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A Bibliometric Analysis of the Most Cited Articles on Short Stature

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Abstract **Background:** Short stature, defined as height significantly below the population average, can arise from various etiologies, including genetic mutations, hormonal imbalances and chronic illnesses. Despite increasing research, a comprehensive assessment of high-impact studies in this field remains lacking. This bibliometric analysis evaluates emerging research trends, methodological quality and the impact of the most-cited articles on short stature, highlighting strengths and identifying gaps in the literature. **Methods:** The 50 most relevant articles on short stature were organized in a bibliometric format and analyzed using R Studio with the packages "BiblioShiny," "Bibliometrix" and "Meta." The analysis included visualizing publication trends, identifying leading countries, authors and affiliations and calculating total citations. A thematic analysis assessed the frequency of key terms and research focus areas. **Results:** The analysis of the top 50 articles reveals that the USA dominates the publication landscape, contributing 58% of the articles, followed by the UK at 11%. Germany, Italy and the Netherlands contribute 5% each, while Switzerland, England and Japan provide 3% and Iran, Finland and France account for 2%. The studies utilize diverse methodologies, with genetic mutation research comprising 17 studies, alongside case-control studies, observational studies and systematic reviews. Genetic studies focusing on mutations in genes like PTPN11, FGFR3 and FGF23 are prevalent, elucidating conditions such as Noonan syndrome and Turner syndrome. Evidence levels vary, with many studies classified as Level 4, while comprehensive reviews and expert consensus papers are categorized as Level 1. Michael Holzer and Niklas Nielsen are the most productive authors, with citations of 1255 and 1231, respectively. The most prolific journal is "The American Journal of Medical Genetics Part A," which published 17 relevant articles. Research volume has steadily increased over the past two decades, reflecting growing awareness of short stature as a significant public health issue. **Conclusions:** This analysis highlights significant contributions to short stature research, emphasizing the need for better translation of genetic findings into clinical outcomes. Future research should prioritize personalized treatments, advanced diagnostic tools and the psychosocial impacts on affected individuals.

Key Words Short Stature, Growth Disorder, Dwarfism, Height Deficiency

INTRODUCTION

Short stature is a common concern in pediatric healthcare, defined as a height more than two standard deviations below the mean for age and sex [1]. It affects approximately 2.5% of the population and is the most frequent reason for referral to pediatric endocrinologists [2]. The etiology of short stature is diverse, encompassing genetic, endocrine and environmental factors.

Growth Hormone Deficiency (GHD) is a significant cause of short stature, accounting for about 28% of cases in some studies [3]. However, normal variants such as familial short stature and constitutional delay of growth and puberty are also common, comprising up to 26% of cases [3]. Other etiologies include chronic diseases, genetic syndromes and nutritional deficiencies [4].

The evaluation of short stature involves a comprehensive approach, including detailed history, physical examination and auxological measurements [5]. Advanced genetic testing has revolutionized the diagnosis of short stature, revealing numerous genetic causes related to growth plate function and the growth hormone-insulin-like growth factor-1 (GH-IGF-1) axis.

Treatment options vary depending on the underlying cause. Growth hormone therapy is the mainstay for GHD and some other conditions, while addressing nutritional deficiencies or underlying medical conditions may be sufficient in other cases (Growth Hormone Research Society, 2019). However, the use of growth hormone in idiopathic short stature remains controversial [5].

The psychosocial impact of short stature is significant, often affecting quality of life and self-esteem [1]. Therefore, a holistic approach to management, addressing both physical and psychological aspects, is crucial.

Recent advancements in understanding the genetic basis of short stature and the development of new therapeutic approaches, such as long-acting growth hormone preparations, are expected to transform the field in coming years [2].

This bibliometric analysis aims to conduct a systematic study of the body of research surrounding short stature with a focus on publication trends, domains of research and emerging themes. We have attempted to bridge these gaps by Find top-cited articles and trends. Analyze which countries, journals and authors lead. Identify main topics and gaps in the research.

METHODS

The bibliometrics analysis of short stature research involves a systematic approach to ensure thorough evaluation. Initially, a comprehensive literature search is conducted using keywords such as "Short Stature," "Growth Disorders," "Dwarfism" and "Height Deficiency" across databases like PubMed and Web of Science, covering publications from inception until September 2024. Inclusion criteria focus on studies specifically related to short stature that are published in peer-reviewed journals and involve human subjects, while exclusion criteria eliminate non-English articles, non-peer-reviewed sources and studies lacking citations. The selection process utilizes Rayyan Software for managing search results, allowing independent review of titles and abstracts by multiple researchers to ensure accurate study selection. Data extraction follows a standardized form in Excel, capturing essential information such as article titles, authors, publication years, journal names, total citations, average citations per year, research settings, financial support, study designs, strength of evidence, primary topics and the application of Patient-Reported Outcome Measures (PROMs). The analysis critically assesses the methodological rigor and relevance of the selected articles, identifying trends, gaps and inconsistencies in the literature. Finally, findings are summarized to provide a comprehensive overview of research trends and quality, along with recommendations for future research directions based on identified gaps. This structured methodology facilitates a deeper understanding of the strengths and weaknesses within the field of short stature research.

Analytics of the data extracted from the first 50 most relevant short-stature articles were arranged in a bibliometrics format. The data were then imported into R studio using the “read.csv” function and analysis was performed in R studio using the packages “BiblioShiny,” “Bibliometrix” and “Meta.” Exploratory analysis includes a time series plot showing the publication trend, most productive country and authors, authors' affiliations and total citation. Thematic analysis such as the most frequent word analysis was carried out.

Ethical Consideration

The ethical committee of KAIMRC has approved the study with IRB number: NRR25/030/3. There was no conflict of interest or funding for this study.

Reporting Standards

This study adheres to the Strengthening the Reporting of Observational Studies in Epidemiology (STROBE) guidelines for observational studies.

RESULTS

Quality Characteristics of Included Studies

Table 1 presents the study properties of the top 50 leading research in short stature, out of which the USA accounts for 58%, followed by the UK with 11% of the total publications. Countries such as Germany, Italy, Netherlands, account for 5% of each. Switzerland, England and Japan have had 3% of the total research in short stature, with Iran, Finland and France having 2% publications (Figure 1).

The studies are characterized by diverse research methodologies, ranging from genetic mutation accounting for 17 studies, 1 Case-control study, 2 case reports, 7 case series studies, 6 observational studies, 20 systematic reviews and 2 expert consensus (Figure 2). Genetic studies dominate the research landscape, with numerous articles analyzing mutations in genes related to growth, such as PTPN11, FGFR3 and FGF23. These genetic insights are crucial in understanding the pathophysiology behind various forms of short stature, including conditions like Noonan syndrome, Turner syndrome and Silver-Russell syndrome. Many of the studies focus on the genetic basis of these syndromes, providing critical information on how mutations in specific genes, such as those affecting growth hormone receptors or regulatory proteins, contribute to short stature (Table 1).

Table 1: Results of the Characteristics of the top 50 articles in Short Stature with their level of evidence

First author	Year	Country	Settings	Level of Evidence	Study design	Outcome Measure
Tartaglia *et al.* [6]	2001	United States	NM	Level 4	Genetics analysis	Mutations in PTPN11 & role of SHP-2 protein
Mundlos *et al.* [7]	1997	United States	Harvard Medical School and universities in Germany	Level 4	Genetic mutation analysis	CBFA1 mutations associated with Cleidocranial Dysplasia
White et al. [8]	2000	United States	Indiana University, School of Medicine (USA), Ludwig-Maximilians-Universität (Germany)	Level 4	Nature Genetics	Mutations in FGF23 linked to ADHR
Yachie et al. [9]	1999	Japan	Kanazawa University, Japan	Level 4	Case study	Oxidative stress effects
Shiang et al. [10]	1994	United States	University of California, Irvine	Level 4	Genetic mutation analysis	Mutations in FGFR3
Holick [11]	2006	United States	Boston University, Medical Center, Massachusetts, USA	Level 5	Review	The role of vitamin D in preventing rickets
Cassidy et al. [12]	2012	United States	University of California, San Francisco; University of Florida, Gainesville	Level 5	Review	Clinical features.
Woods et al. [13]	1996	United Kingdom	St. Bartholomew's Hospital, London	Level 1	Case study	Intrauterine and postnatal growth failure
Varon [14]	1998	Germany	Charitè ,Berlin	Level 3	Genetic linkage analysis	Identify gene mutation
Hill *et al.* [15]	2005	USA	North America	Level 3	Systematic review	Diagnosis and treatment
Gelb *et al.* [16]	1996	USA	New York,Birgham and women's hospital	Level 4	Genetic study	Mutation in cathepsin K gene
Galanello *et al.* [17]	2010	Italy	Italian medical institutions	Level 3	Narrative review	Genetic mutations
Rao *et al.* [18]	1997	Netherlands	Dutch research institutions	Level 4	Genetic study	Genetic interaction
Prasad *et al.* [19]	1961	iran	Nemazee hospital and saadi hospital,Shiraz	Level 4	Case series study	Syndrome
Zapf *et al.*[20]	1981	Switzerland	Conducted at the university of Zurich	NA	Radioimmunological study	Immune
Gravholt *et al.*[21]	2017	UK	Cincinnati international turner syndrome meeting,USA	Level 3	Systematic review	Diagnosis, treatment and management
de Onis and Branca [22]	2016	Switzerland	Global perspective	Level 5	Review article	Cause and solution
Hästbacka *et al.* [23]	1994	United States.	Primarily based on Finnish genetic population data.	Level 4	genetic association study	Diastrophic dysplasia gene
Godowski *et al.* [24]	1989	United States.	United States and Israel	Level 4	genetic observational study	Gene deletions in the growth hormone
Baxter and Martin [25]	1986	Australia	Royal Prince Alfred Hospital, Camperdown, New South Wales, Australia	NA	Experimental study	RIA for growth hormone
Bione *et al.* [26]	1996	Italy	Primarily Pavia, Italy	Level 4	Genetic stuy	dentifying G4.5 gene's role in Barth syndrome.
Tartaglia *et al.* [27]	2002	United States	Various international locations	Level 4	Genetic study	PTPN11 mutations in Noonan Syndrome.
Schubbert *et al.* [28]	2006	United States	United States (California), Germany, and the Netherlands.	Level 4	Genetic analysis	Germline KRAS mutations as causative in Noonan syndrome
Roberts *et al.* [29]	2013	United States	Multiple international locations including the United States, Canada and Italy.	Level 5	Review article	Noonan syndrome
Pandit *et al.* [30]	2007	UK	E.O. Lawrence Berkeley, National Laboratory	Level 4	Genetic linkage	Genetic mutation
Embleton *et al.* [31]	2001	USA	Newcastle on Tyne	Level 2	observational study	Nutritional care
Karlberg and Albertsson-Wikland [32]	1995	USA	Goteborg	Level 3	Retrospective cohort	final height
Schipani *et al.* [33]	1995	USA	not mentioned	Level 4	genetic linkage and mutation analysis	Genetic mutation

Table 1: Continue

Prendergast and Humphrey [34]	2014	UK	Global perspective	Level 5	Review article	Overview of stunting
Clayton *et al.* [35]	2007	USA	Global perspective	Level 5	Expert consensus	Treatment
Tavormina *et al.* [36]	1995	UK	New Jersey	Level 4	genetic linkage and mutation analysis	Mutations may cause TD
Cohen *et al.* [37]	2008	USA	Global perspective	Level 5	review	treatment
Spiliotis *et al.* [38]	1984	USA	Study of growth hormone secretion in short children with GH neurosecretory dysfunction	observationa l evidence	Observational study	Neurosecretory dysfunction
Silventoinen *et al.* [39]	2003	Finland	Twin cohorts from eight different countries: Australia, Denmark, Finland, Norway, Sweden, UK, and the United States.	3 observe and comperative	Comparative study	Heritability of adult body height.
Abuzzzahab *et al.* [40]	2003	USA	• Cincinnati Children's, Hospital Medical Center, Cincinnati, Ohio, USA • Hospital for Children and Adolescents, University of Leipzig, Leipzig, Germany	Level 4	Case report	IGF-I receptor mutations
Tartaglia *et al.* [41]	2007	Italy	Dipartimento di Biologia Cellulare e Neuroscienze, Istituto Superiore di Sanità, Rome, Italy	Level 3	Case control design	Mutations in the SOS1 gene
Horwitz [42]	2001	United States	St. Jude Children's Research Hospital in Memphis, Tennessee, United States	Level 4	Case series design	linear growth, bone mineralization, and fracture rate
Grimberg *et al.* [43]	2016	United States	Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA, USA	Unknown	Systematic review	Primary insulin-like growth factorI deficiency
Briggs *et al.* [44]	1995	USA	United States (Ahmanson Department of Pediatrics, Steven Spielberg Pediatric Research Center, Cedars-Sinai Research Institute, Los Angeles, California)	Maybe 3	Case studies	Mutations in the cartilage oligomeric matrix protein (COMP)
van der Eerden *et al.* [45]	2003	Netherlands	Leiden University Medical Center, Leiden, The Netherlands	Maybe 4	review article	Factors influencing chondrocyte proliferation and differentiation
Novelli *et al.* [46]	2002	United States	University of Rome "Tor Vergata"	Level 3	genetic linkage	Mandibuloacral Dysplasia (MAD)/identification of (R527H) in the LMNA gene
Romano *et al.* [47]	2010	United States	New York Medical College, Valhalla, NY	Level 5	Expert consensus	Genetic findings and multidisciplinary care recommendations.
Spence *et al.* [48]	1988	United States	Baylor College of Medicine, Houston, Texas	Level 4	Case study	Uniparental disomy.
Rosenfeld *et al.* [49]	1994	United States	Doernbecher Memorial Hospital for Children, Oregon Health Sciences University, Portland, Oregon	Level 4	Review	Growth Hormone (GH) Insensitivity due to Primary GH Receptor Deficiency.
Baumann [50]	1991	United States	Northwestern University Medical School	Level 4	Review article	Growth hormone (GH) heterogeneity
Hennekam [51]	2006	United Kingdom	Clinical and Molecular Genetics Unit, Institute of Child Health, Great Ormond Street Hospital, London, UK	Level 4	Review	Hutchinson-Gilford Progeria Syndrome (HGPS)
Niikawa *et al.* [52]	1981	Japan	Department of Pediatrics, Hokkaido University School of Medicine, Sapporo, Japan	Level 4	clinical observational study.	Kabuki make-up syndrome
Hokken-Koelega *et al.* [53]	1995	Netherlands	Sophia Children's Hospital, Erasmus University Rotterdam	Level 2	Retrospective cohort	Postnatal growth of infants born small for gestational age (SGA)

Table 1: Continue

Gicquel *et al.* [54]	2005	France	Laboratoire d'Explorations Fonctionnelles Endocriniennes, Inserm U515 et UPMC Paris 6, Hôpital Armand Trousseau, APHP, Paris	Level 3	Observational study	Epigenetic Mechanisms in SilverRussell Syndrome (SRS)
Angulo *et al.* [55]	2015	United States	Winthrop University Hospital	Level 5	literature review	Prader-Willi syndrome (PWS), focusing on its clinical, genetic, and endocrine findings
Horton *et al.* [56]	2007	United Kingdom	Research Center, Shriners Hospital for Children, Portland, Oregon, USA (William A. Horton)	Level 5	seminar-style review	The most common form of shortlimb dwarfism in humans
Hofman *et al.* [57]	1997	USA	New Zealend	Level 2	Controlled experimental	Etiology, Complication
Lassarre *et al.* [58]	1991	England	France	Level 2	Controlled observational	Etiology, Complication
Razzaque *et al.* [59]	2007	Germany	Tokyo	Level 4	In vitro experiment study	Genetic
Saenger *et al.* [60]	2007	USA	NA	Level 1	Comprehensive Review	Complication
Ahn *et al.* [61]	1995	Germany	Housten	Level 4	Case Reports	Genetic
El-Hattab *et al.* [62]	2015	USA	NA	Level 4	Comprehensive Review	Syndrome
Sandstea *et al.* [63]	1967	USA	NA	Level 2	Prospective cohort	Complication, therapeutic
Holick [64]	2005	USA	Bethesda Maryland	Level 1	Comprehensive Review	Complication, therapeutic
Martorell *et al.* [65]	1994	England	London	Level 1	Comprehensive Review	Therapeutic
Patten *et al.* [66]	1990	USA	Maryland	Level 4	Genetic mutation analysis	Genetic
Prasad *et al.* [67]	1963	USA	Iran	Level 4	Case Series	Complication

The evidence levels vary, with many studies falling under Level 4, which includes genetic studies and case reports, while some high-quality studies are categorized as Level 1, such as comprehensive reviews and expert consensus papers. These Level 1 studies provide broader overviews, offering a synthesis of research findings and clinical recommendations for managing short stature (Table 1).

Analyses of the Annual Scientific Production and Growth

Figure 3 shows the Annual scientific production of research related to short stature between 1961 and 2023, by examining the trend, there has been a steady increase in research volume over the past two decades, particularly since the early 90s. This increase corresponds to growing awareness of research on short stature as a significant public health issue, causing controversy on the causes and appropriate interventions to reduce the prevalence. This trend is particularly evident in countries with Advanced healthcare infrastructures such as the two most productive countries (USA and UK) (Figure 1), where large literature review and systematic review studies have become more feasible. As shown in the graph, research on short stature was at its peak in 1993 and 2004, indicating that research on short stature is receiving increasing attention from academic researchers (Figure 3).

Analyses of the Most Productive Author's Country in the Research on Short Stature

According to Table 2, the most performing author is MICHAEL HOLZER with a total citation of 1255 and an average citation per year of 52.29, followed by the author NIKLAS NIELSEN has 1231 TC with an average total citations per year of 43.96. These two authors are from the United States, meaning that, the United States of America is the leading country with 58% of publications (Figure 1) and authors in the USA published more articles than authors in other countries. The United Kingdom took the

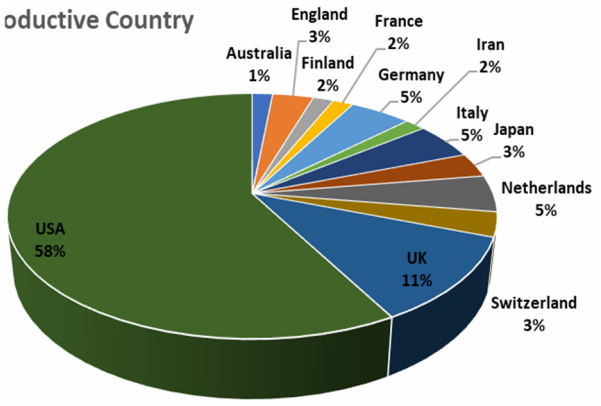


Figure 1: Pie Chart distribution of the Most Productive Country in short stature research

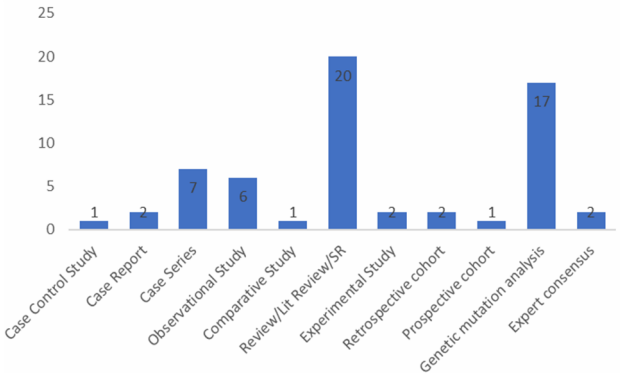


Figure 2: Distribution of study design in the field of short stature research

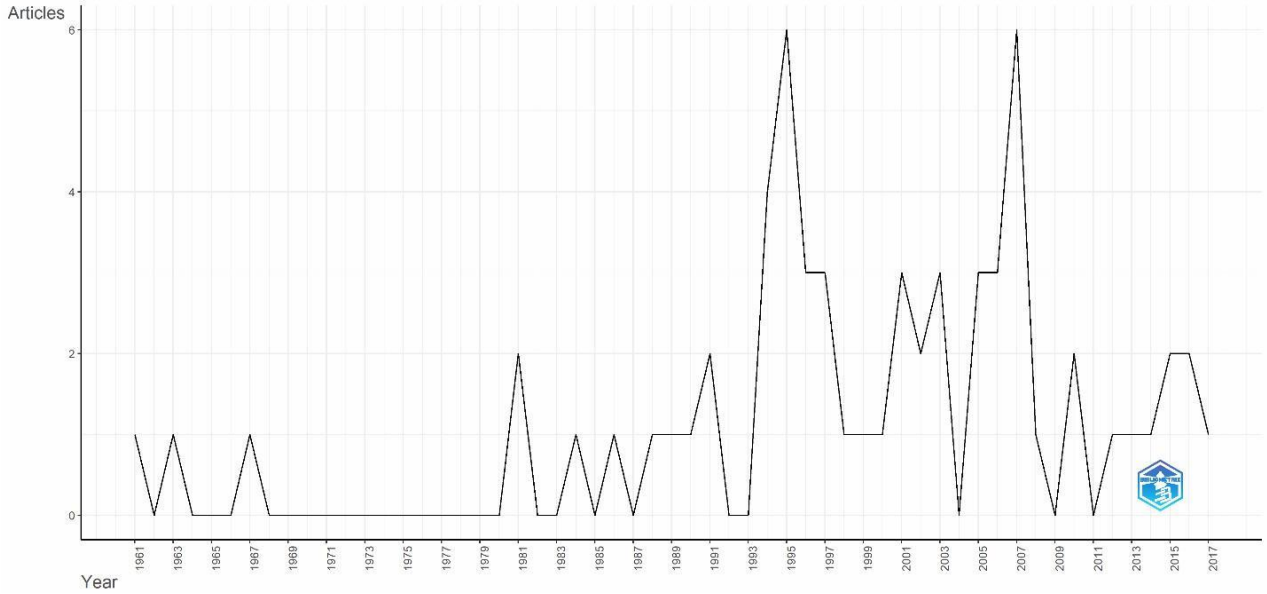


Figure 3: Time series plot of scientific production growth of research in the area of short stature

Table 2: Most Relevant and Productive Authors

Author	Total Citations	TC per Year	Normalized TC
Michael Holzer	1255	52.29	1.46
Niklas Nielsen	1231	43.96	1.62
Comilla Sasson	1150	46.00	1.00
Graham Nichol 1	1087	41.81	1.00
Christie Atwood	1052	28.43	1.00
J. Berdowski	1011	32.61	-
Karl-Heinz Kuck	980	51.58	1.84
Graham Nichol 2	901	69.31	1.00
Michel Haïssaguerre	892	33.04	1.00
David S. Siscovick	887	73.92	1.00
Jerry P. Nolan	880	38.26	1.32
Alfred Hallstrom	867	27.97	-
Terence D. Valenzuela	844	29.10	1.14
Benjamin S. Abella	815	40.75	1.88
Packer, Douglas L.	809	27.90	1.09
Saket Girotra	805	33.54	0.94
E.F.M. Wijdicks, M.D.	800	53.33	1.71
Jan-Thorsten Gräsnee	800	36.36	1.02
Syed S. Mahmood	795	36.14	1.02
Mary P. Larsen	777	21.58	1.00
Lan G. Stiell	753	34.23	0.96
Mads Wissenberg	675	24.11	0.89
Gavin D. Perkins	675	10.55	1.00
Vinay M. Nadkarni	675	15.34	1.38
Stuart J. Connolly	670	17.18	1.00
Gary B. Smith	655	36.39	1.41
Ingela Hasselqvist-Ax	594	66.00	1.09
Peter J. Schwartz	586	19.53	-
Philippe Ryvlin	577	19.90	0.78
Stephen Laver	549	28.89	1.03
Koenraad G. Monsieurs	517	28.72	1.11
Peter A. Meaney	517	21.54	0.60
Raymond H. Chan	515	17.17	-
Yih-Sharng Chen	500	62.50	1.00
Steven M. Frank, M.D.	500	55.56	0.91
Christine M. Albert	486	16.20	-
Barry J. Maron	479	15.97	-
Kjetil Sunde	473	26.28	1.01
Peter J. Kudenchuk	457	19.87	0.68
Ian Jacobs	454	26.71	1.00
Zhengming Chen	440	10.73	1.00
Jim Christenson 6	423	23.50	0.91
Thomas C. Mort	422	38.36	1.00
J. Hope Kilgannon	137	9.13	0.29
Jim Christenson 1	66	3.47	0.12
Tom P. Aufderheide	23	0.68	0.12

second position with 11% of publications contributing significantly to the research in short stature. Germany, Netherlands and Italy accounted for 5% of the publications each, meaning that, few researchers in the countries were interested in research in short stature, followed by Finland, Australia, England, France and Japan with 3% of Publications each.

Analyses of the Most Productive Journals in the Area of Short Stature

The graphical representation of the most productive journals where the Top 50 articles in the area of short stature were published is presented in Figure 4. The figure below depicts the first 10 most productive journals, such that, "The American Journal of Medical Genetics Part A" had published 17 articles in the field of short stature, followed by "NATURE GENETICS" with 8 articles and CELL, JOURNAL OF CLINICAL INVESTIGATION, SCIENCE, THE AMERICAN JOURNAL OF HUMAN GENETICS and THE AMERICAN JOURNAL ENDOCRINE REVIEWS had just published 2 articles each between 1963 to 2019 as shown in (Figure 5). We can say that the journal "THE AMERICAN JOURNAL OF MEDICAL GENETICS PART A" is the most productive and relevant journal in the field of research on short stature from the USA.

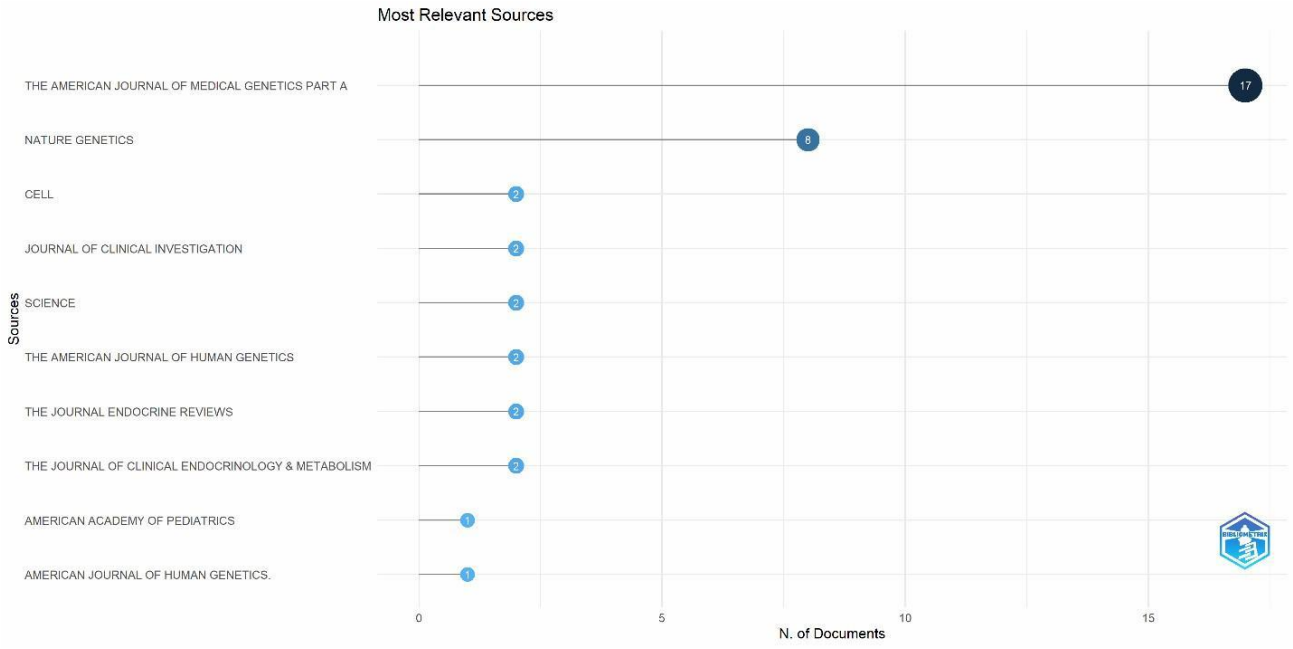


Figure 4: Trend line of the most production Journals in the field of short stature research

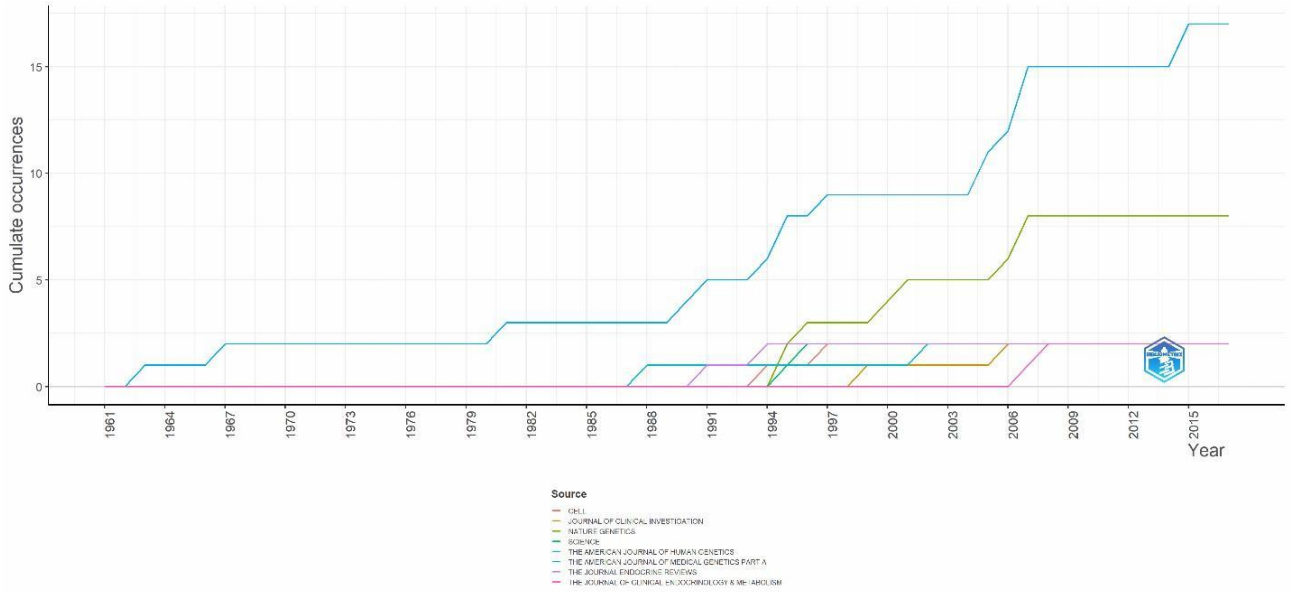


Figure 5: The trend line of journal production over time

DISCUSSION

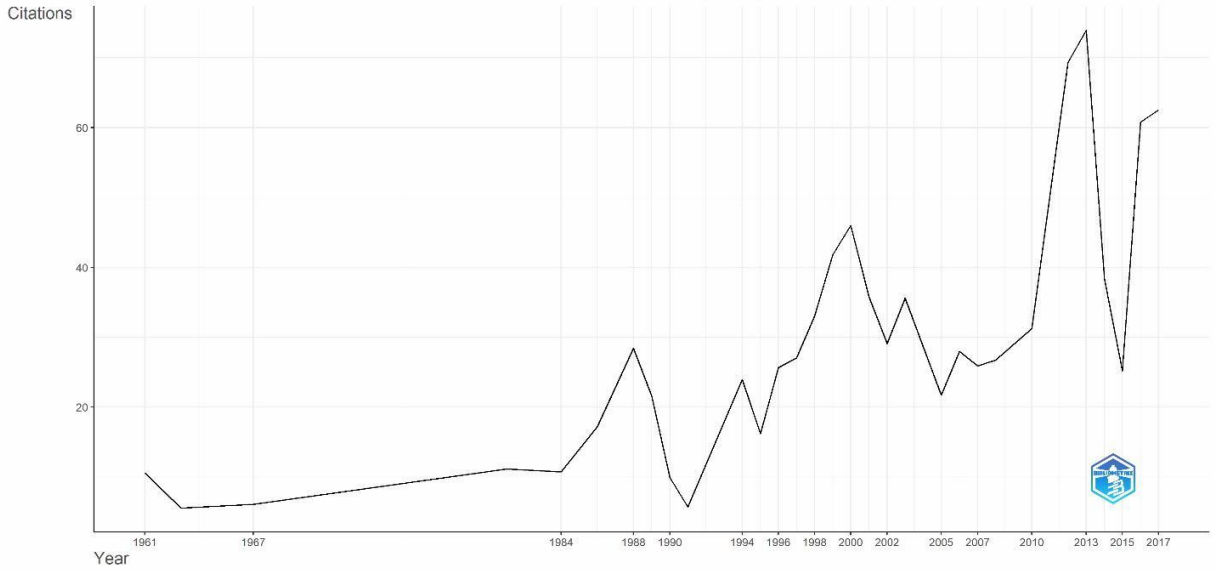
We found that research on short stature has steadily increased over the past two decades, particularly since the early 1990s, peaking in 1993 and 2004, reflecting growing academic interest and its significance as a public health issue. This rise in publications followed the World Health Organization's (WHO) 1993 review, which identified flaws in the existing growth references and recommended new growth curves. The WHO Multicenter Growth Reference Study conducted between 1997 and 2003, developed these updated growth standards to better assess global childhood growth and development [68]. In response to many cases remaining undiagnosed and labeled as Idiopathic Short Stature (ISS), the Growth Hormone Research Society, along with other pediatric endocrine organizations, organized an international workshop in 2007 to review evidence on the evaluation and management of ISS. Experts from around the world contributed to the development of a consensus document, addressing key issues through a structured model to guide clinical practice in managing children with ISS [69,70].

The study reveals that the research landscape on short stature is predominantly shaped by the Americas (AMRO) and Europe (EURO) regions of the WHO, collectively accounting for nearly two-thirds of all related publications. The United States, within AMRO, leads with a substantial 58.0% of the global research output, followed by the United Kingdom in EURO with 11.0%. The two most cited authors in this field are Michael Holzer, with a total of 1,255 citations and Niklas Nielsen, with 1,231 citations, both from the United States. Several factors contribute to the leading role of these regions in short stature related research. One key reason is the presence of influential institutions such as the US Centers for Disease Control and Prevention (CDC), the European CDC and various other public health organizations that drive research efforts. These institutions facilitate collaboration and funding, ensuring sustained research development [71]. The emphasis on reducing health disparities and ensuring inclusive healthcare aligns with the Sustainable Development Goals (SDGs), (3) particularly Goal 3: "Good Health and Well-being," which seeks to promote healthy lives and well-being for all individuals, regardless of age [72]. This goal addresses a wide range of health issues, including maternal and child health, infectious diseases, non-communicable diseases, mental health and access to essential healthcare [73]. Both AMRO and EURO regions prioritize health equity and focus on tackling noncommunicable diseases, positioning them at the forefront of global health research efforts [73,74].

Short stature, arising from causes such as chronic conditions, genetic disorders, familial short stature or consensus papers, dominate the research landscape by providing clinical insights and recommendations for management. Level 4 studies (17.0%) also play a pivotal role, with *The American Journal of Medical Genetics Part A* emerging as the most cited journal, publishing 17 articles highlighting the genetic basis of conditions like Noonan syndrome, Turner syndrome and Silver-Russell syndrome. These studies offer critical information on mutations in genes such as PTPN11, FGFR3 and FGF23, shedding light on how alterations in growth hormone receptors or regulatory proteins contribute to short stature.

However, significant research gaps remain, particularly in translating genetic discoveries into clinical applications. While genetic studies have provided insights into the mutations responsible for growth disorders, further research is needed to improve the understanding of the long-term effects of these conditions and the effectiveness of treatments like growth hormone therapy. Additionally, the psychosocial impact of short stature is underexplored, with limited research on the emotional toll, social stigmas and mental health challenges faced by affected individuals and their families.

Supplementary Figure 1: Most Frequent Words



Supplementary Figure 2: Trend line of average total citations over the year

CONCLUSIONS

This bibliometric analysis provides a comprehensive overview of the most influential research in the field of short stature, offering valuable insights into publication trends, research methodologies and global contributions. The main results reveal that the United States and the United Kingdom leads global efforts in short stature research, collectively accounting for nearly 70% of the analyzed studies. Genetic investigations dominate the research landscape, with mutations in key genes such as PTPN11, FGFR3 and FGF23 being pivotal in understanding the underlying mechanisms of conditions like Turner syndrome and Noonan syndrome. Moreover, the prominence of systematic reviews and consensus papers demonstrates the ongoing need for comprehensive syntheses of evidence to guide clinical decision-making. The dominance of genetic studies underscores the critical role of molecular insights in understanding the etiology and management of short stature-related conditions, such as Noonan syndrome and Turner syndrome.

Despite these advancements, significant gaps remain in translating these findings into improved clinical outcomes for patients. developing personalized treatment strategies, refining diagnostic tools and exploring the psychosocial impacts of short stature.

Limitations

The present study has several limitations that could affect its comprehensiveness. First, the exclusion of non-English articles may limit the review's global scope, potentially missing valuable research and introducing language bias. Second, by excluding grey literature and nonpeer-reviewed sources, we may overlook important findings that often present emerging trends or valuable insights. Additionally, focusing only on the most cited articles introduces citation bias, as highly cited studies may not always represent the most recent or innovative research, leaving newer studies with fewer citations underrepresented. Lastly, exclusion of psychosocial angles too.

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