



## Artificial Intelligence Driven Bibliometric Insights: Pioneering Down Syndrome Research

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**Abstract:** The present bibliometric analysis investigates the scholarly output from 2013 to 2022 to explore the use of artificial intelligence (AI) in Down syndrome research. The analysis demonstrates a significant and rapid growth in publications, starting from a minimal number of 17 articles in 2013 and reaching a peak of 2162 by 2022. This indicates a notable increase in interest and dedication to this topic. Upon analysing national contributions, the United States stands up as a frontrunner in terms of research output, citations, and collaborations. This underscores its crucial role in influencing the discussion and influence of AI-driven Down syndrome research. The dynamics of collaboration, especially between the United States and countries like the United Kingdom, China, and Germany, illustrate a vast worldwide network that facilitates the exchange of information. The congruence of these findings with past research highlights the regularity in exponential growth tendencies, which can be attributed to technical discoveries and interdisciplinary cooperation. Moreover, the prevalence of dominant nations, the significance of renowned publications, and the sway of prolific authors underscore the firmly established connections between research productivity and influence within specific fields of study. The study's findings suggest that future developments in AI-driven Down syndrome research will focus on integrating AI more deeply, fostering interdisciplinary collaborations, and prioritizing ethical considerations. These trends align with the anticipated paths and ethical obligations in this field.

### Introduction

Down Syndrome (DS), a condition marked by cognitive impairments and a range of medical complications, has generated much scientific attention with the aim of improving the well-being of individuals affected by it (Antonarakis et al., 2020). The emergence of artificial intelligence (AI) in DS research has the potential to revolutionize our understanding of the condition, enabling early detection and personalized treatment (Gupta et al., 2022; Rivera-Bonet, 2023; Subramanian et al., 2020). The rapid and extensive generation of data from diverse sources has become a prevalent phenomenon in the modern era of technology

(Lombardi and Vannuccini, 2022). Data is a vital resource that enables informed decision-making and operational optimization across several businesses. Handling this substantial influx of data has presented challenges, requiring the implementation of technologies such as big data analysis and text mining (Feldman et al., 2018; Kaur, 2023; Reddy and Khanaa, 2023). Text mining is a cognitively demanding task that facilitates the identification of patterns and the discovery of information inside collections of documents. Moreover, bibliometric analysis offers a quantitative approach to evaluate developing patterns and measure the research productivity in a certain subject (Ellegaard and Wallin,



2015; Haloi et al., 2023; Bulawit et al., 2023). Bibliometric analysis is a useful method for analysing scientific productivity and research trends. Bibliometric analysis can provide insights into the quantity and caliber of publications, as well as the geographic dispersion and collaborative tendencies of researchers in this field. Prior studies employing bibliometric analysis have identified significant patterns and advancements in the utilization of artificial intelligence (AI) in healthcare (Jana et al., 2009; Kammer et al., 2022). The researchers utilized databases such as SCI, SSCI, Scopus, and WOS, in addition to doing temporal and spatial studies, as well as co-occurrence, co-country, and co-authorship analyses. This study discovered that AI applications possess significant promise in enhancing clinical prognosis, reducing analytics expenses and developing clinical decision support systems (Jana et al., 2009; Kammer et al., 2022).

The present bibliometric analysis seeks to examine the emerging convergence of artificial intelligence (AI) applications in Down syndrome research. The effort is motivated by the increasing fascination and advancements in AI technology, which have sparked a more thorough examination of its potential implications and contributions to the comprehension, diagnosis, and treatment of conditions like Down syndrome (Koul et al., 2023). The rationale stems from a recognized deficiency in comprehending the rapid development patterns observed in AI-driven research on Down syndrome during the past decade (Windsperger and Hoehl, 2021). Comprehending and recording these patterns of growth may provide insight into emerging areas of focus and signal possible changes in research objectives within this interdisciplinary field (Windsperger and Hoehl, 2021). Moreover, the study aims to analyse worldwide research patterns by evaluating the contributions of different countries, collaborations, and influential entities such as leading journals and prolific authors. The objective is to gain insights into the importance of nations and entities in advancing this field. The study seeks to offer valuable guidance for future research attempts by identifying notable collaborators and potential opportunities for collaborative research projects through the characterization of these contributions. The study aims to contribute to the existing knowledge by extending and verifying the findings from previous studies, thus enhancing the collective comprehension of AI applications in Down syndrome research. Moreover, the study is anticipated to aid in policy-making and financial planning, as bibliometric analyses provide valuable guidance for allocating resources to future research and development endeavors.

## Materials and Methods

Consistent with previous studies, we employ bibliometric techniques, such as scientific mapping and performance analysis, to gain a deeper understanding of the development of artificial intelligence (AI) applications in the data science (DS) research field. We conducted a Systematic Literature Review (SLR) to initiate our analysis, enhancing the transparency, rigour, and comprehensiveness of our research. The search of Scopus and WoS databases ensured the inclusion of relevant material on the issue of 'AI applications in Down Syndrome research' due to its comprehensiveness, regular updates, and adaptability to facilitate bibliometric analysis. The literature searches and study selection processes were meticulously conducted to ensure the inclusion of pertinent publications while excluding those that did not contribute to our research objectives. This systematic approach served as the foundation for the subsequent bibliometric analysis of AI applications in Down Syndrome research.

### Selection of Database

We conducted a thorough search using two reputable databases, Scopus and Web of Science (WoS). The selection of these resources was based on their extensive inclusion of scholarly material in the fields of AI and Down Syndrome research, encompassing articles, conference proceedings, and journals. In order to enhance our bibliometric analysis, we have integrated advanced metrics like the h-index and i10-index. These metrics offer a deeper understanding of the impact and influence of research in the field of AI applications for Down syndrome. In addition, we have incorporated an analysis of alt metrics better to understand the changing landscape of research dissemination and impact. This approach provides a wider view of the impact and involvement of research beyond the usual academic references, showcasing the influence of the digital age on scholarly communication.

### Search Strategy

We implemented a predetermined search strategy that incorporated pertinent keywords and phrases. The search queries included terms such as "AI," "Artificial Intelligence," "Down Syndrome," and related subjects. Boolean operators were utilized to refine search queries, guaranteeing the retrieval of pertinent publications. The inclusion criteria of our search were limited to papers that were published in peer-reviewed journals and conference proceedings. We focused exclusively on papers written in English and included those published up until the most recent date at the time of the study (knowledge cutoff date: September 2021).

### Duration of Search and Preliminary Assessment

The search was restricted to papers that made a valuable contribution to the expanding field of AI applications in Down Syndrome research during a specific time frame that aligns with the objectives of our study. We obtained relevant bibliographic information, including titles, authors, publication dates, abstracts, and citation details, from the retrieved papers. This data provided the basis for subsequent bibliometric analysis. The selection of the study topic was based on a comprehensive evaluation of the existing literature to identify those that satisfied our criteria for inclusion. The sequence of actions undertaken was as: We commenced by scrutinizing all recognized publications to eliminate any duplicates and verify the uniqueness of each publication.

### Evaluation of the Title, Abstract, Textual analysis and Data Extraction

We conducted an analysis of the titles and abstracts of the remaining papers in order to ascertain their pertinence to the research subject. Publications that were plainly

irrelevant to AI applications in Down Syndrome research were excluded. We conducted a comprehensive analysis of the complete texts of publications that met the requirements of the title and abstract review in order to determine if they met the specified inclusion criteria. Excluded from consideration were studies that did not align with the research objectives. The pertinent data from the chosen papers, encompassing publication particulars and citation details, were extracted for subsequent bibliometric analysis.

### Dataset description

The bibliometric analysis utilizes an extensive collection of scientific articles sourced from 2078 different sources. The collection comprises 7896 articles, conference papers, and other scientific publications that were meticulously acquired from reputable journals and conference proceedings. A total of 7804 articles underwent thorough preprocessing, which involved meticulous quality checks and data cleansing. However, around 93 entries were excluded due to reasons such as

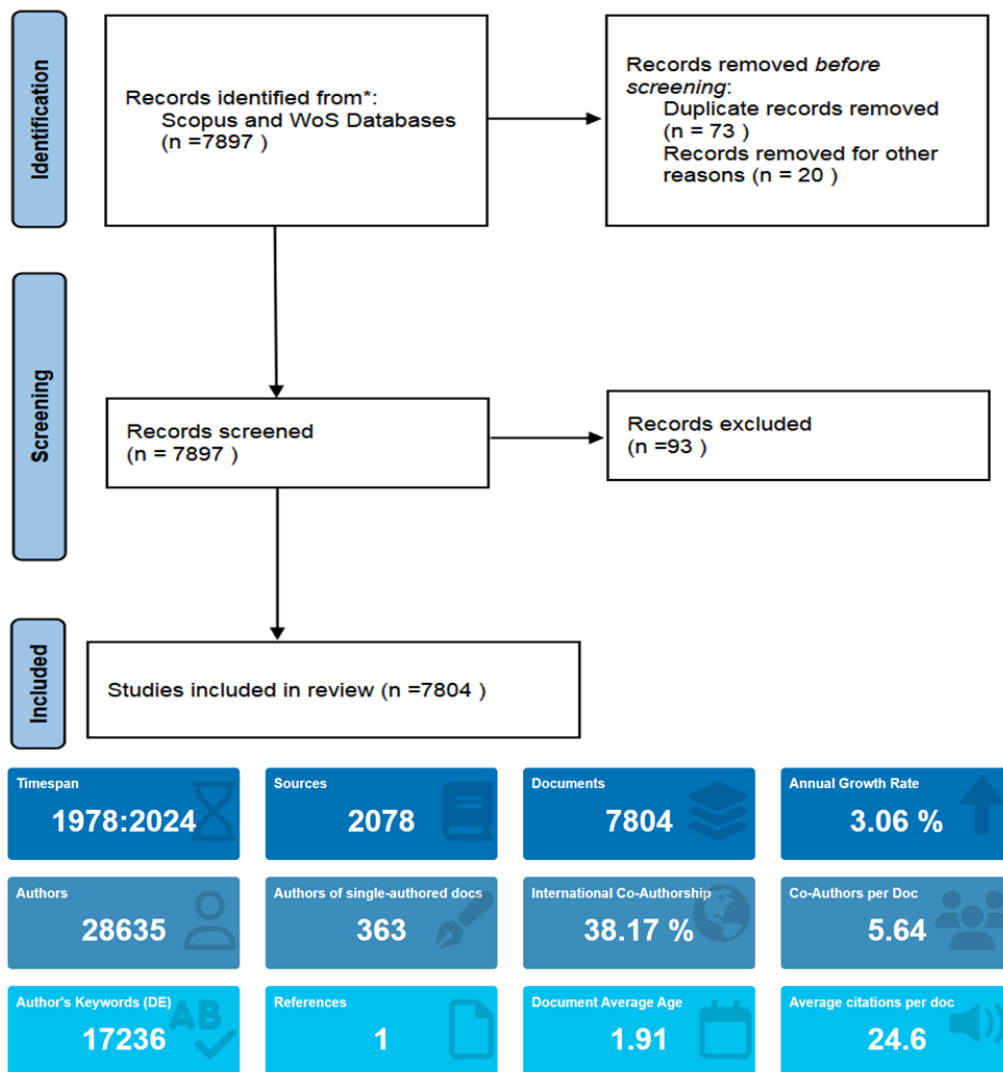


Figure 1. Flowchart of the PRISMA methodology.

duplication, missing information, or failure to meet the required criteria. The collection currently consists of 7804 pieces, offering a thorough overview of research trends and intellectual activity that have occurred throughout several decades. Figure 1 illustrates the PRISMA flow diagram of the investigation, while Table 1 offers further details about the dataset.

**Table 1. Detailed Dataset Description.**

<b>Main Information</b>		
<b>S. No</b>	<b>Description</b>	<b>Results</b>
1	Timespan	1978:2024
2	Sources (Journals, Books, etc)	2078
3	Documents	7804
4	Annual Growth rate %	3.06
5	Document Average age	1.91
6	Average citation per document	24.6
7	References	1
<b>Document Content</b>		
	Keywords Plus (ID)	20021
	Authors Keywords (DE)	17236
<b>Authors</b>		
1	Authors	28635
2	Single authored Docs	363
<b>Authors Collaboration</b>		
1	Single author	381
2	Co-authored per documents	5.64
3	International co-authorship %	38.17
<b>Document Types</b>		
1	Article	80
2	Conference papers	38
3	Review	7428
4	Review articles	1
5	Review, Book Chapter	16
6	Review, early access	241

## Results

The objective of the literature searches conducted for this study was to identify pertinent scholarly articles pertaining to the utilization of artificial intelligence in the field of Down Syndrome research. We adhered to a clearly outlined procedure to guarantee a thorough and systematic approach: The Systematic Literature Review (SLR) was executed with great attention to detail, adhering to a precise and rigorous methodology tailored to our specific research topic. Special attention must be given to the significance of search criteria and search refinement in bibliometric analysis. With this consideration in mind, we have partitioned the operation into three primary segments, commencing with the initial database search. In the second step, we employed search

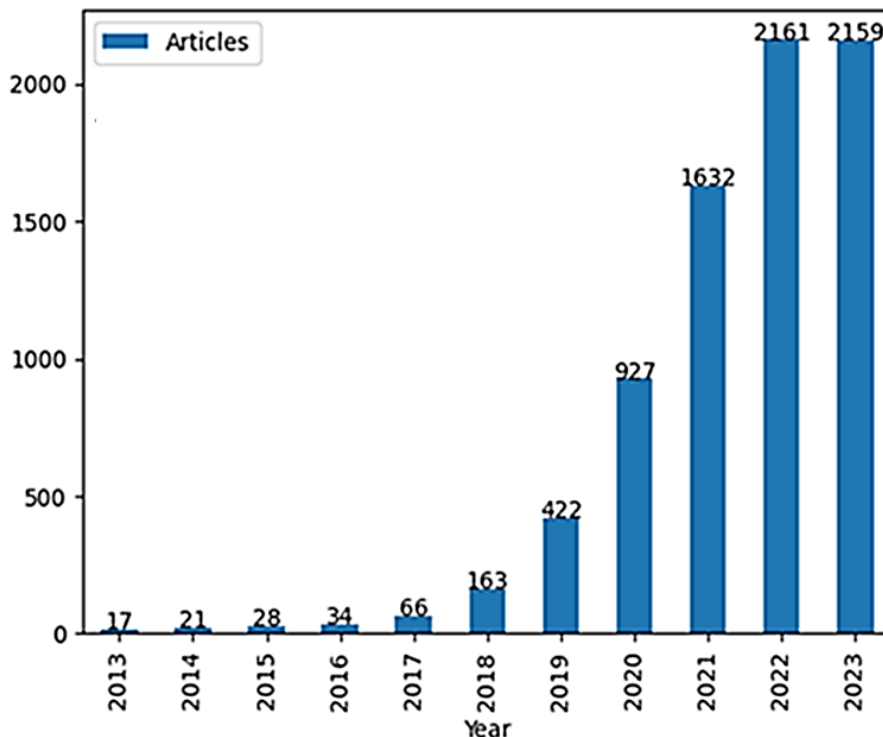
criteria to determine which studies should be selected for further study. Ultimately, we conducted a bibliometric analysis of scientific article production, presenting the findings for performance analysis and science mapping analysis. Figure 1 illustrates the visual depiction of the approach employed in this inquiry, as depicted in the PRISMA flow diagram.

### Annual scientific output

The landscape of AI and Down syndrome research has seen a significant transformation in the past decade, witnessing a substantial surge in scientific productivity. Annually, the quantity of publications experiences exponential growth during this timeframe. The bibliometric analysis provides a comprehensive overview of the yearly scientific output from 2013 to the present, spanning a period of ten years. In the initial year of 2013, a mere 17 articles were published. However, the quantity of publications on this subject has doubled each year since then. Consequently, the quantity of articles released in the research domain has surged to 2162 in 2022. Figure 2 illustrates the annual scientific output and the upward trend in scientific production for the selected time frame of the study.

### Country-wise Production

Different bibliometric indicators, such as the quantity of published papers, the number of citations received, the patterns of collaboration, and the impact factors, can be employed to evaluate the research article output of a country. The investigation has primarily focused on three pivotal indicators: the quantity of published articles, the overall count of citations, and the mean number of citations per country. Based on the statistics presented in Table 2, the United States is the frontrunner in terms of the quantity of papers published, with a total of 5646 articles. China follows closely with 4163 articles, and the United Kingdom comes in third with 2537 research articles produced in the research field. The leading ten countries based on their overall citations and average article citations (Table 2). Regarding this particular trend, the United States is at the forefront with 42533 citations and an average article citation score of 33.9, surpassing all other countries. China and the United Kingdom follow closely behind. Citation counts serve as a measure of a country's scientific impact and importance. Identifying the most cited countries involves considering both the quantity of publications and the frequency with which other experts reference these papers. High citation counts are indicative of a country's research being widely recognized and making a substantial impact on the progress of knowledge.



**Figure 2. Visual Representation of Scientific Output and Production Trends. Panel A illustrates the annual scientific output, while Panel B depicts the trend line indicating the increase in production over time.**

**Table 2. Ranking of Top 10 Countries in Scientific Publications. The table presents the top 10 countries ranked according to the quantity of their published papers, alongside metrics for their overall citations and average citations per article.**

Sl. No	Country	Number of Articles	Total Citations	Average Article Citations
1	USA	5646	42533	33.9
2	China	4163	26696	20.6
3	UK	2537	24973	42
4	Italy	2113	7376	15.4
5	India	1360	4560	14.8
6	Germany	1334	6813	22.9
7	Australia	1080	5982	20.4
8	France	1027	4992	22.6
9	Canada	919	5506	25.3
10	Spain	839	5061	23.6

**International collaborations**

International cooperation is crucial in bibliometric analysis to reveal the interconnected network of research activity among various countries. Table 3 presents a comprehensive overview of global research cooperation. The results were derived by considering cooperation between nations that occurred more than 50 times, as measured by the frequency count of such collaborations. The research findings indicate that the United States has the highest number of research collaborations with the United Kingdom (275), followed by relationships with

China and Germany. The collaborative ecosystem is visually depicted as a graph where nodes symbolize individual countries and directed edges signify the exchange of research collaborations. Figure 3 illustrates the network of collaborations at a national level within the selected research field. Every node in the diagram represents a country that engages in bibliometric collaboration. Nodes are connected by directed edges, represented by arrows, which indicate the direction of international bibliometric interactions. The graph illustrating the nation's cooperation showcases the

**Table 3. International Collaboration Among Countries. The table details the extent and nature of collaborative research efforts between different countries.**

Sl. No	From Country	To Country	Frequency
1	USA	UNITED KINGDOM	275
2	USA	CHINA	197
3	USA	GERMANY	177
4	USA	ITALY	175
5	USA	CANADA	149
6	CHINA	UNITED KINGDOM	148
7	UNITED KINGDOM	ITALY	136
8	UNITED KINGDOM	GERMANY	125
9	USA	INDIA	123
10	CHINA	AUSTRALIA	104
11	USA	AUSTRALIA	100
12	USA	FRANCE	98
13	USA	NETHERLANDS	98
14	ITALY	GERMANY	94
15	UNITED KINGDOM	INDIA	92
16	UNITED KINGDOM	NETHERLANDS	88
17	UNITED KINGDOM	AUSTRALIA	87
18	UNITED KINGDOM	SPAIN	84
19	USA	SPAIN	79
20	UNITED KINGDOM	FRANCE	75
21	USA	SWITZERLAND	75
22	UNITED KINGDOM	CANADA	74
23	ITALY	SPAIN	73
24	ITALY	FRANCE	66
25	GERMANY	FRANCE	65
26	GERMANY	SWITZERLAND	65
27	CHINA	SINGAPORE	63
28	GERMANY	NETHERLANDS	60
29	CHINA	INDIA	59
30	CHINA	CANADA	57
31	USA	BELGIUM	56
32	GERMANY	SPAIN	55
33	ITALY	NETHERLANDS	54
34	INDIA	AUSTRALIA	52
35	CHINA	GERMANY	52
36	GERMANY	CANADA	51
37	UNITED KINGDOM	SWITZERLAND	51

dynamics of research collaboration, where the sizes of the nodes correspond to the in-degree and out-degree. The size of the nodes corresponds to the level of engagement of countries in collaborations. For instance, the node representing the United States, followed by the United Kingdom and China, indicates their significant involvement as either initiators or recipients. The labels on the edges indicate the frequency of collaboration between the interconnected countries. The numerical values displayed on the edges represent the level of

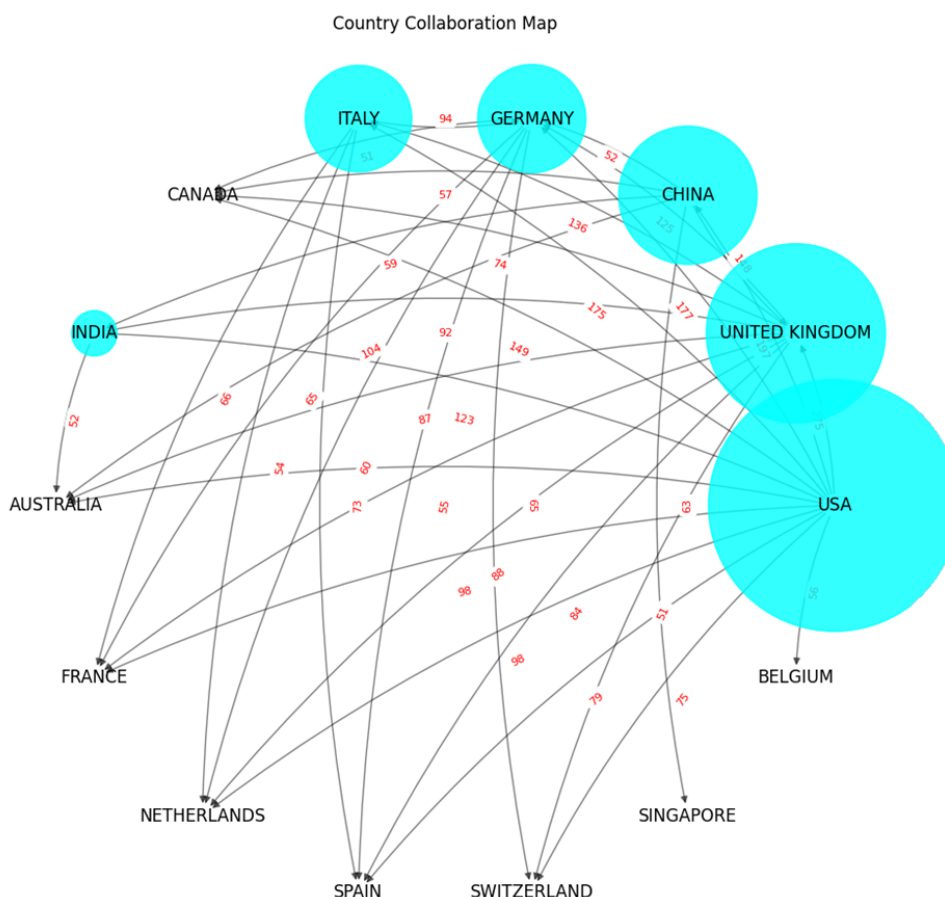
collaboration in terms of research articles. The combined dimensions of nodes and edge labels offer insights into the intensity and orientation of global partnerships.

Ensuring the strength and reliability of the study through a comprehensive bibliometric analysis is crucial to accurately identifying and utilizing the most pertinent sources. In this case, the phrase "most relevant sources" denotes a meticulous selection process where journals listed in esteemed databases like WoS and Scopus were chosen based on their direct pertinence to the research

objectives and the specific characteristics of the study. Figure 5 illustrates the top ten journals that are considered the most pertinent sources in the present research. According to the results of the analysis, the magazine Sensors holds the most significance in the research field, having published a total of 253 articles. Following Sensors, 244 research papers were published in the field of diagnostics and 202 research articles were published in Applied Sciences-BASEL, along with other prominent journals mentioned in the findings.

contributions to the area of interest. This information is crucial for allocating resources, enabling more efficient literature research, and ensuring that the selected sources are representative of the wider scholarly landscape. By adhering to Bradford's Law, bibliometric studies employ a systematic framework that enhances accuracy and significance in the identification and utilization of primary sources, hence enhancing the thoroughness and credibility of the study outcomes. Bradford's Law is commonly expressed in numerical form as (1).

In represents the quantity of journals in the nth zone,



**Figure 3. Collaborative Research Landscape Graph by Country.** The figure provides a graphical representation illustrating the interconnected research landscapes and collaborative networks among different countries.

### Bradford's Rule

The concept of Bradford's Law, a fundamental principle in bibliometric analysis, is crucial for comprehending the spread of knowledge across academic publications. As per this legislation, a limited number of journals or sources contribute significantly to the total number of publications on a specific topic, followed by a region with lesser production, and finally, a few irregularly referenced or peripheral journals. Bradford's Law aids in the strategic selection of sources for a thorough study in the field of bibliometric analysis, enabling researchers to concentrate their attention on the key journals that provide the most substantial

whereas  $C$  is a fixed value.  $r$  represents the ratio between the number of journals in one zone and the number of journals in the previous zone. The numerical value representing the zone is indicated by the variable " $n$ ". Figure 5 illustrates the findings of the analysis conducted on Bradford's law in the proposed study. The graph highlights the prominent position of top journals such as Sensors, Diagnostics, Applied Science, IEEE Access, and Sustainability, which are highly relevant sources of research.

### Authors with the Highest Relevance

When doing bibliometric analysis, identifying the most pertinent authors necessitates both a substantial

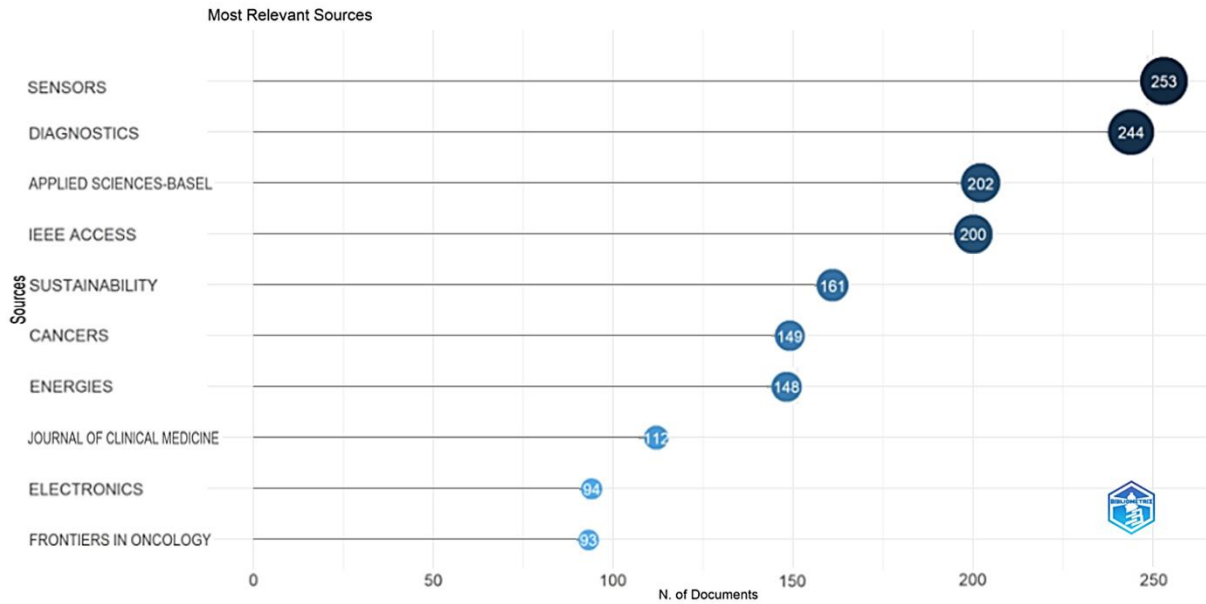


Figure 4. The figure represents the ten most influential journals, ranking them based on their significance and contribution to the ongoing research inquiry in the field.

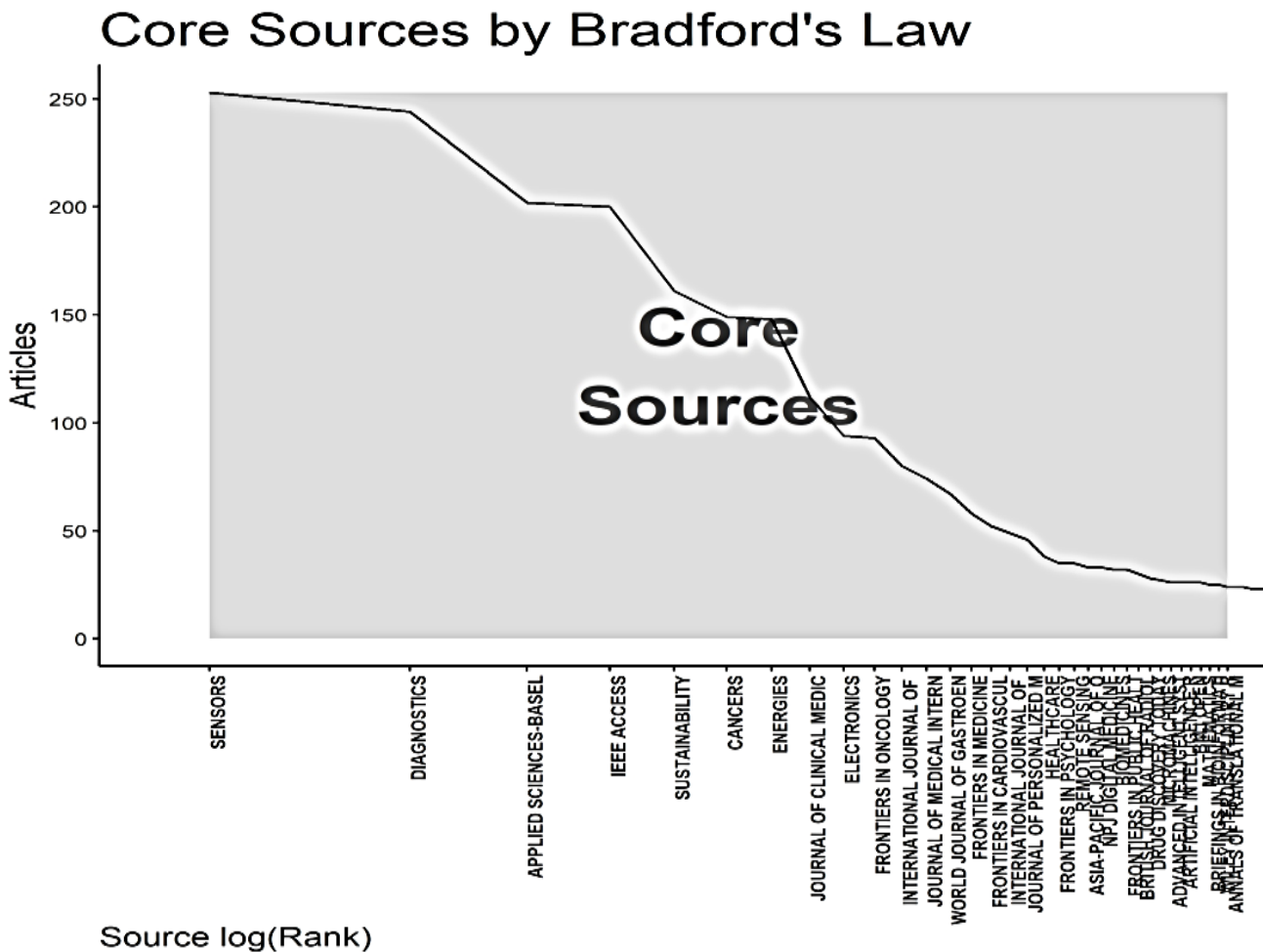


Figure 5. The figure illustrates the outcomes observed from the application of Bradford's Law in the analysis of scientific literature distribution.



quantity of articles and the use of fractional counting. Fractionalization refers to situations in which an author collaborates with others on a publication rather than just counting the number of articles. This approach acknowledges the cooperative aspect of authorship and provides a fairer representation of an individual's intellectual importance. Bibliometric analysis can effectively identify writers who have made substantial joint contributions to the field by including fractionalized publications. This advanced evaluation enables a comprehensive understanding of an author's impact, considering both their individual works and collaborative endeavors. Therefore, incorporating fragmented works into the analysis of relevant authors enhances the quantitative evaluation, offering a more comprehensive view of scholarly influence within a certain field. According to the analysis results provided in Table 4, Wang Y is at the top of the list with 82 articles and a fractionalized value of 15.71. Liu Y follows with 73 articles and a fractionalized value of 14.02, while Zhang Y also has 73 articles with a fractionalized value of 12.96.

words such as classification, artificial intelligence and prediction is 904,878, and 832, respectively. Since all these three words are very closely related to the artificial intelligence. The next word with maximum frequency is down-syndrome, with the frequency of 574.

### Tree Map Analysis

The results in Figure 7 depict a tree map analysis representing bibliometric data in a hierarchical format. This tree map employs nested rectangles to convey the results about categories or subsets of data, where the size of each rectangle corresponds to a quantitative measure of the frequency of specific terms within the collected literature. The varying sizes of rectangles within the map reflect the prevalence of terms, showing the importance of different words based on their frequency. The tree map provides hierarchical relationships among terms more efficiently, facilitating a deeper understanding of patterns and structures within the scholarly literature. As shown in the results, down-syndrome is contagious to artificial intelligence, prediction and classification, contributing 7% of the total word occurrences in the selected sphere. These patterns provide a clear proof of how the two areas

**Table 4. The data represents the ten most significant authors based on their relevance in the field, highlighting their contributions and impact.**

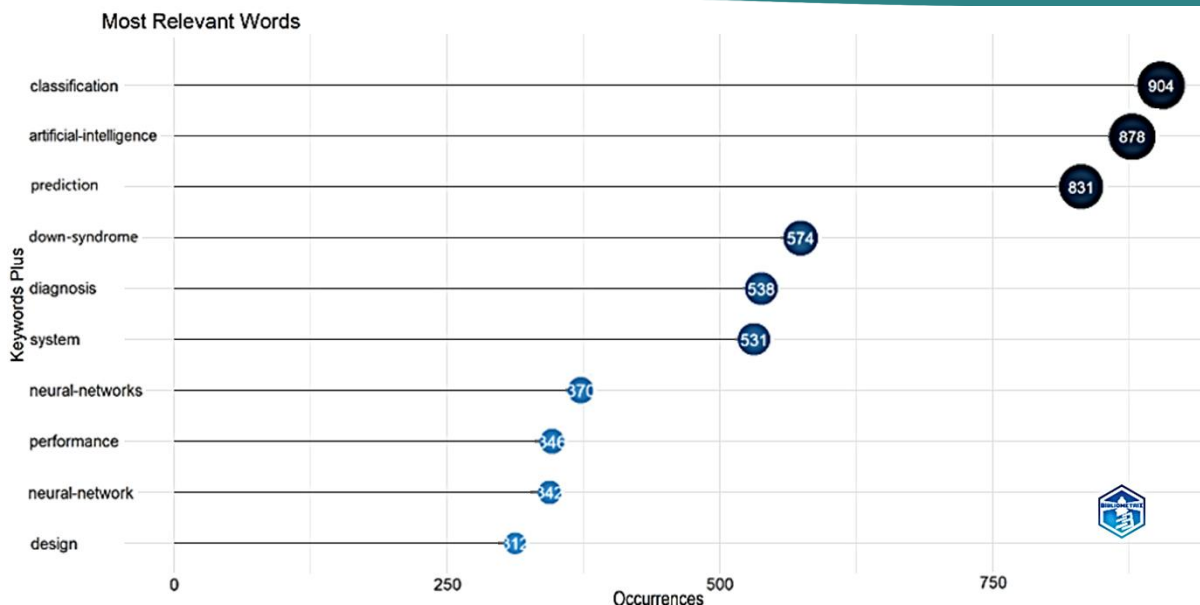
Sl. No	Authors	Articles	Articles Fractionalized
1	WANG Y	82	15.71
2	LIU Y	73	14.02
3	ZHANG Y	73	12.96
4	WANG J	80	12.18
5	WANG Z	63	12.04
6	LI J	74	11.83
7	LI Y	69	11.12
8	ZHANG J	55	11.05
9	WANG X	61	10.52
10	CHEN J	49	10.43

One of the important results of the bibliometric analysis reveals a comprehensive depiction of the most frequent words within the scholarly literature under consideration []. The graphical representation given in Figure 6 highlights key terms that dominate the discourse in the examined corpus, offering valuable insights into prevalent themes and areas of focus. Notably, words such as “classification”, “artificial intelligence”, “prediction” down syndrome,” emerge as leading words, reflecting the significance of the relationship in the fields under investigation. The visual graph provides a clear and intuitive illustration of the relative frequencies of these words, allowing researchers to discern patterns, trends, and the overall emphasis within the academic discourse. As can be seen in the graph, the frequency of related

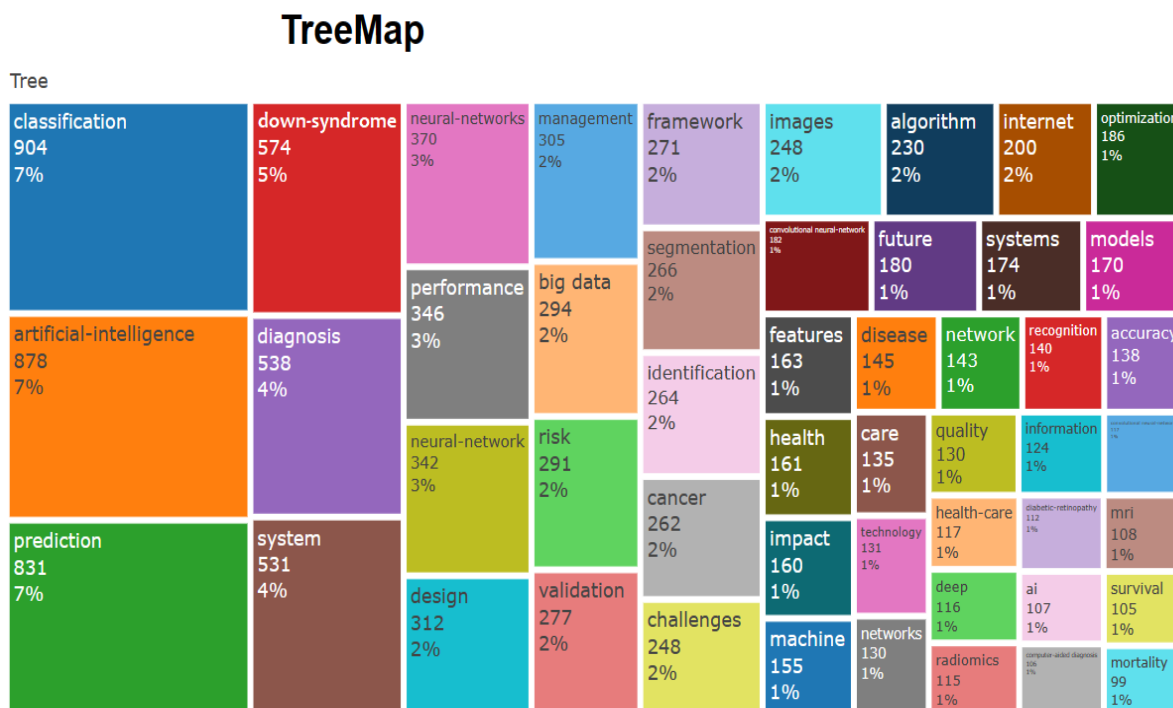
are significantly emerged in the past decade.

### Trend Topics

The purpose of incorporating "Trend Topics" analysis in Bibliometric Analysis is to provide researchers and scholars with dynamic and visually intuitive insights for exploring the evolving patterns and themes within a specific field of study. By leveraging advanced bibliometric techniques, the analysis identifies and highlights the most pertinent and trending topics within the scholarly literature, enabling users to stay abreast of the latest developments in their respective fields. This feature serves as a valuable aid for researchers seeking to understand the current intellectual landscape, identify emerging areas of interest, and pinpoint key themes driving research discussions. The trend topics in Figure 8 related to AI Applications in Down Syndrome Research



**Figure 4.** This figure graphically represents the most relevant words identified in the study, highlighting their frequency and significance in the context of the research.



**Figure 5.** The figure presents a tree map visualization, analyzing the distribution and relationships of keywords within the research context.

include Artificial Intelligence, classification and prediction diagnosis. Artificial Intelligence is the recent trend, along with topics like Down syndrome, trisomy 21, developmental disorder and parental diagnosis, which have appeared a significant number of times each, implying the prominence of using AI and related technologies in Down syndrome. The word “artificial

intelligence” appears 878 times, implying extensive relevance in using AI technologies in developmental disorders and Down syndrome. This emphasizes the need for more study and discussions in this field of study. In light of the fact that AI, down syndrome, classification and prediction are emerging topics of research, this suggests that using AI technologies in Down syndrome diagnosis, treatment and management is highly relevant.

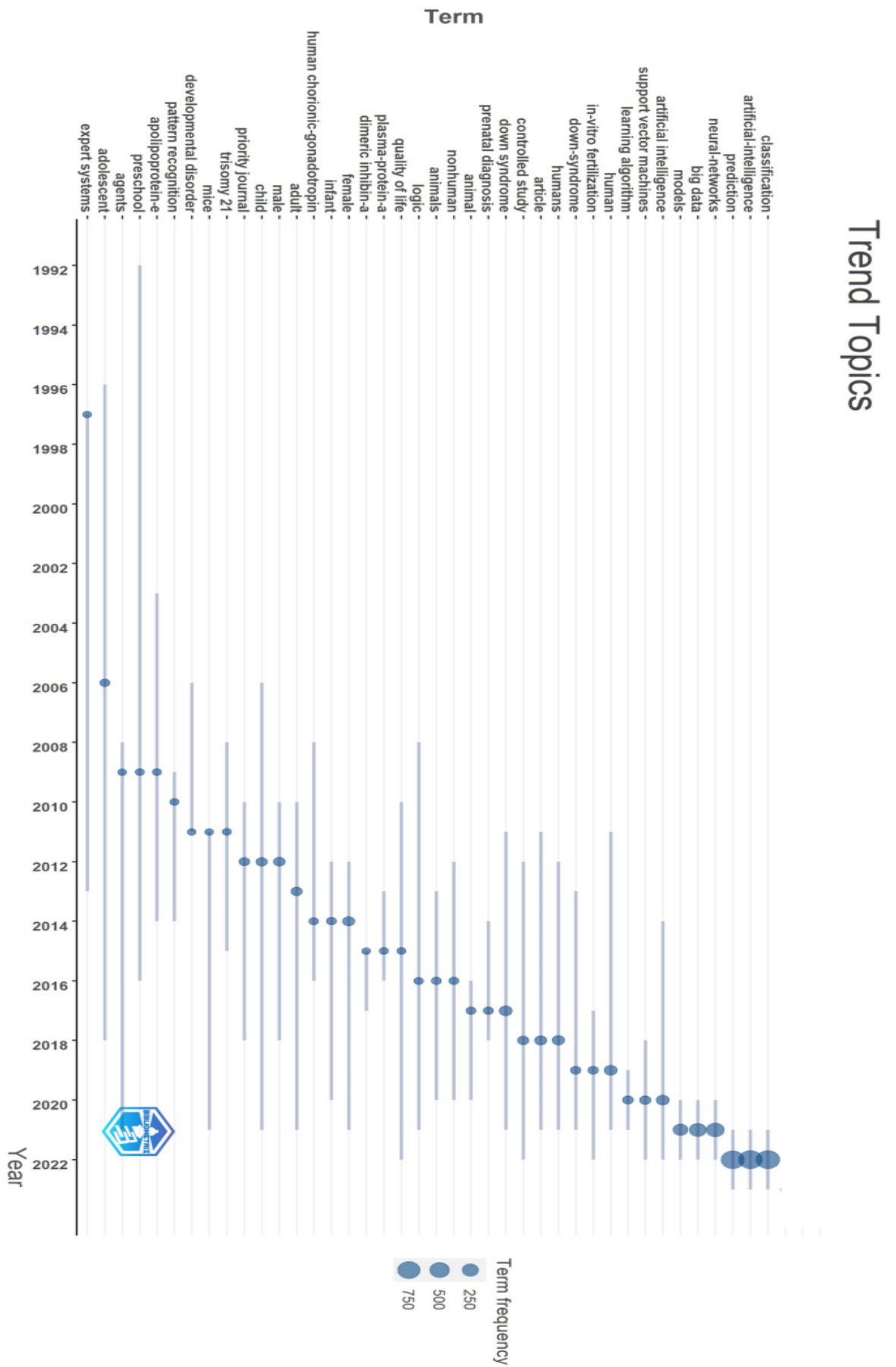


Figure 6. The figure provides a visual representation of the most prominent and trending topics in the field, highlighting their evolution and significance over time.

## Discussion

In the past ten years, there has been a significant surge in scientific publications focusing on the use of artificial intelligence (AI) in Down syndrome research (Paredes et al., 2022; Zhang et al., 2021). The exponential growth, starting from 17 articles in 2013 and reaching a significant 2162 in 2022, clearly illustrates the increasing interest and dedication to studying the intersection of artificial intelligence and Down syndrome. The significant increase in scientific output demonstrates the quick progress in technology and the increasing acknowledgement of AI's ability to tackle the various difficulties related to Down syndrome (Bohr & Memarzadeh, 2020; Fernandez-Batanero, Montenegro-Rueda, Fernandez-Cerero, & Garcia-Martinez, 2020). In various other domains, research has shown similar patterns of exponential growth, which can be attributed to advancements in technology and increased collaboration across multiple disciplines (Bohr and Memarzadeh, 2020; Reshi, Rustam et al., 2021). Upon examining the AI and Down syndrome research landscape, it becomes evident that several countries have played significant roles in advancing this field (Kalis et al., 2018; Zhang et al., 2021). The United States, China, and the United Kingdom are at the forefront in terms of research output, citation impact, and collaborative endeavors. Significantly, the United States not only has the highest number of research contributions in the field but also has a strong influence and recognition in terms of citation impact, demonstrating the wide acceptability and significance of its study. The dominance of this entity can be attributed to robust funding channels, a well-developed research infrastructure, and a conducive climate that encourages innovation and collaboration (Bohr and Memarzadeh, 2020). These findings corroborate previous bibliometric analyses that highlight the correlation between research productivity and impact, underscoring the importance of dominant countries in shaping discussions and advancing scientific fields (Okubo, 1997).

The collaboration patterns among nations further emphasize the interconnectedness of research activities within the AI and Down syndrome research landscape. The extensive cooperation, including those between the United States and the United Kingdom, China, and Germany, illustrates a worldwide network of information exchange. Collaborative techniques facilitate the consolidation of diverse knowledge, resources, and perspectives, leading to a comprehensive comprehension of Down syndrome and the use of AI to tackle its challenges (Alvino et al., 2020; Bohr and Memarzadeh,

2020). This aligns with previous studies that have shown the advantages and results of international cooperation in scientific research, highlighting the combined power and impact of collaborative endeavors (Wang et al., 2022). Specific publications are prominent sources that influence the discourse and dissemination of research within this academic field. The platforms of Sensors, Diagnostics, and Applied Sciences-BASEL have emerged as important venues for publishing research on the uses of AI in Down syndrome (Joshi et al., 2023; Murillo-Llorente et al., 2023; Torres-Nunes et al., 2023). Their extensive array of publications not only illustrates their significance but also their role in stimulating conversations and advancements in the field. Prior research has emphasized the significance of top-tier journals in determining research trajectories and fostering a cohesive scientific community, which in turn facilitates progress in certain areas of study (Merigo and Nunez, 2016).

The highly productive authors, whose contributions are crucial in shaping the research environment, play a vital role in the dissemination of information. Authors play a vital role in the AI and Down syndrome research sectors, not only by producing a large amount of work but also by making important contributions that have a measurable impact (Baldo et al., 2023; Chen et al., 2020; Paredes et al., 2022; Zhang et al., 2021). Their portfolio is expected to encompass a diverse array of AI applications in comprehending and managing Down syndrome, showcasing their significant contributions in shaping the discourse and advancements. Research has shown that prolific authors and their joint contributions have a significant impact on scientific achievements. This emphasizes the relevance of their work in advancing research agendas and promoting innovation (Chen et al., 2020). Based on current observations, the trends in Down syndrome research led by artificial intelligence suggest ongoing growth and potential avenues for exploration. In the future, more advanced AI technologies, such as machine learning and predictive analytics, could be employed to enhance diagnostic and therapeutic strategies. Interdisciplinary collaboration and ethical considerations will play a crucial role in the long-term progress of this industry (Kondo et al., 2023). Recent studies and foresight articles on the future implications and likely directions in AI and medical research shed light on the potentialities and challenges that lie ahead, offering valuable insights into the trajectory of this field (Reshi et al., 2021).

The recent discoveries reveal a significant and rapid growth in the utilization of artificial intelligence (AI) in Down syndrome research. We have included a variety of

case studies and practical examples to demonstrate the tangible effects of AI in Down syndrome research. These narratives highlight the practical advantages and progress that AI technologies have enabled in the fields of research and treatment, providing readers with a clear grasp of the capabilities and potential of this technology. The number of publications on this topic has surged from 17 in 2013 to a substantial 2162 in 2022, indicating an exponential increase. These findings align with the current body of literature on technological fields. This agreement aligns with previous research that highlights the exponential growth in scientific output due to technological advancements, underscoring the increasing interest and commitment to this field of study. Moreover, the prevalence of leading nations in terms of research output, citations, and collaborations demonstrates established connections between research productivity and impact as evidenced by bibliometric analysis (Kumari et al., 2023). These findings, in line with prior research, emphasize the pivotal roles of leading nations in shaping scientific discussions and enhancing specific subjects. Moreover, the recognition of influential journals like *Sensors*, *Diagnostics*, and *Applied Sciences-BASEL*, along with prolific authors, aligns with previous bibliometric studies that highlight the significant contributions of these journals and authors in influencing research directions and impacting scientific fields (Joshi et al., 2023; Murillo-Llorente et al., 2023; Torres-Nunes et al., 2023). The consistent identification of influential actors in the research ecosystem underscores the crucial role these entities play in guiding and shaping the trajectory of AI-driven Down syndrome research. Moreover, the study's suggestions for future endeavors, such as enhanced integration of AI and interdisciplinary collaborations with a focus on ethical considerations, align with the ideas expressed in prior foresight papers and reviews regarding the future implications and potential pathways in AI and medical research (Kondo et al., 2023; Lewis and Mercer, 2023). This alignment highlights the importance of maintaining consistent expected paths and ethical principles throughout the expanding range of AI applications in Down syndrome research.

As a result of our bibliometric analysis, we are thoroughly investigating the use of AI in Down Syndrome research. We are exploring the current trends and perspectives that shape this evolving field. The examination of numerical data, guided by rigorous bibliometric standards, has yielded a valuable understanding of the wide range of scholarly work, establishing the foundation for informed discussions and

future research endeavors. We have discovered several significant findings during our examination. There has been a noticeable evolution in research output throughout time, which demonstrates the academic community's adaptability to technological improvements and changing objectives. By analysing prolific periods and evaluating the H-index and total publications of sources, we successfully identified notable contributors and determined the concentration of scholarly impact inside specific journals and among particular writers. The analysis conducted on a decade-by-decade basis revealed temporal patterns that emphasized the emergence of certain themes and changes in attention. The correlation between Down Syndrome research and AI applications has experienced fluctuations in activity, aligning with advancements in technology, changes in legal frameworks, and variations in funding endeavors. Our bibliometric analysis's findings, in particular the frequency of phrases like "classification," "artificial intelligence," "prediction," and "down syndrome," highlight a noteworthy pattern in the use of AI in research on the condition. These terms are frequently used, which not only emphasizes AI's expanding significance in diagnostic and predictive techniques but also points to the appearance of hotspots in predictive analyses and diagnostic tools driven by AI. An interdisciplinary strategy combining artificial intelligence (AI) with conventional medical research is further revealed by the trend topic analyses and tree map. This points to a changing field in which artificial intelligence (AI) plays a key role in improving our knowledge of and ability to treat Down syndrome, rather than only serving as a helpful tool. Our bibliometric analysis's findings are consistent with the growing corpus of research highlighting the use of artificial intelligence (AI) in genetic and medical sciences. It has been demonstrated how AI might improve diagnosis procedures, especially for genetic diseases like Down syndrome (Alrefaei et al., 2022; Koul et al., 2023). This aligns with our findings about the frequency of phrases like "classification" and "prediction." Additionally, research on the trans-disciplinary applications of AI in medical research is consistent with our observations of a growing trend in which AI is becoming a more central, rather than merely supportive, component of Down syndrome research (Gupta et al., 2022). These parallels highlight the ways in which AI is becoming more and more important in both understanding and treating Down syndrome. They also point to a possible direction for future research that will concentrate on AI-driven innovations for better diagnostics and predictive analyses, which could

completely change the course of care and outcomes for those who have this condition. In order to improve the quality of life for people with Down syndrome, future research should concentrate on utilizing these insights to support creative, AI-enabled solutions that can increase diagnostic accuracy and predict developmental outcomes.

The present analysis examined the historical development of the area and provided valuable insights into the flexible nature of research at the intersection of AI and Down Syndrome. The findings and perspectives shown in this analysis have extensive implications for future research. Identifying influential sources, including their geographical influence and temporal contributions, serves as a guide for researchers navigating the ever-changing landscape of AI applications in Down Syndrome research. By identifying emerging trends, technologies, and multidisciplinary collaborations, scholars can get a forward-thinking perspective and focus their attention on areas that warrant further investigation. It is crucial to acknowledge the constraints of any bibliometric analysis. Although the provided data is comprehensive, it may not encompass the entirety of the research environment. Moreover, the dynamic and ever-evolving nature of AI technology and research on Down Syndrome necessitates a persistent reassessment of patterns and perspectives. There are abundant prospects for further academic pursuits. Potential research projects could explore the impact of certain AI methodologies, explore cooperative networks, and investigate the convergence of AI applications with other emerging technologies.

The integration of AI into Down syndrome research offers transformative potential, yet several challenges hinder its full realization. Key among these are the scarcity of high-quality datasets, privacy concerns related to medical data, and a skills gap in applying AI techniques among researchers and clinicians. To overcome these obstacles, a comprehensive strategy is essential. Enhancing data access while ensuring privacy can be achieved through secure sharing platforms and advocating for open science. Bridging the knowledge gap requires fostering collaborations between AI specialists, geneticists, and medical professionals, alongside investing in AI education for healthcare providers. Moreover, involving policymakers in creating supportive regulatory environments that prioritize ethical considerations is crucial. Embracing these approaches will unlock AI's promise in Down syndrome research, accelerating breakthroughs and leading to personalized care that improves life quality for affected individuals. The recent literature reflects the complexity and potential

of integrating AI into Down syndrome research. For instance, a study highlights the use of machine learning techniques to address cognitive challenges in Down syndrome individuals, suggesting the need for improvement in mental challenges and enhancing academic, social collaboration, and employment capabilities (Leghari and Ali, 2023). Another review focuses on the integration of AI and machine learning technologies for improved diagnosis and management of Down syndrome, emphasizing the role of these technologies in enhancing accuracy in diagnosis and understanding the complex interplay between genetics and environmental factors (Koul et al., 2023). These studies underscore the importance of a comprehensive approach to overcoming the challenges faced in integrating AI into Down syndrome research. By enhancing data access, fostering collaborations, investing in AI education, and involving policymakers, the full potential of AI in this field can be realized, leading to significant advancements in the care and treatment of individuals with Down syndrome.

### Conclusion and Future Prospects

Overall, this bibliometric analysis offered a thorough and detailed examination of the use of artificial intelligence (AI) in Down Syndrome research. It effectively combined quantitative metrics with nuanced observations to create a full understanding. This research study presents trends and opinions that provide a foundation for ongoing conversation, collaboration, and the stimulation of future revolutionary contributions to this important and dynamic issue. This study emphasizes the importance of utilizing advanced technology, such as AI, in Down syndrome research. It showcases the potential benefits and advancements that can be achieved, ultimately contributing to improved healthcare outcomes and aligning with the goals of Saudi Vision 2030. By incorporating AI into medical research, Saudi Arabia has the potential to establish itself as a center for cutting-edge healthcare solutions. This will not only support the Kingdom's goal of creating a dynamic society but also contribute to a flourishing economy. In addition, the valuable insights obtained from our bibliometric analysis can serve as a guide for Saudi Arabian researchers and policymakers to identify crucial areas for investment and growth. This holds significant importance for the strategic health initiatives outlined in Vision 2030, which place a strong emphasis on research, innovation, and the integration of cutting-edge technologies in the healthcare sector. The potential for integrating AI in medical research is limitless, offering exciting possibilities for the

future. Investing in AI in Saudi Arabia has the potential to make great advancements in personalized medicine, early diagnosis, and treatment plans for conditions such as Down syndrome. This is in line with the goals of Vision 2030, which aims to improve the overall quality of life and promote the well-being of its citizens. In addition, this study can help facilitate international partnerships in research and development. Collaborations with global research communities in AI and healthcare can greatly enhance Saudi Arabia's research capabilities, knowledge base, and innovation capacity. This, in turn, will contribute to the Kingdom's overall contribution to global medical advancements, aligning with Vision 2030. To sum it up, the incorporation of AI in Down syndrome research brings about immediate advantages for medical science and sets the stage for Saudi Arabia to fulfill its Vision 2030 goals in healthcare innovation and societal well-being. The potential for AI to revolutionize healthcare in Saudi Arabia is enormous, and our study lays the groundwork for future research further to advance the nation's progress toward a prosperous future.

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#### Declaration of competing interest

The authors declare no conflicts of interest.

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