

Genomic Disease Surveillance 2006 -2024: Current Trends as Revealed by a Bibliometric Analysis

Festus Mulakoli

School of Nursing and Midwifery, Aga Khan University, Nairobi, Kenya

Correspondence to Festus Mulakoli (mulakolifesto@gmail.com)

ABSTRACT

Background: Genomic disease surveillance has become an essential and transformative tool in public health. It enhances our ability to detect, monitor, and control infectious diseases by providing real-time genetic information about pathogens. This advanced approach allows for more accurate identification of disease outbreaks, tracking the spread of infections, and understanding the evolution of pathogens over time. Global genomic surveillance faces challenges with a lack of standardized data protocols and insufficient funding. Addressing privacy and ethical concerns about genetic data is crucial for building stakeholder trust and cooperation. Overcoming these obstacles is vital for maximizing the benefits of genomic surveillance in global public health protection.

Aim: This study aimed to identify key research trends and authors in genomic disease surveillance.

Methods: This study employed bibliometric analysis using data from the Scopus database, which was analyzed quantitatively with the biblioshiny software for bibliometric analysis. The R package offers functions for analyzing yearly outputs, country outputs, citations, collaboration networking, and publication trends.

Results: Two thousand and sixty-two (2062) articles were retrieved from the Scopus database. The United States leads with a total of four hundred and three (403) publications. Giovanetti M. was the lead author with twenty (20) publications. The field of Medicine had the highest number of articles (867 publications). Notably, there was a significant increase in research output between the years 2017 and 2023, with the highest number of publications (202) in the latter. The top-ranked article, written by Quick J, has garnered an impressive 998 citations.

Conclusion: This bibliometric analysis of scientific publications on genomic disease surveillance from 2006 to 2024 identified significant research trends, thematic hotspots, and patterns of international collaboration. The study observed a notable increase in research output from developed countries such as the United States, United Kingdom, China, Australia, and Brazil, with less output from developing countries. These findings highlight the critical role of genomic surveillance in the detection, monitoring, and response to infectious diseases, particularly in the context of future pandemics.

BACKGROUND

Genomic disease surveillance has revolutionized the detection, monitoring, and control of infectious diseases worldwide.¹ By integrating genomics into traditional surveillance methods, public health officials have gained deeper insights into pathogen evolution, transmission patterns, and antimicrobial resistance (AMR).² This approach has been important during the coronavirus disease (COVID-19) pandemic, when genomic epidemiology dashboards played a crucial role in presenting complex information to policymakers and the public.

Integrated genomic disease surveillance has been implemented to varying degrees across different countries, with some countries making considerable progress and others facing challenges. Several countries have attempted to integrate genomic surveillance into their public health systems.^{3,4} For instance, the number of countries with in-country SARS-CoV-2 genomic sequencing capabilities

increased by 40% from February 2021 to July 2022.^{5,6} Countries such as Canada, Sweden, and England have more advanced surveillance systems, although they are not fully integrated.⁷

Developed countries have more advanced systems, whereas resource-limited countries strengthen their capabilities. The World Health Organization (WHO) supports country-driven efforts to strengthen genomic surveillance in its 194 Member States, with activities reflecting regional specificities.⁸ However, challenges such as obtaining sequencing equipment, a shortage of skilled staff, and obstacles to maximising the utility of genomic data persist and must be addressed for effective global implementation.

Data collected by the WHO show that 54% of the countries had this capacity in March 2021. By January 2022, owing to the major investments made during the COVID-19 pandemic, this number increased to 68%. Even greater gains were made in the public sharing of sequence data; in January 2022, 43% more

countries published their sequence data than in the previous year.⁹

Recent advancements in high-throughput deoxyribonucleic acid (DNA) sequencing technologies have enabled the rapid and precise identification and characterization of emerging pathogens, revolutionizing public health responses to outbreaks.¹⁰ The integration of genomic surveillance with wastewater monitoring has gained prominence, particularly during the COVID-19 pandemic, as it provides a complementary passive monitoring system for clinical surveillance.¹¹

Adaptive sampling strategies have shown promise in uncovering new variants of infectious diseases up to five weeks earlier than constant sampling methods, particularly in countries with limited resources.¹² Trends in genomic disease surveillance highlight the need for a unified global pathogen surveillance system with standardized processes.²

The establishment of networks such as the Global Emerging Infections Surveillance program demonstrates the importance of committed laboratories and expert consortiums in building and maintaining genomic surveillance capabilities.¹³ As genomic technologies become more accessible, their integration with traditional surveillance methods and other innovative approaches, such as wastewater monitoring, will be crucial for enhancing our ability to detect, monitor, and respond to emerging infectious diseases and antimicrobial resistance.¹⁴

Whole-genome sequencing (WGS) has become a prevalent technique for identifying and tracking pathogens, establishing transmission routes, and managing outbreaks.⁶ The coronavirus disease pandemic has highlighted the importance of genomic surveillance with large-scale genome sequencing analysis of SARS-CoV-2 variants to guide monitoring and public health response.¹⁵

The application of genomic surveillance extends beyond traditional methods and has been integrated with other technologies to enhance its effectiveness. For instance, cloud-based platforms, such as Solu, have been developed to integrate WGS reads or assembled genomes into real-time, user-friendly surveillance systems, bridging the gap between cutting-edge research and practical applications in healthcare settings.^{16,17} Genomic epidemiology dashboards have emerged as powerful tools for presenting complex genomic data with easily interpretable visualizations, thereby increasing transparency among scientists, policymakers, and the public.

Most European countries, with prominent examples including France, Germany, Italy, Spain, the United Kingdom, the Netherlands, Sweden, Denmark, Norway, Finland, and Switzerland, actively participate in genomic disease surveillance. This is largely facilitated by the European Centre for Disease Prevention and Control (ECDC), which coordinates surveillance efforts across the European Union and European Economic Area (EEA) countries, enabling data sharing and analysis across the region.^{18,19}

Most EU/EEA countries utilize whole-genome sequencing (WGS) for pathogen surveillance and antibiotic resistance

tracking.²⁰ The ECDC is central to coordinating genomic surveillance activities by facilitating data sharing, harmonizing methodologies, and supporting laboratory networks across Europe. Countries are particularly focused on the genomic surveillance of pathogens such as SARS-CoV-2, West Nile virus, and antibiotic-resistant bacteria.²¹ European countries have contributed genomic data to international platforms such as the Global Initiative on Sharing All Influenza Data (GISAID) to enable global comparisons and outbreak investigations.²²

Several Asian countries, including India, Indonesia, Malaysia, and Thailand, have established systems for genomic disease surveillance. The Asia Pathogen Genomics Initiative (Asia PGI) is a collaborative network aimed at accelerating the genomic sequencing of pathogens. Asia PGI was established in late 2021 with support from the Bill and Melinda Gates Foundation.^{8,23}

However, a Duke-NUS study found that outbreak detection efforts in South and Southeast Asia are under-resourced.²⁴ This study found that only approximately half of the countries reviewed had integrated pathogen genomic surveillance initiatives into their national plans. Challenges to pathogen genomic surveillance in Asia include limited sequencing capacity, shortages of trained personnel and reagents, inadequate budgets, and a lack of national plans and guidelines for pathogen genomic surveillance.²⁵

In South America, several countries have established robust genomic surveillance systems to tackle infectious diseases, including COVID-19.²⁶ Argentina is at the forefront of genomic surveillance in the region. The country has established a national genomic surveillance network that collaborates with international organizations to monitor the spread of infectious diseases. This network has been instrumental in identifying and responding to disease outbreaks, such as COVID-19 and influenza.²⁷

Brazil has a well-developed genomic surveillance program that plays a crucial role in monitoring the spread of SARS-CoV-2. The country has contributed significantly to global databases, such as GISAID, which tracks the genetic sequences of pathogens. Brazil's efforts have helped understand the evolution of the virus and inform public health strategies.^{28,29}

Chile has implemented a comprehensive genomic surveillance system to monitor infectious diseases and antimicrobial resistance. The country has focused on building its capacity for genomic sequencing and analysis, which is essential for detecting and responding to infectious disease outbreaks. Chile's collaboration with international partners has strengthened its ability to manage public health threats.^{30,31}

Colombia has established a regional genomic surveillance network in collaboration with neighboring countries. This network is vital for tracking the spread of diseases across borders and implementing coordinated response strategies to address them. Colombia's efforts have contributed to a better understanding of the epidemiology of infectious diseases in this region.^{32,33}

Other countries, such as Uruguay, Venezuela, and Peru, have made significant strides in genomic surveillance, particularly in COVID-19 tracking. The country has

developed a national strategy for genomic surveillance, which includes the collection, sequencing, and analysis of pathogen genome data. Their efforts have been crucial for identifying and controlling infectious diseases.³⁴

Genomic disease surveillance is a vital tool for tracking and responding to infectious diseases in South America, particularly in Brazil. Countries such as Argentina, Brazil, Chile, Colombia, Peru, Uruguay, and Venezuela have contributed to a better understanding of the epidemiology of infectious diseases and informed public health strategies. Continued collaboration and investment in genomic surveillance are essential to ensure the health and safety of populations in the region.^{35,36}

In Africa, Mali has established an advanced molecular surveillance system aligned with the WHO's global strategy for genomic surveillance through collaboration with research institutions in Benin.³⁷ However, there are contradictions and challenges in implementing genomic surveillance in Africa. Despite these advances, none of the countries have fully integrated surveillance systems, and surveillance is often fragmented across regions.

Several African countries have established robust genomic surveillance systems to address infectious diseases. South Africa has been the leader in genomic surveillance. Several high-capacity sequencing centres have been established, such as the Centre for Epidemic Response and Innovation (CERI) at Stellenbosch University and the KwaZulu-Natal Research and Innovation Sequencing Platform (KRISP) at the University of KwaZulu-Natal.^{38,39} These centres have played crucial roles in tracking the spread of SARS-CoV-2 variants and informing public health responses. Gambia has been the leader in genomic surveillance in West Africa. The country shares sequences with approximately 9% of all confirmed COVID-19 cases, making it one of the top contributors to the region.⁴⁰ This has enhanced the country's and the region's pandemic response. This level of sequencing enabled the early detection and monitoring of SARS-CoV-2 variants, contributing valuable data to global platforms such as GISAID. These efforts supported the regional tracking of variant spread and informed public health strategies, such as targeted vaccination and containment measures. Moreover, Gambia's participation in genomic surveillance initiatives fostered capacity building, strengthened laboratory infrastructure, and demonstrated a commitment to collaborative global health efforts, positioning the country as an active player in regional disease monitoring and preparedness efforts. This has been instrumental in tracking the spread of variants and implementing effective public health measures.^{41,42}

Kenya, Nigeria, Ghana, and Ethiopia have developed national genomic surveillance strategies.⁴³⁻⁴⁵ Kenya, for instance, has made considerable progress in genomic surveillance during the COVID-19 pandemic, developing a strategy aligned with the WHO's global framework to enhance sequencing capacity and integrate genomic data into public health decisions. Kenya's efforts are part of a WHO-supported regional initiative for timely access to pathogen sequencing.⁴⁶ Nigeria has expanded its genomic surveillance beyond that of COVID-19. The Federal Ministry of Health, along with the WHO and its partners, is implementing a strategy to strengthen sequencing

capacity through enhanced laboratory infrastructure and personnel training. Ghana is developing its genomic surveillance capabilities by building on COVID-19 momentum. Ghana participates in WHO initiatives to strengthen surveillance infrastructure through capacity building and data integration.⁴⁷ Ethiopia has established sequencing capabilities and is developing a five-year strategy (2022–2026) for SARS-CoV-2 and other pathogens. This strategy addresses the challenges of laboratory infrastructure and supports public health responses through genomic data analysis. The country collaborates with international partners to enhance surveillance capabilities and track infectious diseases.^{48,49}

In Africa, surveillance of genomic diseases is vital for tracking and responding to infectious diseases. The efforts of countries such as South Africa, Nigeria, Gambia, Kenya, Ghana, and Ethiopia have contributed to a better understanding of the epidemiology of infectious diseases and informed public health strategies. Continued investment in genomic surveillance and collaboration with international partners will be essential to ensure the health and safety of African populations.^{50,51}

Another field that has seen an increased use of genomic surveillance is antimicrobial resistance (AMR). Several countries have made significant strides in the use of genomic surveillance to combat antimicrobial resistance, which is a growing global health threat. In Kenya, the Ministry of Health partnered with the World Health Organization (WHO) through the Global Antimicrobial Resistance and Use Surveillance System (GLASS) to monitor resistance patterns in key bacterial pathogens and integrate genomic data to inform treatment guidelines.⁵² In India, genomic surveillance has been scaled up to track resistance in hospital-acquired infections, with data informing national AMR action plans and influencing antibiotic stewardship programs. Brazil has incorporated genomic tools into its national surveillance network to detect emerging resistance genes in clinical and environmental samples, helping prevent outbreaks. Sweden and the Netherlands have long-standing genomic surveillance systems that monitor resistance and guide public health interventions and antibiotic prescription practices. These efforts, coordinated by GLASS, underscore the importance of standardized cross-border genomic data sharing in addressing AMR globally.^{53,54}

Countries with limited resources face significant challenges in establishing advanced molecular surveillance systems.³⁷ In countries that depend on external funding, the focus on a single disease has limited integration, and parallel systems have been created to address other diseases.⁴ African countries have weak clinical and microbiological laboratory services. This has compromised the effectiveness of the genomic epidemiology of the pathogen in this region. Other challenges, such as sampling bias, data quality, and the need for standardized computational pipelines, hinder their implementation.^{55,56}

These contradictions highlight the importance of investing in infrastructure, capacity building, and training to fully leverage the potential of genomic surveillance. Future genomic disease surveillance represents a significant

advancement in public health practices, offering real-time monitoring of pathogens and informing targeted interventions in the fight against communicable diseases.⁶ As we move forward, it is crucial to adopt a One Health approach that integrates surveillance across human, animal, and environmental health sectors.⁵⁷ By addressing the current challenges and fostering international collaboration, genomic surveillance has become an even more powerful tool for preventing and controlling infectious diseases.

Genomic disease surveillance has revolutionized public health by providing detailed insights into the evolution and transmission of pathogens. To fully leverage its potential, universal access to real-time WGS data, integration of various data sources, and cross-sectoral collaboration using a single health approach are required.⁵⁸ As genomic technologies become more accessible, their integration into routine clinical microbiology and public health surveillance is crucial for effective disease prevention and control.

Genomic disease surveillance has emerged as a critical tool for monitoring and managing infectious diseases, particularly coronavirus disease (COVID-19). However, several challenges have hindered the implementation of effective global genomic surveillance systems. One significant gap is the lack of standardization and coordination in wastewater surveillance. A unified catalogue of software tools and services is required to streamline the end-to-end genomic wastewater surveillance pipelines.⁵⁹

Global disparities in SARS-CoV-2 genomic surveillance have revealed serious geographical gaps, highlighting the need for a more equitable distribution of resources and expertise.⁶⁰ Another challenge is the integration of wildlife surveillance into existing systems. Many emerging zoonotic diseases have wildlife origins; however, only 54.4% of the surveyed countries have wildlife disease surveillance programs.⁶¹ The lack of dedicated budgets affects outbreak investigations, sample collection, and diagnostic testing in wildlife surveillance.⁶¹ Furthermore, the complex ecology of spillover and spillback between domestic and wild animals poses additional challenges for managing shared pathogens.⁶² Addressing these gaps requires a multifaceted approach. Overcoming these challenges is crucial for improving global preparedness and responses to future disease outbreaks.

Genomic disease surveillance has emerged as a pivotal tool in global health, enabling the detection, monitoring, and characterization of infectious pathogens in humans. This approach has gained traction in response to major health crises, including the Ebola, Zika, and COVID-19 pandemics. This field has undergone rapid growth, resulting in a diverse body of scientific research. Bibliometric analysis offers a rigorous methodology for systematically exploring this domain.⁶³ By analyzing publication trends, co-authorship networks, and citation patterns, bibliometrics provides a structured overview of the intellectual landscape and development of the research field. This approach facilitates the identification of influential contributors, thematic evolution, and collaboration patterns. This study aimed to conduct a bibliometric analysis of scientific publications on genomic

disease surveillance from 2006 to 2024. By mapping scholarly output over nearly two decades, this study sought to uncover the dynamics and emerging frontiers of genomic surveillance research. These findings will inform research priorities, guide policy development, and support the development of global disease-surveillance systems.

Aim

This study aimed to identify major research trends, thematic hotspots, and patterns of international collaboration in genomic disease surveillance.

METHODS

Study Design

The bibliometric analysis methodology was used in this study. Data from the Scopus database were quantitatively analyzed using Biblioshiny software for bibliometric analysis. The author intentionally selected this database because of the comprehensive institutional access provided by their affiliated organization. Furthermore, the database is recognized for its comprehensive coverage, rigorous indexing standards, and advanced citation-tracking capabilities, making it a reliable and robust resource for scholarly research. The R package provides various functions for bibliometric analysis, including citation, keyword, and publication trend analyses.

Data Collection

Inclusion and Exclusion Criteria

Articles from Scopus, the leading database for academic research, were examined along with comprehensive citation data. Only indexed published articles were included in bibliometric analyses. The Boolean operators “Genomics” AND “Disease surveillance” were used to search for terms in the titles, abstracts, or keywords. Articles that were unavailable online were excluded.

Database Selection

The Scopus database was selected because of its research focus and availability of relevant publications. The authors searched the Scopus database for studies on genomic surveillance of diseases. Careful attention was paid to selecting keywords (“Genomics” AND “Disease surveillance”) that encompassed the full range of pertinent research while minimizing irrelevant results.

Search Strategy

A comprehensive and systematic search was performed in the Scopus database, focusing on the keywords “Genomic” and “Disease surveillance.” This process involved carefully establishing the inclusion and exclusion criteria to ensure that the most relevant studies were selected. To further refine the search results, various Boolean operators, such as “AND,” “OR,” and the asterisk symbol for wildcard searches, were employed, thereby enhancing the thoroughness and accuracy of the search.

Data Extraction and Preprocessing

We systematically extracted key components from each article, including the title, author names, author affiliations, abstracts, publication dates, and relevant keywords. Two independent authors evaluated the completeness and consistency of the metric data derived

from the manuscript to ensure data quality and reliability. This thorough assessment helped us determine which articles were relevant to the analysis.

During the review process, a comprehensive search was conducted for duplicate entries that were subsequently identified and eliminated from the dataset to maintain integrity. Articles that lacked the essential metrics required for our analysis were excluded to ensure robust and accurate evaluation. Following these steps, we aimed to create a well-defined and reliable collection of articles for this study.

Statistical Analysis

Co-authorship, co-citation, and keyword co-occurrence networks were created and visualized using Biblioshiny in the Bibliometrix package to identify and display research themes and clusters. Biblioshiny is a web-based graphical interface for the Bibliometrix R package designed to make bibliometric analysis more accessible to users who may not be familiar with R programming. Bibliometric analysis involves a quantitative study of academic literature, such as publication trends, citation patterns, authorship networks, and research themes. Each cluster was labeled to represent the core research topics and subfields of the corpus. Data preprocessing, bibliometric indicator computation, and visualization were conducted using R Studio version 4.4.2 (R Core Team, Vienna, Austria), which calculates collaboration indices, h-indices, and citation counts to assess research productivity and impact.

Researchers used the Bibliometrix package in R Studio to generate a comprehensive tag cloud by extracting and cleaning titles, abstracts, and keywords. This was followed by a bibliometric analysis using the Biblioshiny function and term frequency visualization through the word cloud function. Bibliometrix also analyzed international collaboration patterns using Scopus datasets, producing a network visualization of collaborative relationships based on author affiliations. In this visualization, the line thickness indicates the strength of the co-authorship connections, whereas the node size represents the publication count for each country. The bibliometric clustering algorithm assigns colors to different collaboration groups, resulting in a network map that effectively illustrates global collaboration trends.

RESULTS

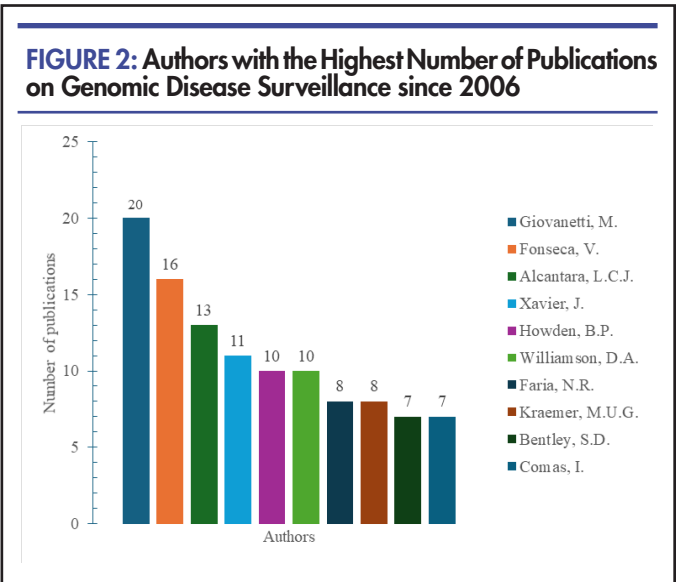
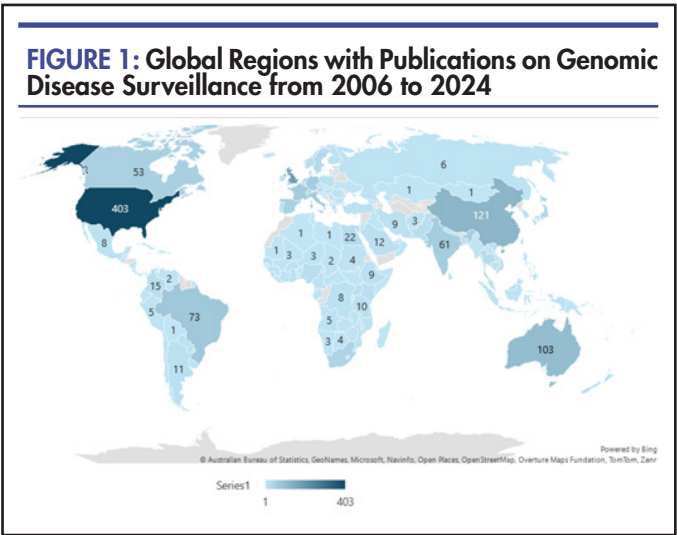
Research Outputs from Different Countries

Figure 1 illustrates the distribution of research publications across countries. The United States stands out as the leader in this domain, boasting an impressive total of four hundred and three (403) publications. The United Kingdom followed at a distance, contributing 181 publications to the global research community. China ranked third with 121 publications, while Australia had one hundred and three (103) publications. Finally, Brazil rounded out the list of the top five countries with seventy-three (73) publications.

Authors with the Highest Number of Publications

Figure 2 provides a detailed overview of the leading authors in this field. Marta Giovanetti holds the top position, with an impressive total of twenty (20) publications.

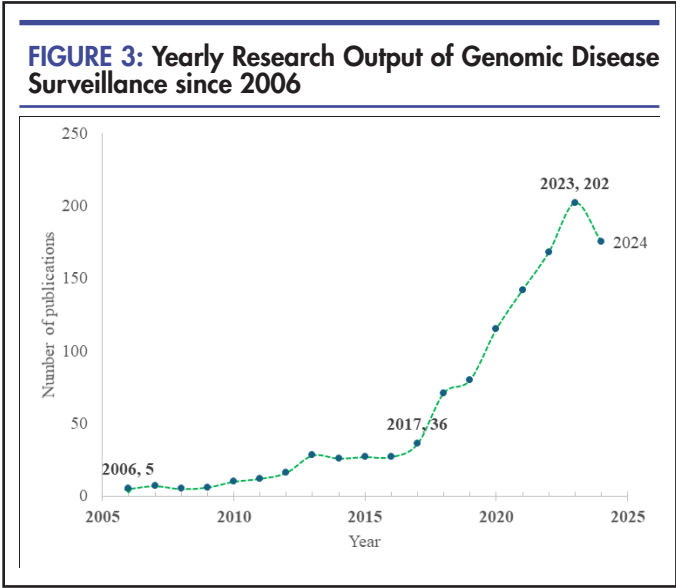
published works, reflecting a significant contribution to the field. Following Giovanetti, Fonseca authored sixteen (16) publications, indicating a strong and influential presence in the literature. Alcantara ranked third, with thirteen (13) publications, highlighting his commitment to advancing knowledge in this domain. Fourth, J. Xavier has eleven (11) publications highlighting impactful research efforts. Finally, Howden B.P. rounds out the list with ten publications, underscoring his valuable contribution.



Yearly Research Output

Figure 3 provides an overview of research publications from 2006 to 2024. Notably, 2023 saw a remarkable surge in research output, with two hundred and two (202) publications. The number of publications increased gradually from 2006, which had five (5) publications, to thirty-six (36) publications in 2017, before peaking in

2023, as previously mentioned. However, a decrease in the number of publications was noted in 2024.



Professional Fields and Scientific Journals with the Highest Number of Publications

The field of medicine led with eight hundred and sixty-seven (867) publications, followed by Immunology and Microbiology four hundred and eighty three (483) publications, Biochemistry, Genetics, and Molecular Biology two hundred and ninety four (294) publications, Agriculture and Biological Sciences eighty five (85) publications, and Veterinary seventy nine (79) publications as shown in Table 1.

Funding Agencies

Table 2 summarizes the leading funding organizations for research publications. The National Institutes of Health led the way with one hundred and twenty-seven (127) publications, making a significant contribution to medical research. The Department of Health and Human Services has one hundred and twenty (120) publications reflecting its vital role in public health and policies. The National Institute of Allergy and Infectious Diseases produced fifty-eight (58) publications, emphasizing its focus on allergies and infectious diseases. The European Commission contributed fifty-seven (57) publications, and the Wellcome Trust, known for its support of innovative research, published fifty-three (53) articles.

Most-Cited Articles

Figure 4 provides an overview of the most-cited articles in this research area. The article authored by Quick J. was ranked first with a remarkable total of 998 citations. Chan’s work has garnered 698 citations and displays its contributions. Additionally, Joensen KG made a notable impression with 632 citations, whereas Ioos S and Fitzgerald RC contributed significantly, receiving 475 and 474 citations, respectively.

TABLE 1: Leading Fields and Journals with Publications on Genomic Disease Surveillance between 2006-2024

Subject Area	Number of publications (n)
Medicine	867
Immunology and Microbiology	483
Biochemistry, Genetics, & Molecular Biology	294
Agricultural and Biological Sciences	85
Veterinary	79
Multidisciplinary	48
Pharmacology, Toxicology, & Pharmaceuticals	47
Environmental Science	45
Chemistry	20
Neuroscience	18
Journal	
Viruses	57
Emerging Infectious Diseases	51
Frontiers In Microbiology	48
Plos Neglected Tropical Diseases	41
Microbial Genomics	39
Plos One	34
Journal Of Medical Virology	22
Microbiology Spectrum	22
Eurosurveillance	21
Journal Of Clinical Microbiology	19

TABLE 2: Key Funding Bodies with the Highest Number of Publications on Genomic Disease Surveillance from 2006 to 2024

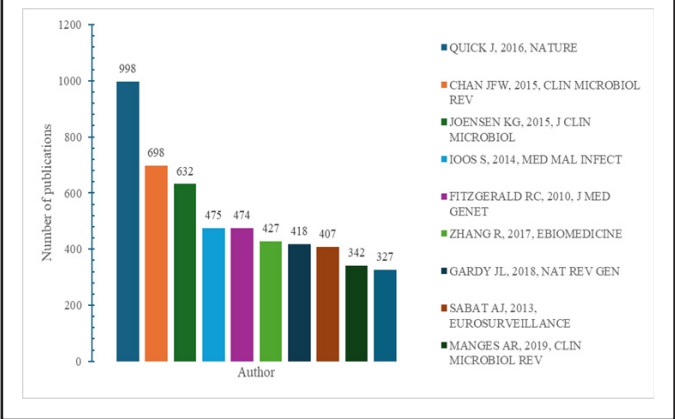
Organization	Number of publications (n)
National Institutes of Health	127
U.S. Department of Health & Human Services	120
National Institute of Allergy & Infectious Diseases	58
European Commission	57
Wellcome Trust	53
Centres for Disease Control and Prevention	49
National Natural Science Foundation of China	49
Horizon 2020 Framework Programme	41
Bill and Melinda Gates Foundation	40
Ministry of Science & Technology of the People's Republic of China	39
Medical Research Council	38
UK Research and Innovation	36
Conselho Nacional de Desenvolvimento Científico e Tecnológico	34
National Key Research and Development Program of China	28

Continue

TABLE 2: Continued

Organization	Number of publications (n)
Coordenação de Aperfeiçoamento de Pessoal de Nível Superior	26
National Health & Medical Research Council	24
Department of Health & Aged Care, Australian Government	21
European Research Council	19
National Institute for Health & Care Research	17
Biotechnology & Biological Sciences Research Council	16

FIGURE 4: Most Cited Research Documents on Genomic Disease Surveillance from 2006 to 2024



Co-citation Networking among Authors

Figure 5 presents a visual representation of five distinct co-citation clusters, each of which reveals the interconnectedness of scholarly articles. Within these clusters, the contributions of prominent authors such as Rambut A, Stanataks A, and Seemann T. stand out, emphasizing their influential roles in the field. Their work enriches academic discourse and highlights the relationships between various publications.

Collaboration and Networking among Authors

Figure 6 provides an overview of the collaborative efforts in genomic disease surveillance. This finding highlights the presence of three distinct collaborative clusters. The first cluster was spearheaded by Giovanetti, who displayed leadership in this area. Li Y and Howden BP guided the other two clusters, indicating a dynamic network of researchers working together to advance the understanding and management of genomic diseases. Each cluster represents a unique partnership that reflects diverse expertise and strategic surveillance approaches.

FIGURE 5: Co-Citation Network between Authors Interested in Genomic Disease Surveillance between 2006-2024

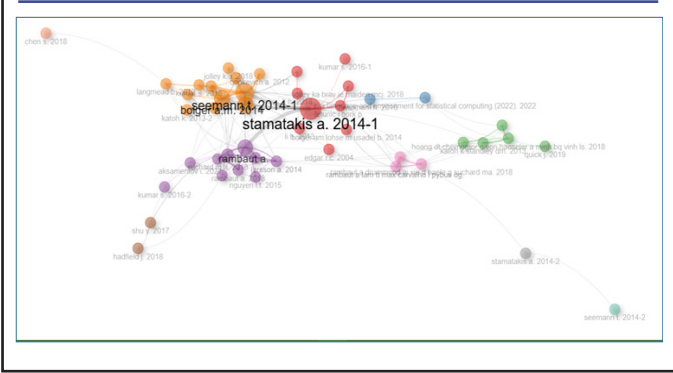
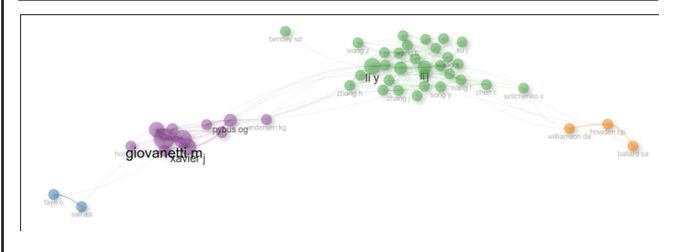


FIGURE 6: Collaboration Network between Authors and Publications in Genomic Disease Surveillance from 2006-2024



DISCUSSION

The results of this bibliometric analysis highlight the transformative impact of genomic technologies on the evolution of disease surveillance practices between 2006 and 2024. The substantial increase in scientific publications, particularly in the aftermath of the COVID-19 pandemic, indicates a global trend towards the integration of genomic data into public health decision-making processes.^{64,65} This trend underscores the enhanced accessibility of sequencing technologies and the growing acknowledgement of their utility in monitoring pathogen evolution, detecting outbreaks, and guiding targeted interventions. As genomic surveillance becomes increasingly integrated into health systems, it is crucial to critically assess its practical implications, including the opportunities it offers for innovation, the challenges it presents in terms of equity and ethics, and the strategies necessary to ensure its sustainability and effectiveness across diverse global contexts.⁶⁶⁻⁶⁹

Bibliometrix is a powerful tool for understanding the development, impact, and trends of research in a specific field.⁷⁰ In this study, a bibliometric analysis provided valuable insights into the growth of genomic surveillance of infectious diseases. This study emphasized the principal contributors who have influenced the field, elucidated seminal research that has molded our comprehension, and identified nascent areas of inquiry with potential for future investigations.

Since 2006, the global publication of genomic surveillance studies has increased significantly, enhancing the ability of researchers, public health officials, and policymakers to share vital data and findings.⁷¹ This growth has been supported by international frameworks such as the World Health Organization's Global Genomic Surveillance Strategy for Pathogens with Pandemic and Epidemic Potential (2022–2032), which aims to strengthen genomic surveillance capacities, promote data sharing, and integrate genomic insights into public health decision-making.⁷¹ Collaborative exchanges are crucial for tracking the spread of pathogens, identifying new variants, and developing targeted interventions to combat diseases. By fostering a global data-sharing network, genomic surveillance can inform public health strategies and improve responses to infectious disease outbreaks.⁷²

Since 2020, there has been a marked increase in publications related to genomic surveillance, a trend that closely aligns with the onset of the COVID-19 pandemic. This surge is well documented in bibliometric analyses and publication tracking systems, such as LitCovid, a curated literature hub developed by the National Centre for Biotechnology Information (NCBI). LitCovid has catalogued tens of thousands of articles related to SARS-CoV-2, many of which have focused on genomic sequencing and surveillance.⁷³ The urgency to monitor viral mutations in real time, especially with the emergence of variants of concern, has significantly accelerated research in this domain.

Globally, publishing studies encourage collaboration between countries and institutions. This collaboration has led to the development of standardized protocols, shared resources, and joint efforts to combat future infectious diseases.⁷⁴ Free access to genomic surveillance data promotes transparency in public health studies. It allows for independent verification of findings, fostering trust in the scientific community and public health authorities.⁷⁵

These findings offer critical insights for global policymakers to make informed decisions regarding public health initiatives. This is particularly important in developing countries, which have limited resources. Utilizing global genomic surveillance data allows governments to understand disease patterns and trends, which is crucial for developing precise, evidence-based public health policies. This approach enhances the efficacy of disease prevention and control, leading to better health outcomes and greater resilience against future public health threats.^{24,76}

Implications for Practice

Genomic surveillance has revolutionized the early detection and monitoring of infectious diseases. By sequencing the genomes of pathogens in real time, scientists have identified new and emerging threats before they become widespread. For instance, during the COVID-19 pandemic, genomic sequencing enabled the rapid identification of SARS-CoV-2 and its variants, such as Alpha, Delta, and Omicron. This early detection allowed governments and health organizations to implement targeted interventions, such as travel restrictions and localized lockdowns, to curb its spread. The ability to monitor changes in pathogen genomes also helps track transmission patterns and outbreak sources, which is

crucial for effective public health responses.^{77,78}

Genomic studies have provided deep insights into pathogen evolution. By comparing genetic sequences across different samples and time points, researchers have identified mutations that may affect the behaviour of a pathogen, such as increased transmissibility, virulence, and drug resistance. For example, genomic surveillance has shown how influenza viruses undergo antigenic drift and shift, necessitating annual updates of influenza vaccines. Similarly, the evolution of antibiotic resistance in bacteria like *Mycobacterium tuberculosis* and *Staphylococcus aureus* has been closely tracked through genomic data, informing treatment guidelines and containment strategies.^{79–82}

Genomic data have significantly accelerated the development of vaccines and therapeutics. By identifying specific genetic markers and protein structures of pathogens, researchers have designed targeted interventions to combat these diseases. mRNA vaccines for COVID-19, such as those developed by Pfizer-BioNTech and Moderna, have been rapidly created using the genetic sequence of the virus's spike protein. Genomic insights also help to monitor vaccine efficacy as new variants emerge, thereby guiding updates to vaccine formulations. Additionally, genomic data supports the development of antiviral drugs by revealing potential targets within the pathogen's genome.^{83–85}

The expansion of genomic surveillance has fostered unprecedented levels of global collaboration. Platforms such as GISAID and GenBank allow researchers worldwide to share genomic data in real-time, thereby promoting transparency and collective action. During the COVID-19 pandemic, millions of viral genomes have been uploaded and analyzed globally, enabling the rapid identification of variants and informing international travel and health policies. This collaborative spirit has also extended to capacity-building efforts, with high-income countries supporting genomic infrastructure in low- and middle-income regions to ensure more equitable surveillance coverage.^{86,87}

Genomic surveillance is increasingly being integrated into national and global health systems. The World Health Organization's Global Genomic Surveillance Strategy (2022–2032) emphasizes the importance of embedding genomic data into routine public health decision-making. These include the use of genomic insights to guide outbreak investigations, inform vaccination strategies, and monitor antimicrobial resistance. Countries such as the UK, through the COVID-19 Genomics UK Consortium (COG-UK), have demonstrated how genomic data have been operationalized within public health frameworks to enhance preparedness and response capabilities.^{76,88,89}

Challenges of Integrating Genomic Disease Surveillance

A major challenge in genomic surveillance is the lack of adequate infrastructure and technical capacity, particularly for LMICs. Many regions face shortages in sequencing equipment, reagents, and trained personnel, limiting their ability to conduct timely and large-scale genomic analyses. This creates significant gaps in global surveillance coverage, making it difficult to detect and respond to emerging threats in a coordinated manner.^{8,90}

Resource and infrastructure disparities among countries explain the lower scientific publication output of developing countries. These gaps prevent researchers in these regions from conducting comprehensive genomic surveillance studies and publishing them in international journals, thereby affecting research visibility and global public health. Targeted international support and capacity-building initiatives are essential to bridge this gap by focusing on enhancing research infrastructure, providing access to advanced technologies, and fostering collaboration between institutions in both developed and developing countries. Addressing these inequalities is crucial for empowering researchers in developing nations and recognizing their contribution to global health.⁹¹

Despite global efforts to promote open data sharing, many countries and institutions still struggle with interoperability issues. Differences in data formats, lack of standardized protocols, and concerns over data ownership and sovereignty hinder seamless genomic information exchange. These issues are compounded by limited digital infrastructure and inconsistent Internet access in some regions, which further restricts real-time data sharing.⁹²⁻⁹⁴ The lack of standardized methods for genomic sequencing and data analysis creates significant challenges in the comparability of studies across various research groups and institutions. Without a unified approach, differing protocols, techniques, and data interpretations have resulted in inconsistent outcomes, hindering reliable conclusions from collective research. The development and implementation of harmonized protocols are crucial for addressing this issue. Such initiatives would ensure consistency in genomic research, enhance collaboration, and facilitate more effective global sharing of findings. A common framework can improve research quality, promote reproducibility, and advance our understanding of genomics.^{56,95}

Genomic surveillance systems often rely on emergency funding during outbreaks, such as the COVID-19 pandemic. However, once an immediate crisis subsides, funding tends to diminish, threatening the sustainability of the systems. The WHO emphasizes the need for long-term investment and integration of genomic surveillance into routine health systems to ensure preparedness for future pandemics.^{91,96} Organizations such as the World Health Organization (WHO) have launched global initiatives, such as the Global Genomic Surveillance Strategy for Pathogens with Pandemic and Epidemic Potential 2022-2032.⁹⁷ Funding from the organizations identified in this study will enable researchers to develop initiatives that enhance genomic surveillance of pathogenic genetic variations, which are vital to public health. This funding also supports the creation of frameworks for effective data sharing and collaboration among academic institutions, healthcare providers, and government agencies, ensuring a coordinated response to health threats and better preparedness for outbreaks. Advances in sequencing technology and bioinformatics will further improve the accuracy and speed of genomic surveillance, allowing for comprehensive real-time pathogen monitoring.^{98,99}

Another challenge is the limited integration of genomic data into broader public health decision-making. In

many countries, genomic insights are not routinely used to inform outbreak responses, vaccination strategies, or antimicrobial resistance monitoring. This disconnection reduces the practical utility of genomic surveillance and highlights the need for stronger links between laboratories, epidemiologists, and policymakers.^{25,100}

Genomic surveillance raises important ethical and legal questions, particularly regarding data privacy, informed consent, and equitable access to technology. There is also a risk of stigmatization or discrimination if genomic data are misused or misinterpreted. The World Health Organization strategy calls for the development of ethical frameworks and governance structures to ensure responsible and inclusive genomic surveillance.¹⁰¹⁻¹⁰³

In summary, investing in capacity-building programs for low-resource countries is essential to enhance their participation in global genomic surveillance. This investment should be backed by major organizations, such as the National Institutes of Health, which are pivotal in funding these initiatives, as suggested by recent studies. Comprehensive training programs have equipped local scientists and healthcare professionals with the skills required for genomic research and disease surveillance. Additionally, developing robust infrastructure, such as laboratories and data management systems, will provide the facilities needed for significant genomic research. Financial support is crucial for sustaining these efforts and enabling local institutions to thrive and contribute to global health initiatives. These measures will bridge the resource and capability gap, promoting a more inclusive approach to genomic surveillance that benefits everyone.^{50,104}

Future Directions for Genomic Surveillance

The global publication of genomic disease surveillance data is crucial for public health and international collaborations. Despite these challenges, standardization methods, ensuring data privacy, and building capacity have improved the efficacy of genomic surveillance. Collaboration enables better detection, monitoring, and response to infectious diseases, thereby enhancing health outcomes. Genomic surveillance is rapidly evolving and driven by technological advances and the urgency of global health threats. The COVID-19 pandemic has highlighted the importance of tracking pathogens and informing public health responses. Future trends and developments will continue to shape genomic disease surveillance.¹⁰⁵

Technological innovations have significantly affected the future of genomic disease surveillance. Advances in next-generation sequencing (NGS) and real-time sequencing have enhanced the speed and precision of genome analysis. These technologies enable rapid identification of pathogens and monitoring of their genetic mutations, offering crucial insights into their evolution and dissemination.^{69,106} The COVID-19 pandemic has underscored the need for global collaboration in genomic surveillance. International bodies such as the World Health Organization (WHO) have devised strategies to enhance these efforts by utilizing existing capacities, overcoming obstacles, and encouraging data sharing and cooperation among nations. Collaborative efforts have enabled countries to better detect and respond to

emerging infectious diseases.^{20,107}

Integrating genomic surveillance into public health systems is essential for its success. This requires enhanced laboratory capacity, training of healthcare workers, and the creation of strong data-sharing networks. Embedding genomic surveillance into public health infrastructure enables a coordinated and efficient response to infectious disease outbreaks.¹⁰³ Genomic surveillance has transformed the battle against antimicrobial resistance (AMR) by monitoring genetic changes in pathogens to identify resistance mechanisms and guide the development of novel antibiotics to combat AMR. This method is crucial for addressing the increasing threat of AMR, which causes numerous deaths worldwide.^{108,109}

The future of genomic disease surveillance presents challenges, including sustained funding, workforce development, and the integration of genomic and health data. However, it also offers opportunities such as improved public health outcomes, better disease prevention strategies, and the potential for personalized medicine.¹¹⁰ The future of genomic disease surveillance is promising owing to technological advancements, global collaboration, and integration with public health systems. Genomic surveillance is crucial for global health, addressing ethical and legal considerations, as well as future challenges. Continued innovation and collaboration highlight the significant potential of genomic surveillance to revolutionize the field of public health.^{106,111}

Limitations

The limitations of this study arise from the exclusive use of data from the Scopus database, excluding other databases such as Web of Science and PubMed, which also contain substantial information on the subject. Variations in journal coverage across databases may lead to the underrepresentation of genomic surveillance research from low- and middle-income countries (LMICs). As highlighted in this study, citation counts and h-indices may have favored older articles with more time to accumulate citations, potentially underrepresenting the impact of recent studies, particularly those from 2023 to 2024. Furthermore, the analyzed metrics may not accurately reflect the quality or practical significance of this study. Although bibliometric tools have revealed publication trends and networks, they cannot capture the full scientific context of studies.

CONCLUSION

This bibliometric analysis of scientific publications on genomic disease surveillance from 2006 to 2024 identified significant research trends, thematic hotspots, and patterns of international collaboration. The study observed a notable increase in research output, with the United States leading in the number of publications, followed by the United Kingdom, China, Australia, and Brazil. These findings highlight the critical role of genomic surveillance in the detection, monitoring, and response to infectious diseases, particularly in the context of future pandemics.

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