Human genomics technologies in clinical studies – the research landscape

Report on the 1990-2024 period

Genomic technologies are becoming increasingly central to clinical practice, with the potential to transform the way diseases are diagnosed, treated and monitored. This snapshot provides a global overview of clinical studies involving human genomic technologies that were registered between 1990 and 2024. It examines patterns in research growth, disease focus, geographical distribution, population inclusion and equity of participation.

KEY MESSAGES

Human genomic data use in clinical studies is accelerating.
Since 1990, over 6500 studies involving human genome analysis have been registered globally, with rapid growth after 2010 owing to improved sequencing technologies, lower costs and expanded clinical applications.

Genomic research is geographically concentrated.
Ten countries account for over 70% of genomic studies, with 68% in high-income settings and less than 0.5% in low-income ones. This concentration reflects global disparities in research capacity and access, even within wealthier nations where access to genomic research and medicine may vary by ethnicity, socioeconomic status or geographical location.

Genomic research is centered on noncommunicable diseases, although infectious diseases represent an untapped potential.
Over 75% of genomic studies concern noncommunicable conditions such as cancer, rare diseases and metabolic disorders, where clinical applications are well established. Given the report's focus on human (rather than pathogen) genomics this is not surprising, but it reveals a missed opportunity to apply human genomic insights to infectious diseases. Genomics could inform targeted treatment, predict susceptibility or guide prevention strategies for high-burden conditions such as tuberculosis, HIV and malaria, especially in low-resource settings where precision approaches may improve impact.

Participation in genomic studies is uneven across the life course.
Most studies to date focus on an adult cohort. Only 4.6% of studies focused on children and 3.3% on older adults. Few studies explicitly included both paediatric and geriatric populations. These gaps may reduce the relevance of genomic findings for populations at different stages of life and limit the development of age-specific interventions.

Efforts to improve equity and impact will require more inclusive, globally aligned research strategies.
Addressing the current imbalances in study geography, disease focus and participant demographics will require stronger global coordination, investment in genomic infrastructure and broader engagement across underrepresented settings. Ensuring that genomic technologies are evaluated in diverse contexts is critical to advance health equity and achieve the full potential of genomics in public health.

Introduction

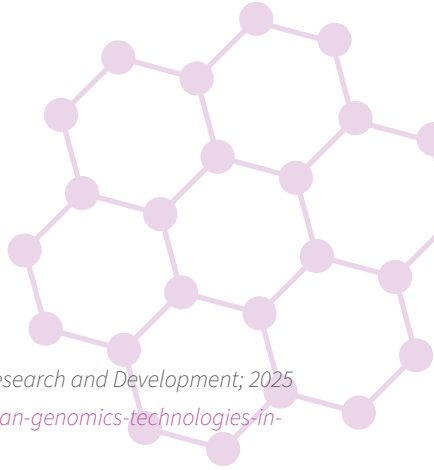
Genomic technologies are increasingly integrated into biomedical research and health care, enabling the identification of disease-associated variants, improving diagnostic precision and supporting the development of personalized medicine[1,2,3]. Advances in sequencing, gene editing and computational analysis have accelerated discoveries across a range of conditions including cancer, rare diseases, infectious diseases and complex noncommunicable disorders[4,5,6].

Over the past three decades, genomic research has shifted from fundamental discovery to widespread clinical applications. The completion of the Human Genome Project in 2003 laid the foundation for modern genomics[7], while subsequent innovations such as next-generation sequencing[8], genome-wide association studies[9,10] and CRISPR-based gene editing[11] have expanded the role of genomics in clinical research and translational science[12]. Falling sequencing costs and improved computational infrastructure have further accelerated this transition[13].

In this report, genomic clinical studies are defined as studies that incorporate human genomic data for the purposes of guiding disease detection, classification, treatment selection or therapeutic development. They include interventional and observational studies to evaluate genetically targeted therapies, pharmacogenomic responses, diagnostic tools and gene therapies, as well as other precision medicine approaches. Studies based solely on pathogen genomics are excluded in order to focus on the human genome*.

Although sequencing technologies have become more accessible, their uptake remains uneven across the globe[14]. Clinical research remains concentrated in high-income settings, with low- and middle-income countries underrepresented in terms of participation and infrastructure. Gaining a better understanding of where studies are taking place, which populations are represented and how research priorities align with global health needs is essential to foster inclusive and equitable genomic science[3,15,16].

This global snapshot analyses over 6500 clinical studies involving human genomic technologies registered between 1990 and 2024 via WHO's International Clinical Trials Registry Platform (ICTRP). It provides insights into trends by disease, geography, inclusion and equity, and assists policymakers, funders and research leaders to set priorities, expand access and ensure that genomic innovation supports diverse populations and settings.

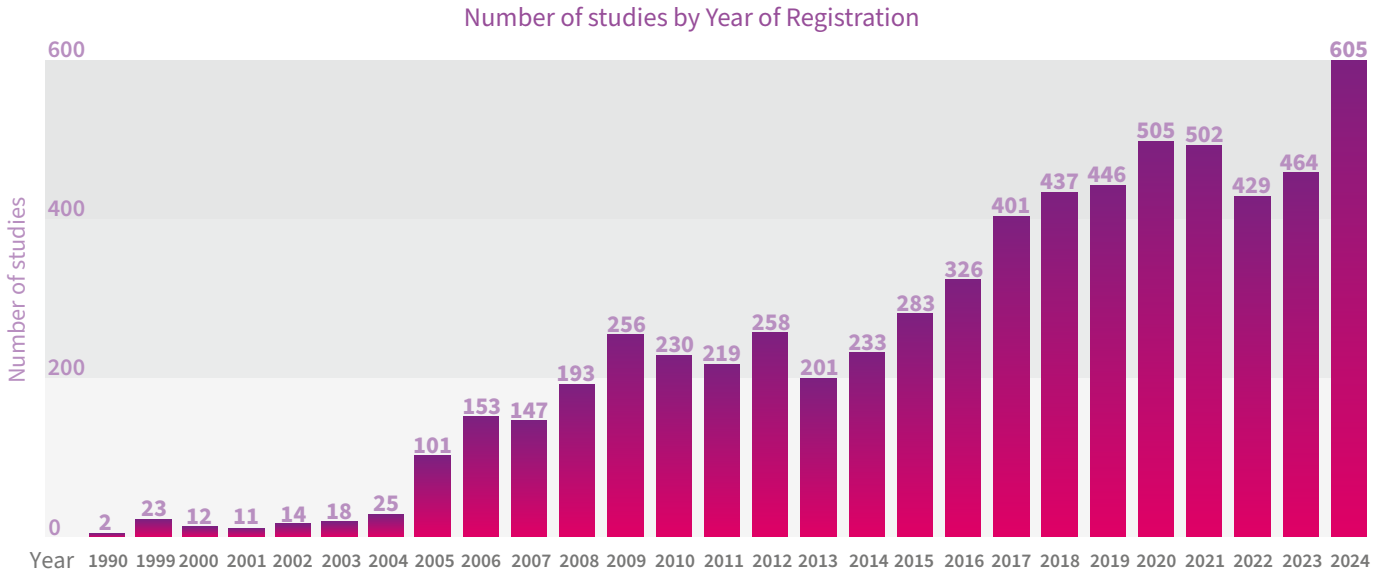


** WHO landscape of human genomics technologies in clinical studies [website]. Global Observatory on Health Research and Development; 2025 (https://www.who.int/observatories/global-observatory-on-health-research-and-development/monitoring/human-genomics-technologies-in-clinical-trials).*

1 Genomic clinical studies are expanding, with a notable increase in cancer and rare disease research

Global research activity in genomic technologies has expanded steadily over the past three decades, with a marked increase since the mid-2000s. This trend has followed the wider integration of next-generation sequencing into research and clinical workflows[1,8], alongside increasing investment in large-scale genomic initiatives across high-income countries[17,18].

Fig. 1. Number of genomic clinical studies registered annually, 1990–2024



Between 1990 and 2024, more than 6500 studies involving human genetic and genomic technologies were registered via WHO's International Clinical Trials Registry Platform (ICTRP). Study numbers began rising sharply after 2012, a pattern that reflects declining costs and improved instrumentation (Fig. 1). A more flexible degree of scalability, widespread availability of commercial reagent kits and comprehensive bioinformatics have made genomic technologies readily adoptable in both clinical and research labs[12,19,20].

Furthermore, the surge in trial registration through the 2010s suggests a turning point in the clinical adoption of genomics, particularly in oncology and rare disease research, which together now account for more than half of all genomic clinical studies. These areas have seen the earliest integration of genomics into clinical care, supported by clear use cases, regulatory incentives and a strong industry pipeline[21,22].

Clinical study activity fell off after 2021, a pattern that may reflect disruptions caused by the COVID-19 pandemic. During this period, global health systems faced widespread delays in clinical trial activation, recruitment and monitoring, with resource reallocation and logistic barriers affecting both new and ongoing studies[23]. In 2024 the number of studies using human genomic technologies appeared to surge, a trend which should be monitored in the future.

To better reflect current clinical research priorities and active development pipelines, we examined the subset of studies registered between 2016 and 2024. This period captures the most recent wave of genomic study activity and parallels typical study completion timelines, offering insight into contemporary applications of genomics in clinical research. The most frequently described uses of genomic technologies in these more recent studies were for screening and diagnosis (1129 studies) and targeted treatment selection (809 studies). Pharmacogenomic approaches were reported in 352 studies, followed by gene therapy and editing (104 studies) and clinical validation of genomic tools (257 studies).*

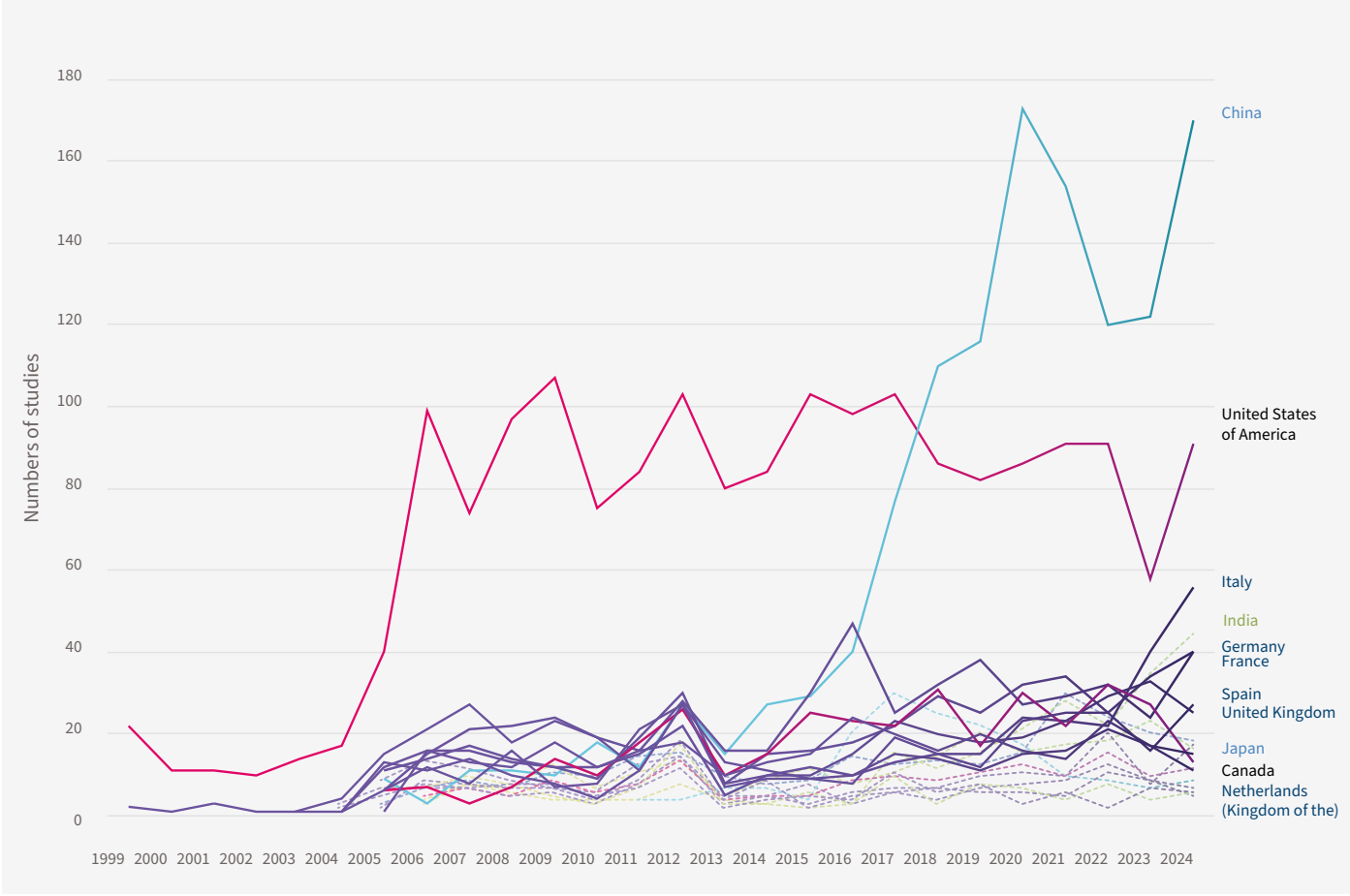
While the overall trajectory of genomics research reflects increasing technological readiness and translational ambition, it also raises critical questions about its focus and distribution, topics explored in the following sections.

**When categorizing genomic applications, only studies with sufficient detail to support clear assignment to a single category were included. Studies with ambiguous descriptions, overlapping applications or limited methodological information were excluded from these counts to avoid overinterpretation.*

2 Genomics clinical research is concentrated in high-income countries, with limited participation from low- and middle-income ones

Genomic clinical studies are overwhelmingly concentrated in a small number of countries. Although more than 6500 studies have been registered since 1990, most activity stems from high-income settings. While some countries have experienced sharp rises in study registrations in recent years, much of the global trajectory is driven by a handful of national programmes and reflects targeted investments in domestic sequencing infrastructure and research funding (Fig. 2).

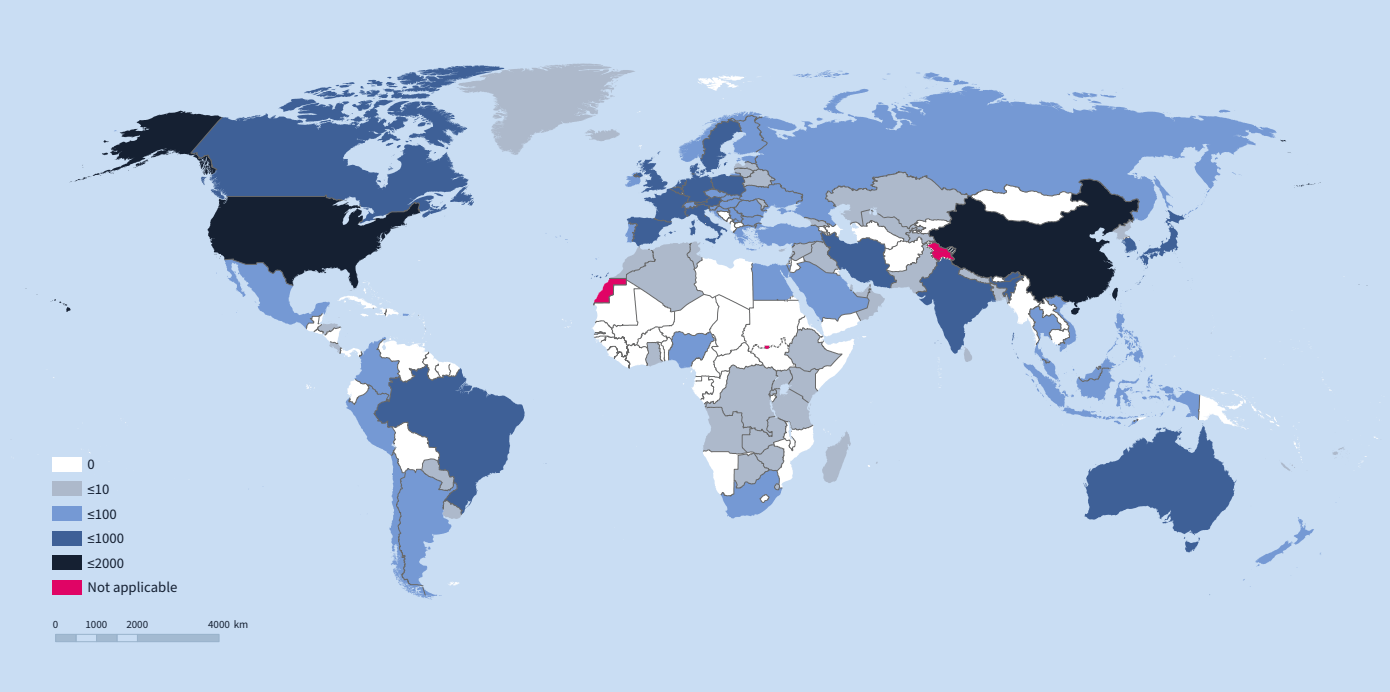
Fig. 2. Annual study counts per country, 1999–2024



Top 10 countries by total number of studies (solid lines) and top 11 to 20 countries by total number of studies (dashed lines).

This concentration is further evident if we look at the distribution of studies across the globe. Most studies are clustered in North America, Western Europe, East Asia and Australia, while large parts of Africa, Latin America, the Middle East and South Asia are minimally represented (Fig. 3).

Fig. 3. Geographical distribution of genomic clinical studies, 1990–2024

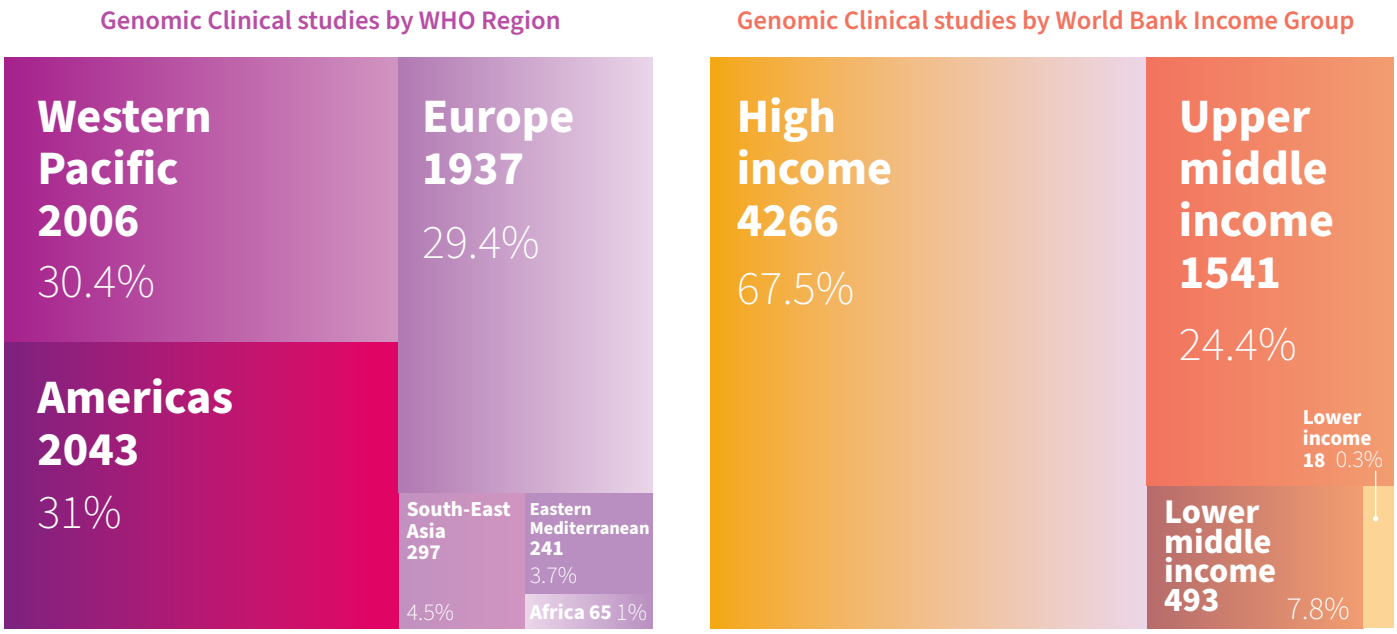


Viewed in terms of WHO regions (Fig. 4), the Region of the Americas, Western Pacific Region and European Region each accounted for roughly 30% of total genomic studies: 31%, 30.4% and 29.4%, respectively*. However, these regional totals mask important internal differences. Study growth in the Region of the Americas and Western Pacific Region has been largely driven by single countries, whereas in the European Region activity is more evenly distributed across countries. This highlights a key contrast in regional participation patterns, with implications for how research capacity is built and shared.

Low- and middle-income countries (LMICs) remain significantly underrepresented. Fewer than 5% of all studies were conducted in lower middle-income and low-income countries combined, while high-income countries accounted for over 80% of all genomic studies. Many LMIC studies took place within multicountry studies, with LMICs included as study sites but rarely as the leading partner: India figured in 235 studies, Egypt in 38, South Africa in 17 and Nigeria in 14. However, other LMICs appeared in fewer than five studies across the entire 34-year window.

**In accordance with resolution WHA78.25 (2025), Indonesia was reassigned to the WHO Western Pacific Region as of 27 May 2025.*

Fig. 4. Clinical studies using human genomic technologies by WHO region and World Bank income group



(Note: box size is proportional to the number of studies per category)

Patterns of disease distribution within LMIC studies also reflected broader disparities. In lower middle-income countries, over 60% of genomic studies focused on noncommunicable diseases such as cancer, rare diseases and metabolic disorders. Only 5% addressed communicable diseases, among which were solely 19 studies addressing infectious diseases in lower middle-income settings and two in low-income countries. This raises concerns about the alignment between genomic studies and local health priorities, especially given the high rates of infectious diseases in these regions as indicated by global burden of disease data.

The types of genomic technologies applied in LMIC studies were similarly skewed. In lower middle-income countries, the most common uses of genomics were for targeted treatment (93 studies), screening and diagnosis (74) and pharmacogenomics (46), with very few studies involving gene therapy (4 studies) or platform validation. These patterns are consistent with broader structural barriers: limited access to advanced sequencing technologies, few local genomic laboratories and constraints on funding for genomic research and development (R&D).

Addressing these gaps will require expanded investment in regional infrastructure, support for LMIC-led research and closer alignment of global research agendas with local disease burdens.

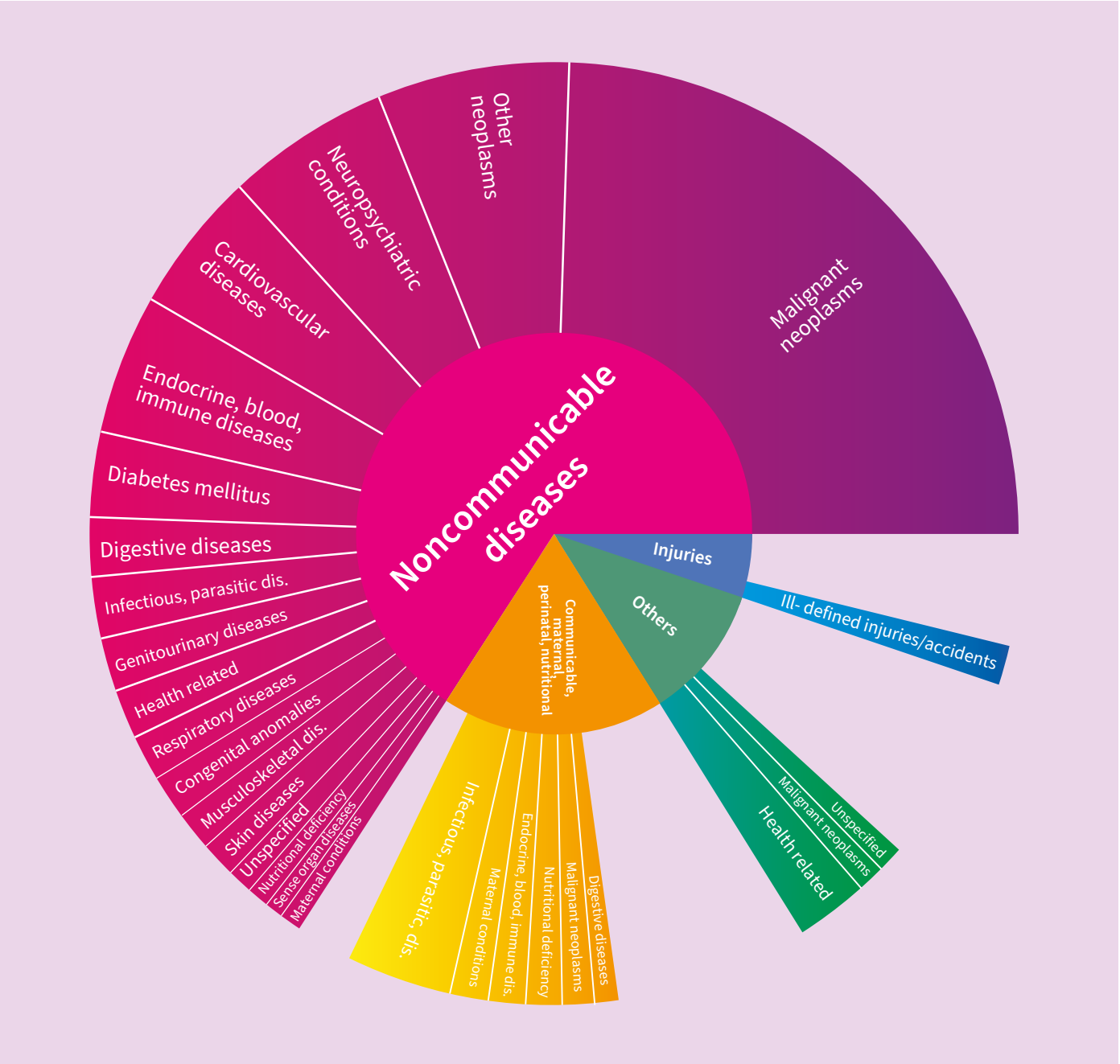
3 Cancer and rare diseases account for most genomic studies, but global health priorities remain underrepresented

The current distribution of genomic clinical studies reflects longstanding disparities in research prioritization across disease areas. While early integration of genomics into oncology and rare disease research has generated meaningful clinical advances, it has also created a narrow focus that may not be in lockstep with broader population health needs, particularly in low- and middle-income countries.

Cancer and rare diseases together accounted for more than half of all genomic studies. These areas were among the first to demonstrate clear use cases for genomics, including molecular subtyping, diagnostic confirmation and treatment selection. Their prominence is furthered by established clinical pathways, evolving regulatory frameworks and expanding precision medicine infrastructure in high-income settings[24,25]. At the same time, genomic technologies offer a unique opportunity to explore variability in disease expression and treatment response, particularly in conditions that do not fit neatly into existing diagnostic frameworks. This potential applies not only to rare and cancerous conditions, but also to more common conditions such as diabetes and infectious diseases, where genomic insights may uncover new biological subtypes or therapeutic targets.

While this study focused on clinical research using human genome data to the exclusion of clinical studies involving pathogen genomics, there are few existing studies of host responses to pathogen infection, or the genetic and genomics characteristics of these responses. In fact, communicable diseases accounted for just 3% of all genomic studies, despite their ongoing contribution to the global disease burden. Conditions such as tuberculosis, HIV and malaria continue to be major public health priorities in many low-resource settings, yet genomic studies investigating human susceptibility, treatment response or host-pathogen interactions are scanty. Where such studies do exist, they are often observational or diagnostic in nature and not always led by researchers in the countries most affected (Fig. 5).

Fig. 5. Distribution of disease categories represented in genomic clinical studies



This narrow range of disease categories reinforces existing inequities in the development and application of genomic tools. Populations affected by high-burden infectious diseases remain underrepresented in both clinical research and reference datasets, limiting the potential of genomics to support more inclusive and context-appropriate health interventions.

Broadening the clinical scope of genomic R&D and ensuring that research priorities better reflect the health challenges faced in low-resource settings will be critical to advance equity and maximize the global relevance of precision medicine.

4 Most studies involve an adult cohort, with a limited representation of children and older adults

Age and sex influence genomic expression, disease susceptibility and treatment response. Nevertheless, genomic clinical studies remain skewed toward adult populations, raising questions about the inclusivity and generalizability of emerging precision medicine approaches.

Of those studies which had available eligibility data, over 75% included adults aged 18–64 years. While many also permitted the enrolment of children or older adults, only 4.6% were explicitly designed to focus on paediatric populations and solely 1% included children alone. Studies designed exclusively for individuals aged 65 and over were similarly rare (3.3%). These patterns reflect longstanding barriers to inclusion, such as recruitment challenges, restrictive eligibility criteria and ethical or regulatory concerns in paediatric and geriatric research[26].

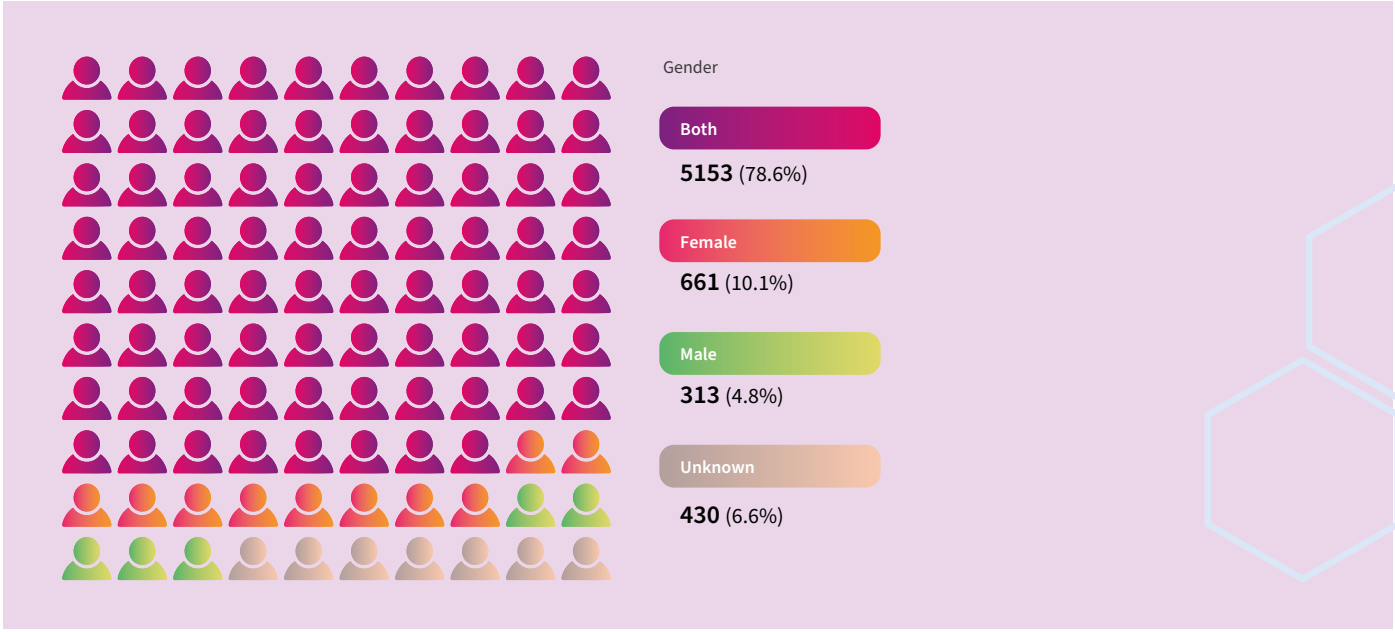
This limited focus was particularly notable in the context of rare disease research, which accounted for a substantial share of genomic studies. While many rare diseases have a paediatric onset, most studies enrolled participants across a wide age-range, rather than specifically targeting children. In these cases, especially for rare or heterogeneous conditions, broad age eligibility is often necessary to ensure adequate statistical power. However, inclusion does not equal representation: studies rarely stratified results by age or analyse age-specific outcomes, limiting the ability to tailor genomic tools to distinct life stages.

The predominance of adult-focused research has implications for downstream implementation. Risk prediction tools and treatment algorithms derived from adult populations may not translate to other age groups, potentially limiting clinical utility and contributing to health inequities[27].

Gender inclusion, by contrast, appeared to be more balanced. Of those studies reporting participant sex, most included both males and females. However, data disaggregation and sex-specific outcome reporting were rare, despite evidence of biological differences in genomic associations and therapeutic response[28,29]. Studies that explicitly addressed sex and gender diversity were virtually absent, limiting insights into these underrepresented populations (Fig. 6).

Improving demographic representation in genomic clinical studies will be essential for ensuring that the benefits of precision medicine are equitably distributed and scientifically valid across all population groups.

Fig. 6. Gender distribution of genomic studies



Conclusion

This analysis bears on both the expansion and uneven distribution of human genomic clinical studies globally. Over the past two decades, studies involving human genomic technologies have grown substantially, particularly in oncology and rare disease areas where clinical applications are well established and supported by evolving regulatory frameworks[24,25].

However, the benefits of this growth are not shared equally. Most studies are concentrated in a small number of high-income countries, and research remains focused on noncommunicable diseases. Low- and middle-income countries are underrepresented both in overall study activity and in genomic studies addressing locally relevant health priorities[3,15]. In addition, specific populations such as children, older adults and gender-diverse individuals remain systematically excluded from many genomic studies or are not explicitly addressed in the analysis of outcomes.

These trends raise concerns about equity, generalizability and missed opportunities to advance health outcomes more broadly. Ensuring that genomic research reflects a wider range of geographical, demographic and disease contexts will be essential to realize its full potential. This includes investing in infrastructure, fostering inclusive study designs and bringing research agendas in line with the public health needs of diverse populations.

As genomic technologies continue to advance, their meaningful integration into clinical research will depend not only on technical readiness but also on a commitment to equity and global relevance. This snapshot provides a foundation for identifying gaps and guiding future strategies to ensure that genomics contributes to health innovation for all.

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Unless otherwise specified, data for all visualizations and key messages in the report are taken from the Global Observatory on Health Research and Development (GOHRD). Data in this brief are derived from the interactive dashboard which can be accessed via the link below:
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