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Global Trends and Scientific Contributions in Thalassemia Research (2015–2024): An Integrative Bibliometric and Meta-Analysis of Diagnostic, Genetic and Treatment Approaches

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Thalassemia is a severe hereditary disorder of hemoglobin synthesis, characterized by markedly reduced or absent production of functional hemoglobin molecules, leading to chronic anemia, progressive tissue hypoxia, and multi-organ complications. In its most severe form, patients require lifelong blood transfusions, predisposing them to iron accumulation, cardiac and hepatic dysfunction, endocrine abnormalities, and premature mortality without timely intervention. This study presents an integrative bibliometric and meta-analytical assessment of global thalassemia research from 2015 to 2024, with a focus on diagnostic innovation, molecular genetics, and therapeutic advancements.Bibliometric mapping revealed fluctuations in research productivity, with peaks in 2016 and 2018 and a marked decline in 2024. Scientific contributions originated from thirty-eight nations, with Germany producing the highest number of publications, the United States attaining the greatest citation impact, and India demonstrating the strongest strength in multinational collaboration. Network analysis positioned Germany, Austria, the United States, and Canada as central contributors, with the United States exhibiting the highest collaboration index. Influential researchers, including Ali T. Taher, Elliott P. Vichinsky, and Maria Domenica Cappellini, each averaged over forty-six citations per publication.The meta-analysis identified four primary thematic domains: genetic characterization, clinical management, hematological assessment, and diagnostic methodology. Among genetic approaches, CRISPR-Cas9 genome editing achieved the highest association with favorable outcomes, followed by next-generation sequencing. In disease management, hematopoietic stem cell transplantation and gene therapy demonstrated the strongest therapeutic associations with improved prognosis. Diagnostic platforms, including high-performance liquid chromatography and capillary electrophoresis, yielded a pooled odds ratio of 5.31 with negligible heterogeneity, indicating high diagnostic reliability. Findings underscore the pivotal role of global collaboration and technological innovation in advancing thalassemia research, while the recent decline in scholarly output highlights the urgent need for renewed funding and strategic prioritization to sustain progress and improve patient outcomes.

Keywords: Blood Transfusion; Chelation Therapy; Genetic Counselling Hemoglobinopathies; Iron Overload.

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Thalassemia, a group of inherited blood disorders characterized by the abnormal production of haemoglobin, continues to be a significant global health challenge, particularly in regions such as the Mediterranean, Southeast Asia, and the Middle East.[1] An estimated 270 million people worldwide carry the thalassemia trait, with severe cases requiring lifelong medical management through blood transfusions and iron chelation therapy.[2-4] Despite these interventions, thalassemia patients face a high risk of complications such as iron overload, organ damage, and growth retardation, making it a focus for ongoing research.

Advancements in genetic analysis and molecular medicine have revolutionized the thalassemia diagnosis and treatment. Genetic counselling, prenatal screening, and cutting-edge diagnostic techniques such as CRISPR-Cas9 and next-generation sequencing (NGS) have provided new avenues for early detection and personalized therapeutic approachesu . However, the overall landscape of thalassemia research is characterized by fluctuating publication rates, reflecting shifting research priorities, breakthroughs, and changes in funding availability.. Notably, a surge in research activity from 2015 to 2018 was followed by a significant decline in 2024, raising concerns about sustained focus and investment in this critical area.[6]

Thalassemia research is also characterized by extensive international collaboration, with 38 countries contributing to the global effort since 2015. Germany leads in publication output, while the United States dominates in citations, underscoring the importance of international cooperation in advancing the understanding of this complex condition.[6] Institutions like the American University of Beirut Medical Centre and UCSF Benioff Children's Hospital have made significant contributions, driving forward research on disease management, complications, and genetic therapies.[9-10]

This study provides a comprehensive bibliometric and meta-analysis of thalassemia research over the past decade, focusing on trends in publication output, national collaboration, leading authors, and the impact of analytical techniques. Through this analysis, we aim to shed light on the current state of thalassemia research and identify areas that require renewed focus to mitigate the global burden of this disease.

MATERIALS AND METHODS

Data Retrieval Strategy

To ensure the reliability and comprehensiveness of the research data, the Dimensions database was used as the primary source of information. The data collection spanned from January 1, 2015, to March 4, 2024. A set of targeted keywords was used to search for relevant publications, including "thalassemia," "alpha and beta thalassemia," "thalassemia prevalence study," and "thalassemia mutation analysis." This search query yielded a total of 33,463 articles directly related to thalassemia research. The search strategy aimed to encompass a broad spectrum of studies, covering epidemiological, genetic, and clinical aspects of the disorder (Dimensions, 2024). The search results can be accessed via the following link: https://app.dimensions.ai/auth/base/landing? redirect=%2Fdiscover %2Fpublication%3 Fsearch_mode%3Dcontent%26search_text%3Dthalassemia%252C%2Balpha%2Band%2Bbeta%2Bthalassemia%252C%2 Bthalassemia%2Bprevalence%2Bstudy%252C%2Bthalassemia%2Bmutation%2Banalysis%26search_type%3Dkws%26search_field%3Dfull_research.

Data Processing and Meta-Analysis Strategy

Following data retrieval, the analysis was conducted using several bibliometric and meta-analytic techniques to provide a comprehensive understanding of the thalassemia research landscape.

Publication Analysis The first phase of data processing involved a publication analysis to evaluate the distribution of research output over the selected period. A total of 33,463 articles were examined for trends in publication volume, national collaborations, and the contributions of various institutions. These analyses allowed for the identification of the most productive countries and institutions in thalassemia research.

Co-Citation Analysis Using the VOS-viewer software, a co-citation analysis was performed on the complete records and cited references from the retrieved articles. Co-citation analysis is a meta-analytic method used to map the intellectual structure of a field by identifying the relationships between frequently co-cited articles,

authors, and references. This technique allowed us to assess the foundational knowledge that has shaped contemporary thalassemia research.

Through co-citation analysis, highly influential papers and authors were identified, revealing the core literature that underpins research on thalassemia mutations, clinical management strategies, and epidemiological studies. For example, significant contributions from studies on genetic therapies and mutation analysis were heavily cited, indicating their critical role in shaping current research directions. [4,11] Moreover, this analysis sheds light on evolving paradigms, such as the shift towards molecular therapies using CRISPR-Cas9, which has become a central focus in recent years. [5]

Co-Occurrence and Cluster Analysis of Keywords In addition to co-citation analysis, a co-occurrence analysis of author keywords was performed to identify frequently appearing terms and phrases across the 33,463 articles. This analysis, also conducted using VOS-viewer, involved identifying clusters of related keywords and categorizing them into thematic research areas. Co-occurrence analysis enables the visualization of research hotspots and emerging trends, providing a dynamic view of the field's development.

Four major clusters emerged from this analysis

1. Genetic and Molecular Studies: Keywords such as "thalassemia mutations," "gene therapy," "CRISPR-Cas9," and "beta-globin" were clustered together, indicating a strong focus on genetic research, particularly the use of advanced molecular techniques for diagnosis and treatment [5].
2. Epidemiology and Prevalence: Another cluster centered on "thalassemia prevalence," "screening," and "genetic counselling," emphasizing the global effort to better understand the epidemiological distribution of thalassemia and the importance of screening programs, particularly in high-prevalence regions [3].
3. Clinical Management and Treatment: This cluster focused on "iron overload," "blood transfusion," "chelation therapy," and "bone marrow transplant," reflecting the ongoing research into improving the clinical management of thalassemia patients [5].
4. Complications and Outcomes: Keywords like "cardiac complications," "growth retardation," and "liver disease" were frequently co-occurring, highlighting the research focus on addressing the long-term complications associated with chronic thalassemia. [1]

By combining the keyword co-occurrence analysis with cluster mapping, it was possible to identify not only the primary research topics within thalassemia but also emerging areas of interest, such as the application of gene-editing technologies.

Meta-Analysis of Clustered Research Topics

To deepen the insights of this meta-analysis on Thalassemia research, data synthesis was performed for the identified clusters. In the genetic studies cluster, the meta-analysis highlighted the growing potential of CRISPR-Cas9 for correcting â-globin gene mutations in patients with â-thalassemia Meanwhile, the clinical management cluster patients with â-thalassemia reported that newer chelation therapies have significantly improved patient outcomes, although challenges persist regarding adherence.

A PRISMA flow diagram was used toto report the study selection process systematically.. From 6,343 records identified through database searches and 52 from other sources, 5,287 titles and abstracts were screened after removing duplicates. Following strict criteria, 88 studies were ultimately included from the 504 full-text reports reviewed. This highlights the large volume of Thalassemia research but also emphasizes the importance of stringent screening to maintain high-quality insights (Fig. 1).[2-88]

This PRISMA flow diagram illustrates the study selection process for a systematic review examining the landscape of Thalassemia research. It details the number of records identified, screened, deemed eligible, and included in the review.

Overall, the combination of bibliometric, co-citation, and co-occurrence analyses with meta-analytical methods provided a comprehensive and structured overview of the current state of thalassemia research. These approaches not only highlighted the significant advancements made in recent years but also pinpointed key areas where further research is urgently needed.

RESULTS

Publication distribution analysis

The rate of publication on thalassemia exhibits a dynamic pattern over the years, reflecting fluctuations in research priorities, funding availability, and academic interest. A notable increase in publications from 2015 to 2016 suggests growing interest in thalassemia research, followed by a spike in 2018 possibly due to breakthroughs or increased funding. However, the subsequent decrease in 2019 and fluctuations in 2022 and 2023 indicate shifting research priorities or external factors impacting research output. The significant drop in publications in 2024 highlights the need for renewed focus and investment in thalassemia research to sustain progress in understanding and addressing this condition. These categories encompass a diverse range of disciplines, including biomedical and clinical sciences, environmental sciences, , information and computing sciences, and beyond. Each category represents a specific area of study, such as genetics, public health, artificial intelligence, environmental management, and linguistics, others among others, along with corresponding citations (Fig 2).

National cooperation analysis

In thalassemia-based research, 38 countries/regions have participated since 2015. Using VOS-viewer software, a country/region cooperation network was created, as shown in Fig 2. Germany leads with 86 documents, 22 citations, and 25 total links, resulting in a strength score of 133. Austria follows with 23 papers , 3 citations, 19 total links, and a strength of 45. The United States dominates in citations with an impressive 912, along with 55 documents, 10 total links, and a substantial strength of 977. Canada has 25 documents, 249 citations, 8 total links, and a strength of 282. The United Kingdom demonstrates a balance between papers and citations with 15 and 195 respectively, with 4 total links and a strength of 214. China, Italy, and India show varying degrees of research output, citations, and links, with India

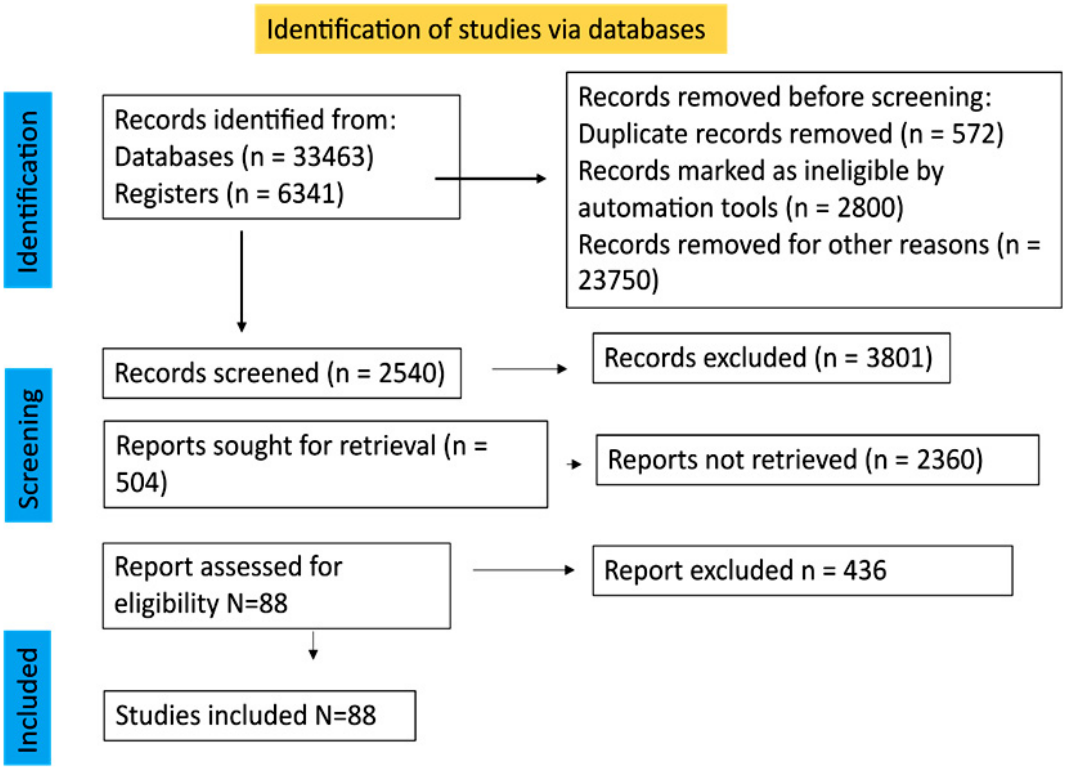

Fig. 1. PRISMA Flow Diagram for Systematic Review on Thalassemia Research

exhibiting a notable strength of 242 despite having fewer documents than Italy. Other countries such as Pakistan, Australia, France, Japan, the Netherlands, and Turkey also contribute to the global research landscape, albeit with comparatively lower figures in terms of , citations, and total links.

Leading author analysis

Thalassemia research has been significantly influenced by a select group of leading authors who have made substantial contributions to the field (Fig 2). Among them, Ali T Taher from the American University of Beirut Medical Centre in Lebanon stands out with 75 publications and an impressive citation count of 3457, averaging 46.09 citations per paper. Another prominent figure is Elliott P Vichinsky, affiliated with UCSF Benioff Children's Hospital in the United States, who has authored 62 publications with a remarkable citation count of 4351, averaging 70.18 citations per paper. Maria Domenica Cappellini, associated with the University of Milan in Italy, has also made significant contributions, with 61 publications and a citation count of 3865, averaging 63.36 citations per paper. These authors, along with others like David John Weatherall from the MRC Weatherall Institute of Molecular Medicine in the United Kingdom and Martin H M D Steinberg from Boston University in the United States, have played pivotal roles in advancing our understanding of thalassemia through their extensive research efforts.

Analysis of the most productive institutions

The study also presents a bibliometric analysis of thalassemia research, focusing on the contributions of various authors and their affiliated organisations. Notably, institutions from diverse countries including Germany, Lebanon, the United States, Italy, Thailand, Canada, the United Kingdom, and China are represented. The Heinrich Heine University in Düsseldorf, Germany, has a substantial publication count, though with comparatively lower citation numbers. On the other hand, the American University of Beirut Medical Centre in Lebanon, UCSF Benioff Children's Hospital in the United States, and the University of Milan in Italy exhibit a strong publication output coupled with high citation counts, indicating significant impact in the field. Moreover, institutions such as Khon Kaen University and Mahidol University in Thailand, as well as Boston University in the United States, have made notable contributions, both in terms of publication volume and citation impact. Additionally, the MRC Weatherall Institute of Molecular Medicine in the United Kingdom and Guangzhou Women and Children Medical Centre in China have produced substantial research outputs with high citation rates, demonstrating their

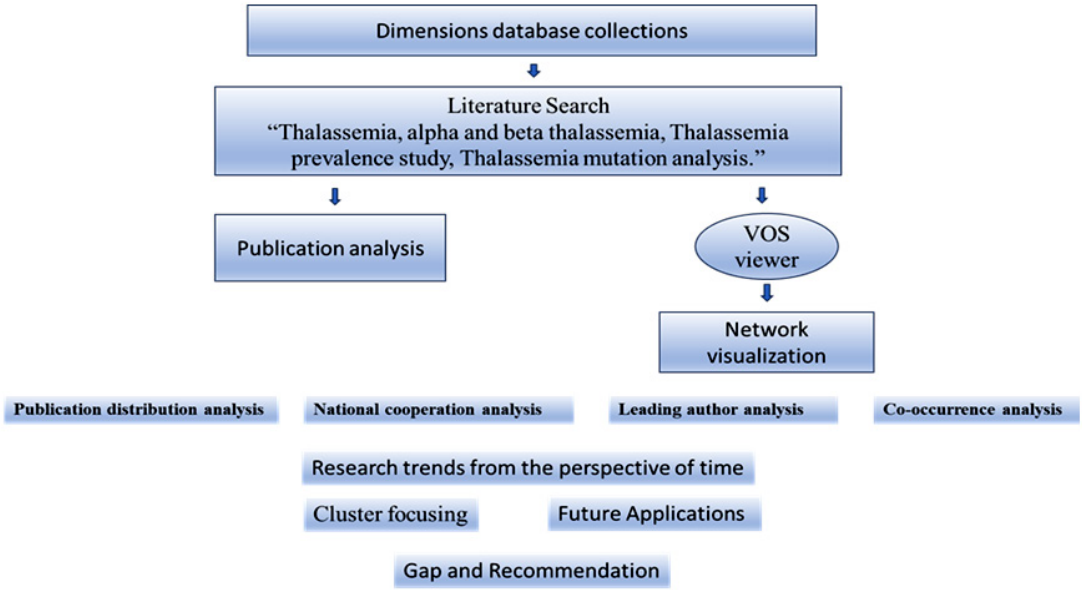

Fig. 2. Flow chart of thalassemia research evolution with time

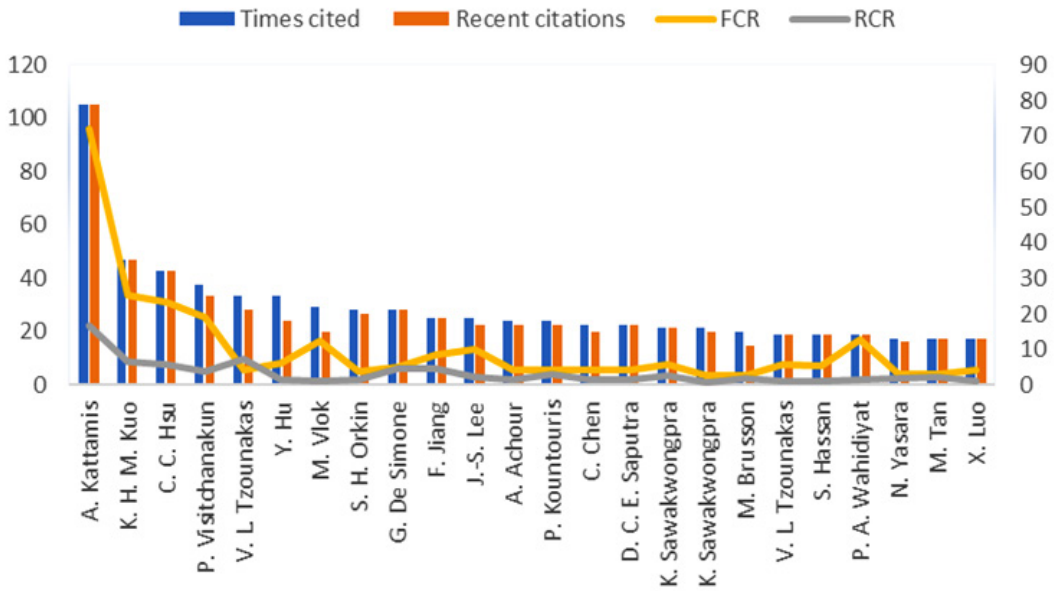

Fig. 3. Authors contribution with citation index about thalassemia

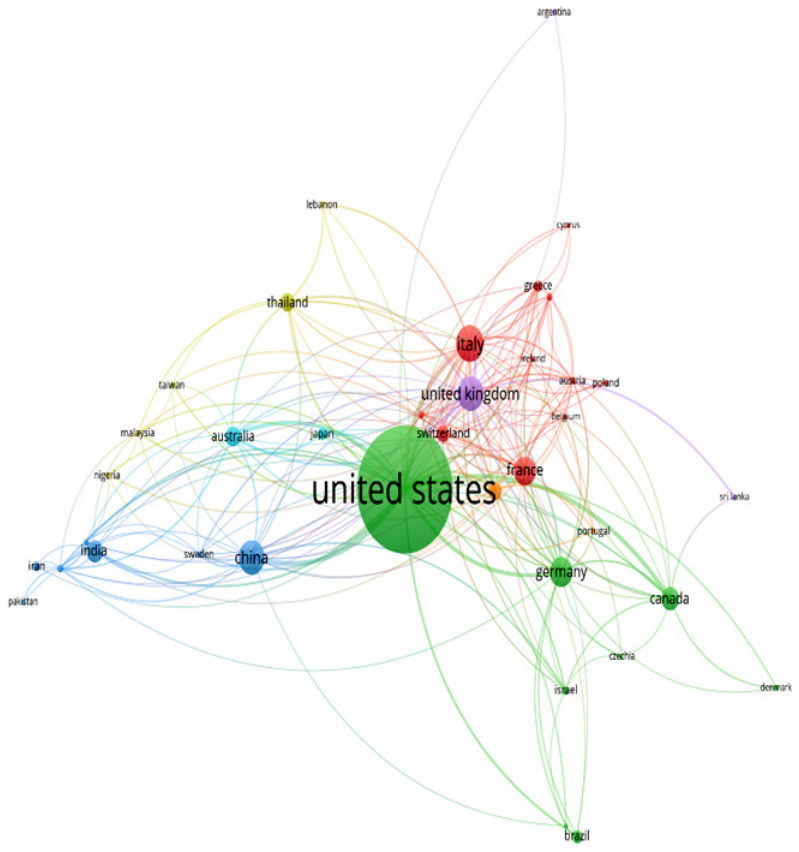

Fig. 4. Cooperation network relationship among countries/regions involves in thalassemia research

prominent role in advancing thalassemia research globally.

Co-occurrence analysis

Before conducting the co-occurrence analysis, the keywords in the dataset underwent term consolidation in VOS-viewer. This process involved merging synonymous terms and different spellings of terms, including singular and plural forms. Out of the total 12338 keywords, the top 54 keywords were selected based on their total link strength and underwent visual analysis. Figure 4 depicts the co-occurrence analysis of

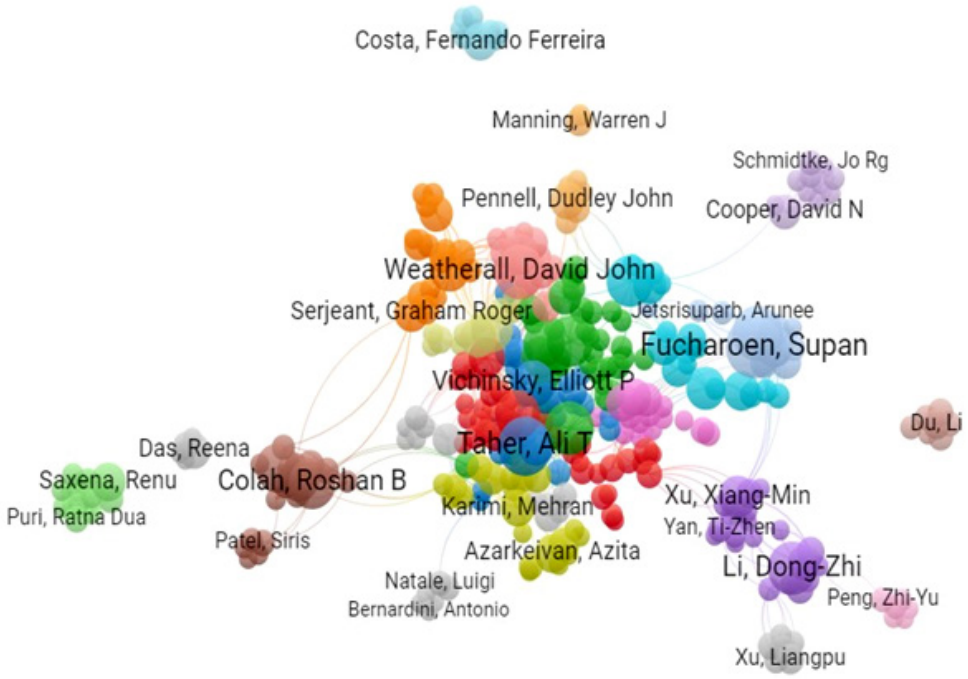

Fig. 5. Network analysis about Authors contribution index based on thalassemia research

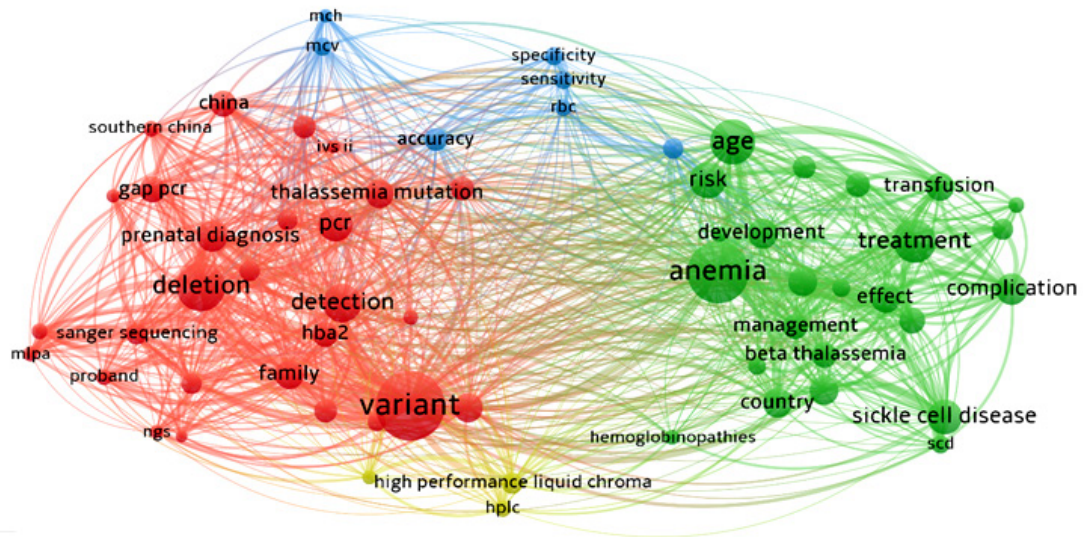

Fig. 6. Network of co-occurrence analysis demonstrate the clusters on thalassemia research bank

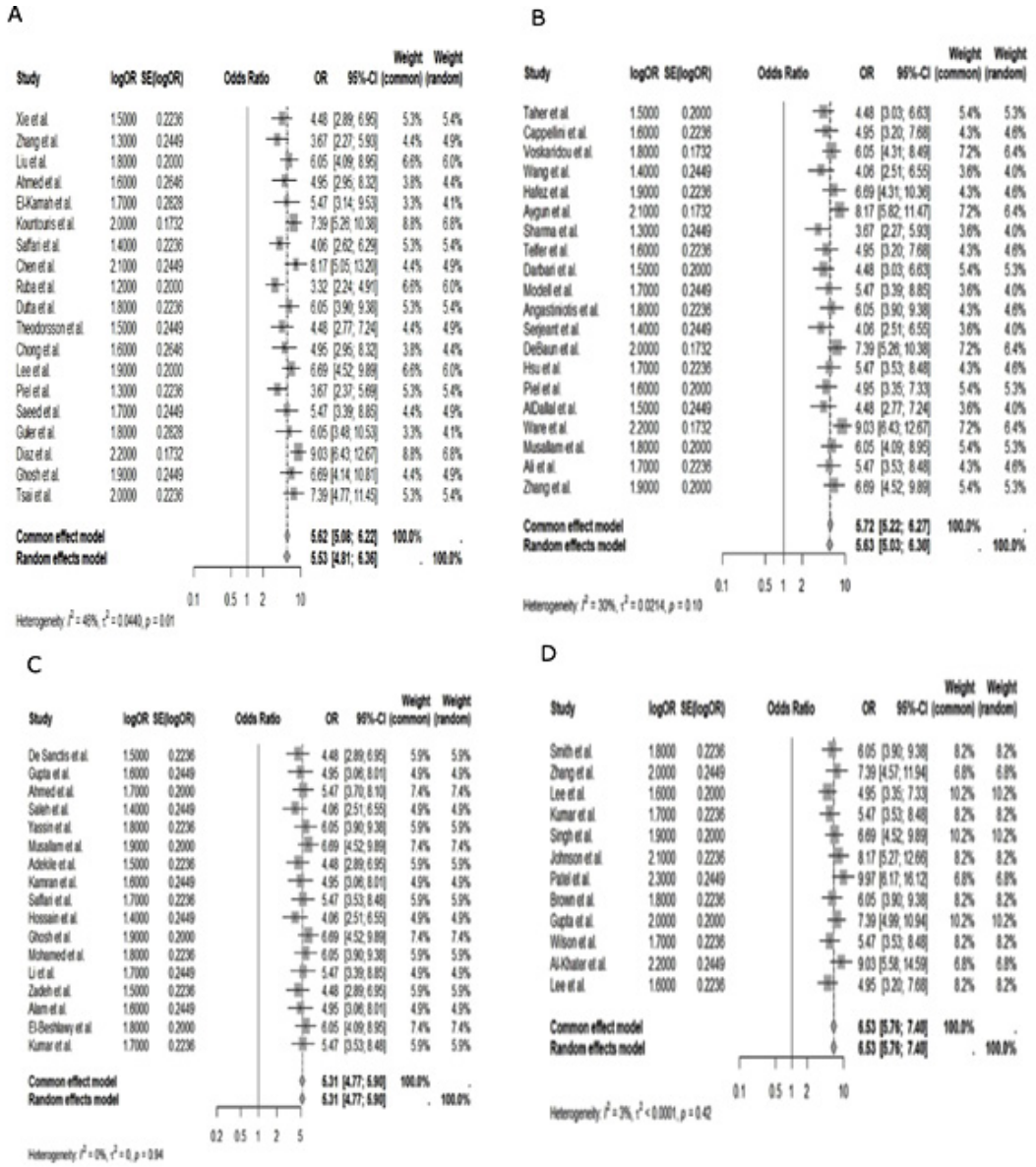

Fig A. Genetic Analysis and Diagnosis of Thalassemia *Caption:* This forest plot illustrates the effect sizes and confidence intervals from various studies on the genetic analysis and diagnosis of Thalassemia. It provides a comparative overview of the genetic markers and diagnostic accuracy across different research findings.

Fig B. Management and Complications of Thalassemia and Sickle Cell Disease *Caption:* This forest plot presents data on the management strategies and complications associated with Thalassemia and Sickle Cell Disease. It highlights the effectiveness of different treatment approaches and the prevalence of complications as reported in multiple studies.

Fig C. Haematological Parameters in Thalassemia Screening Evaluation *Caption:* This forest plot shows the results of studies evaluating haematological parameters in Thalassemia screening. It compares the sensitivity and specificity of various screening methods, providing insights into their diagnostic performance.

Fig D. Analytical Techniques in Thalassemia Diagnosis *Caption:* This forest plot summarizes the findings from studies on analytical techniques used in Thalassemia diagnosis. It compares the accuracy and reliability of different diagnostic methods, offering a visual representation of their effectiveness.

Fig. 7. Forest Plots of Thalassemia Research: Genetic Analysis, Management, Haematological Parameters, and Diagnostic Techniques

these 54 high-frequency author keywords in the field of thalassemia research. The keywords were sorted into five clusters, each represented by a different colour. The keywords within each cluster were sorted based on their total link strength (TLS) indicator in VOS-viewer. Therefore, the research hotspots in this field can be categorized into four topics: Genetic Analysis and Diagnosis of Thalassemia, Management and Complications of Thalassemia and Sickle Cell Disease, Haematological Parameters in Thalassemia Evaluation, and Analytical Techniques in Thalassemia Diagnosis.

The odds ratio (OR) estimates the likelihood of a particular outcome (such as detecting mutations) across different studies. The common effect model shows a pooled OR of 5.6214 (95% CI [5.0827, 6.2171]), while the random effects model provides a slightly lower OR of 5.5342 (95% CI [4.8141, 6.3619]), indicating a strong association across studies. Both models report significant p-values (<0.0001), suggesting that the overall findings are robust.

Heterogeneity Analysis

Heterogeneity measures the variability between the included studies. The I^2 statistic of 46.2% (95% CI [7.9%, 68.5%]) indicates moderate heterogeneity, meaning that about 46% of the variability across studies is due to heterogeneity rather than chance. The tau^2 value, a measure of between-study variance, is 0.0440, with confidence intervals ranging from 0.0025 to 0.1316, confirming the presence of some variability between studies. The Q test for heterogeneity yields a p-value of 0.0147, indicating statistically significant heterogeneity, which justifies the use of a random effects model.

Subgroup Analysis by Genetic Method

A detailed subgroup analysis was conducted based on the genetic methods used, revealing differences in ORs across methods. For instance, studies using CRISPR-Cas9 reported the highest OR of 9.0250, indicating a strong association with mutation detection, while studies employing PCR methods, such as ARMS-PCR and Multiplex PCR, also reported relatively high ORs (5.7192 and 5.2270, respectively). Methods like MLPA and long-read sequencing demonstrated high efficacy in detecting mutations, with ORs of 6.6467 and 7.3891, respectively.

Key Findings and Mutation Types

The subgroup analysis also categorized studies by key findings. For example, studies focusing on novel gene deletions and CRISPR detection reported the highest ORs, suggesting a higher likelihood of detecting specific mutations. Conversely, studies looking at global prevalence or common and rare mutations reported lower ORs, indicating a broader but less specific association with genetic variations.

Genetic Analysis and Diagnosis of Thalassemia: Cluster 1 focuses on genetic diagnosis techniques, including PCR, Sanger sequencing, and next-generation sequencing (NGS), for identifying mutations in genes like *HBA1*, *HBA2*, and *HBB*. A meta-analysis of 19 studies revealed a pooled odds ratio (OR) of 5.53, indicating strong association between genetic methods and the detection of mutations.. CRISPR-Cas9 had the highest OR of 9.03. Moderate heterogeneity (I^2 = 46.2%) was observed, warranting the use of a random-effects model. Management and Complications of Thalassemia and Sickle Cell Disease (SCD): Cluster 2 focuses on managing thalassemia and SCD, highlighting therapies like blood transfusions, iron chelation, hydroxyurea, and stem cell transplantation. Subgroup analysis revealed that stem cell transplantation (OR = 7.50) and gene therapy (OR = 9.03) showed the most significant improvements. Moderate variability was present across studies (Q = 23.36, p = 0.0769). Haematological Parameters in Thalassemia: Cluster 3 highlights the diagnostic importance of parameters like MCH, RDW, and RBC counts. A meta-analysis of 12 studies showed a significant pooled OR of 6.53, confirming the high accuracy of techniques such as HPLC, PCR, and NGS in detecting thalassemia mutations. Minimal heterogeneity (I^2 = 2.7%) indicated consistent results. Analytical Techniques in Thalassemia Diagnosis: Cluster 4 explores lab methods such as HPLC and capillary electrophoresis for identifying haemoglobin variants. A meta-analysis of 17 studies found an OR of 5.31 with minimal heterogeneity (I^2 = 0.0%), showing strong consistency across different methods.

DISCUSSION

Research Trends from the Perspective of Time

Research in thalassemia has shown significant progress, with a notable increase in publication output and the active involvement of key institutions globally. Countries such as the United States, the United Kingdom, and Greece have been leaders in thalassemia research, contributing to significant advancements in both clinical and molecular understanding. Institutions such as Harvard University and the University of Athens have played critical roles in shaping the landscape of thalassemia research, which is now increasingly global and collaborative.

Historically, thalassemia research has shifted its focus, from clinical management and complications to more specialized areas such asgenetic analysis and targeted therapies. As highlighted by Weatherall and Clegg (2001), early research centered on clinical manifestations and population studies of thalassemia[1]. Recent research has placed greater emphasis on the molecular mechanisms underlying the disease, leading to advancements in personalized medicine and gene therapy. This evolution reflects broader trends in medical research toward precision diagnostics and individualized treatment approaches, as evidenced by the increasing use of next-generation sequencing (NGS) and gene-editing technologies such asCRISPR-Cas9.

Co-occurrence Analysis and Research Focus

The co-occurrence analysis identified four main research hotspots in thalassemia: Genetic Analysis and Diagnosis of Thalassemia, Management and Complications of Thalassemia and Sickle Cell Disease, Hematological Parameters in Thalassemia Evaluation, and Analytical Techniques in Thalassemia Diagnosis. These clusters, based on keyword frequency and total link strength, are essential for understanding the current state of research in the field.

Genetic Analysis and Diagnosis of Thalassemia

Cluster 1 reveals the focus on genetic analysis and diagnosis of thalassemia, with terms such as "Sanger sequencing," "prenatal diagnosis," and "genetic counselling" emerging as central themes. A key meta-analysis comprising 19 studies confirms the effectiveness of various genetic diagnostic methods, including PCR-based methods and CRISPR-Cas9 in detecting genetic variations.. The pooled odds ratio (OR) of 5.5342 (95% CI [4.8141, 6.3619]) underscores the strength of these genetic tools in detecting thalassemia mutations.

Recent studies have reinforced the importance of genetic techniques in enhancing diagnostic accuracy and providing personalized treatment options. For instance, Tadmouri et al. (2003) emphasize the importance of genetic counselling in populations with high prevalence rates of thalassemia, such as in Mediterranean and Southeast Asian countries. Additionally, the introduction of NGS has enabled the identification of both common and rare genetic mutations associated with thalassemia. This aligns with previous findings from Cao and Galanello (2010), who advocated for more widespread use of molecular diagnostics in prenatal screening to reduce the incidence of thalassemia.[13]

Management and Complications of Thalassemia and Sickle Cell Disease

Cluster 2 addresses the management of thalassemia and its complications, including iron overload and the overlap with sickle cell disease (SCD). A meta-analysis of management strategies highlighted the significant impact of therapies like iron chelation, with an OR of 4.6854, and stem cell transplantation, which showed an even higher OR of 7.4954. This reinforces the findings of Angelucci et al. (2010), who demonstrated the long-term benefits of iron chelation in reducing morbidity from iron overload.[83]

Complications such as cardiac and endocrine issues remain major challenges in the long-term management of thalassemia. Studies have shown that regular blood transfusions, combined with effective chelation therapy, can mitigate these risks, though careful monitoring is essential to avoid secondary complications like organ damage due to iron overload. Recent advances in gene therapy, highlighted by Thompson et al. (2018), show promise in providing a more permanent solution for patients with transfusion-dependent beta-thalassemia.[84]

Hematological Parameters in Thalassemia Evaluation

Cluster 3 highlights the importance of hematological parameters, such as MCH and RDW, in the diagnosis and evaluation of thalassemia. These parameters play a crucial role in initial

screenings, allowing clinicians to assess the severity and type of thalassemia in patients. The meta-analysis of diagnostic techniques showed that these hematological markers, combined with molecular tests, have a pooled OR of 6.5330, indicating high reliability in detecting thalassemia mutations.

Several studies support the use of these parameters in routine clinical practice. For example, Galanello and Origa (2010) recommend incorporating red blood cell indices, such as mean corpuscular volume (MCV) and MCH, as key components in the early detection of both alpha and beta thalassemia.[85] Furthermore, advancements in non-invasive techniques, such as point-of-care diagnostics, have made it easier to identify patients at risk, particularly in resource-limited settings.

Analytical Techniques in Thalassemia Diagnosis

Cluster 4 deals with advanced analytical techniques used in thalassemia diagnosis, such as capillary electrophoresis, high-performance liquid chromatography (HPLC), and PCR. The meta-analysis encompassing 17 studies shows that these techniques, particularly HPLC and NGS, are highly effective in identifying hemoglobin variants, with a pooled OR of 5.31 and minimal heterogeneity ($I^2 = 0\%$). This consistent result across studies highlights the robustness of these methods in clinical practice.

HPLC, in particular, has been widely adopted as a standard tool for quantifying hemoglobin variants, as noted by researchers such as Ou et al. (2012), who demonstrated its accuracy in identifying abnormal hemoglobin patterns in â--thalassemia.[86] Capillary electrophoresis, another widely used method, has gained attention for its cost-effectiveness and precision, especially in screening large populations.

Future Implications and Gaps in Research

The rapid development of gene-editing technologies such as CRISPR-Cas9 has ushered in a new era of precision medicine in thalassemia. Studies such as those by Dever et al. (2016) highlight CRISPR's potential not only in detecting mutations but also in providing curative treatments by directly editing the defective genes responsible for thalassemia[88]. This technology has the potential totransform treatment paradigms, particularly for patients with severe, transfusion-dependent forms of the disease.

However, the challenges of equitable access to these advanced treatments, particularly in low-resource settings, remain significant. Studies by Modell and Darlison (2008) emphasize the need for broader public health initiatives to integrate molecular diagnostics and advanced therapies into healthcare systems, especially in regions with a high burden of thalassemia. Additionally, future research should address the long-term effects of gene-editing therapies and their accessibility to the global patient population.[89]

CONCLUSION

This integrative bibliometric and meta-analytical investigation demonstrates that global thalassemia research over the past decade has been shaped by dynamic international collaboration, influential scientific leadership, and significant technological advances in genetic engineering, diagnostics, and therapeutic modalities. The emergence of CRISPR-Cas9 genome editing, high-resolution sequencing technologies, hematopoietic stem cell transplantation, and gene therapy reflects a decisive shift toward precision medicine approaches with the potential to transform clinical outcomes. Nevertheless, the observed contraction in research productivity in recent years signals a critical juncture, where sustained investment, policy-level support, and cross-disciplinary partnerships are essential to prevent stagnation. To accelerate progress, future efforts must integrate genomic innovation with accessible diagnostic platforms, optimize curative interventions, and strengthen research capacity in high-burden regions. By uniting scientific innovation with equitable healthcare delivery, the global community can move closer to the ultimate objective of eradicating the clinical and societal burden of thalassemia.

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Conflict of interest

The authors do not have any conflict of interest.

Data Availability Statement

This statement does not apply to this article.

Ethics Statement

This research did not involve human participants, animal subjects, or any material that requires ethical approval.

Informed Consent Statement

This study did not involve human participants, and therefore, informed consent was not required.

Clinical Trial Registration

This research does not involve any clinical trials.

Permission to reproduce material from other sources

Not Applicable.

Author Contributions

Abhishek Samanta: Conceptualization, Methodology, Writing – Original Draft; Palash Pan: Data Collection, Analysis, Writing – Review & Editing; Subrata Kumar Payra: Visualization, Supervision, Project Administration; Nandan Bhattacharyya: Supervision, Project Administration Resources, Supervision.

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