobin, and gene editing. These findings indicate that gene mutation research in thalassemia is no longer limited to identifying pathogenic variants but increasingly connects molecular characterization with diagnosis, reproductive health, and emerging therapeutic strategies.

The publication trend showed substantial and sustained research activity throughout the study period, with the highest number of publications observed in 2020 and relatively stable output thereafter. This pattern is consistent with broader bibliometric evidence showing continued expansion of thalassemia research. A previous bibliometric analysis of β-thalassemia research covering 2013–2023 identified a steady increase in scientific publications and reported gene mutations among the major research hotspots, together with iron overload, anemia, and ineffective erythropoiesis (Lv et al., 2024). The present analysis provides a more focused perspective by specifically examining the genetic mutation dimension of thalassemia. The sustained publication activity observed in our study suggests that molecular genetics remains a fundamental research domain within the broader thalassemia field.

The strong dominance of Hemoglobin as the most productive source is another important finding. Hemoglobin contributed substantially more publications than other sources, followed by Annals of Hematology, Hematology, Frontiers in Genetics, and Scientific Reports. Bradford's Law further demonstrated a marked concentration of publications within a relatively small group of core journals. This pattern indicates that thalassemia mutation research is anchored primarily in specialized hematology and genetics literature, while also being disseminated through broader molecular and multidisciplinary journals. A similar pattern has been reported in previous bibliometric research, in which Hemoglobin was identified as one of the leading publication sources for α-thalassemia research (Ren et al., 2023). The concentration of research in specialized journals may facilitate knowledge accumulation within established scientific communities while simultaneously highlighting opportunities for greater interdisciplinary integration.

The thematic structure provides further insight into the scientific priorities of the field. The central themes surrounding mutation, genotype, alleles, sequence analysis, and α-thalassemia indicate that characterization of genetic variation remains a foundational research activity. This emphasis is biologically meaningful because thalassemia is fundamentally caused by pathogenic variants affecting globin-chain production, while the clinical phenotype may vary substantially according to the specific mutation, genotype, and co-inherited modifiers. A recent review of the global distribution of β-thalassemia mutations reported more than 350 disease-causing mutations and emphasized substantial geographical clustering, with a relatively small number of common mutations accounting for a large proportion of mutations in particular populations (Zyontz & Pomeroy-Carter, 2021). Thus, the prominence of genotype, alleles, and sequence analysis in the present network reflects an ongoing need to define population-specific mutation spectra and improve genotype–phenotype understanding.

The prominence of genetic testing and prenatal diagnosis represents another important dimension of the research landscape. The co-occurrence network showed connections among genetic testing, pregnancy, prenatal diagnosis, and high-throughput nucleotide sequencing, suggesting that molecular research is increasingly linked to reproductive and preventive applications. This direction has direct clinical relevance because accurate identification of parental carrier status and pathogenic variants enables risk assessment and reproductive counseling. Recent research has demonstrated the value of combining multiple molecular techniques, including MLPA, Sanger sequencing, targeted next-generation sequencing, and single-molecule real-time sequencing, particularly for rare or complex thalassemia variants encountered in prenatal diagnosis (Wu et al., 2022). The convergence of mutation research and prenatal diagnosis therefore suggests a translational pathway from molecular discovery to prevention of severe thalassemia.

The thematic presence of gene expression, fetal hemoglobin, erythropoiesis, and gene editing indicates that the field is also moving toward therapeutic genomics. Rather than focusing exclusively on correcting or identifying the primary globin mutation, research increasingly considers molecular mechanisms that can modify disease severity, including regulation of fetal hemoglobin and erythropoiesis. The broader bibliometric analysis of β-thalassemia similarly identified gene editing as an emerging research direction. More recent bibliometric evidence on gene therapy for inherited hematologic disorders has also demonstrated rapid growth in research involving genome editing, CRISPR-based approaches,

and clinical translation, with thalassemia forming an important component of this research landscape (Gamage et al., 2023). These developments suggest that mutation research is increasingly connected to therapeutic strategies capable of addressing the underlying molecular mechanisms of disease.

The international collaboration network further demonstrates that gene mutation research in thalassemia is geographically distributed and supported by cross-national scientific collaboration. The involvement of countries from multiple regions is particularly relevant because thalassemia mutation spectra vary considerably between populations and geographical areas (Yu et al., 2022). International collaboration can therefore facilitate comparison of mutation patterns, validation of molecular diagnostic approaches, and development of more comprehensive genomic databases. The global distribution of β-thalassemia mutations is influenced by geographical clustering and population migration, making collaborative research important for understanding changing mutation patterns across populations. The international nature of the research network also supports the need for harmonized genetic testing and data-sharing approaches that can improve the comparability of findings across populations (Torres-Espín & Ferguson, 2022).

From a clinical and translational perspective, these findings suggest that future thalassemia research should increasingly integrate mutation characterization with genetic counseling, carrier screening, prenatal diagnosis, and individualized management. The identification of specific mutation patterns is not merely a molecular research objective; it can influence diagnostic strategies, reproductive decision-making, prognosis, and eligibility for emerging molecular therapies. Research from regional populations has continued to identify distinct mutation frequencies and distributions, reinforcing the importance of population-specific molecular data for thalassemia prevention and genetic counseling (Enni, 2023). Consequently, the research landscape identified in this study supports a broader genomic medicine framework in which molecular information is translated into prevention, diagnosis, counseling, and treatment.

Several limitations should be considered when interpreting these findings. First, the analysis was based exclusively on PubMed, and therefore publications indexed in other databases may not have been captured. Second, the search strategy used the specific combination “gene mutation AND thalassemia,” which may have excluded relevant studies using alternative terminology such as genetic variant, mutation spectrum, molecular characterization, or specific globin-gene terminology without explicitly using the term gene mutation. Third, bibliometric indicators reflect patterns of publication and terminology rather than the methodological quality or clinical impact of individual studies. Finally, keyword-based thematic and co-occurrence analyses are dependent on the metadata available in the retrieved records and may be influenced by differences in indexing and terminology.

Future research should therefore extend bibliometric analyses across multiple databases and employ more comprehensive search strategies incorporating related genetic and genomic terminology. Longitudinal analyses could also examine how research priorities shift from mutation identification toward genomic diagnosis, gene editing, and clinical translation. Particular attention should be given to the integration of population-specific mutation databases, artificial intelligence-assisted variant interpretation, genomic carrier screening, and precision genetic counseling. Such developments may help translate the

rapidly expanding molecular knowledge of thalassemia into more effective prevention and personalized care strategies.

Conclusion

This bibliometric analysis demonstrates that research on gene mutations in thalassemia has developed into a broad and interconnected field, with sustained scientific productivity and strong contributions from core hematology and genetics journals. The research landscape is primarily centered on mutation, genotype, alleles, sequence analysis, and α- and β-thalassemia, while genetic testing, prenatal diagnosis, gene expression, fetal hemoglobin, and gene editing represent important interconnected research areas. The findings indicate a continuing shift from genetic mutation characterization toward the clinical and translational application of genomic knowledge. Strengthening international collaboration, expanding population-specific genetic evidence, and integrating molecular research with genetic screening, counseling, diagnosis, and emerging genomic therapies may further support the translation of thalassemia genetic research into more effective prevention and personalized care.

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Author Contribution

HS: conceptualization, methodology, data analysis, investigation, and writing of the original manuscript. LLS: data analysis, investigation, and writing of the original manuscript. AF: review, critical revision, and editing of the manuscript. Both authors approved the final version of the manuscript.

Conflict of Interest

The authors declare that they have no conflict of interest.

Ethical Clearance

Ethical clearance was not required for this study because it involved the analysis of bibliographic records from the PubMed database and did not involve human participants, patient-level data, or identifiable personal information.

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