

Review Article

From genomic insights to public health impact: opportunities, challenges, and future directions in personalized disease prevention

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ABSTRACT

Genomic science is rapidly reshaping healthcare, offering unprecedented opportunities to prevent disease through precision approaches. When combined with traditional public health strategies, genomic data can enable earlier detection, refined risk stratification, and more targeted interventions, a vision often described as precision public health. Despite this promise, real-world integration remains limited. Methodological challenges such as ancestry bias, outcome heterogeneity, and inconsistent study designs undermine generalizability. Technical barriers, including fragmented data systems and poor interoperability, restrict scalability. Ethical concerns around privacy, equity, and governance further complicate implementation, particularly in resource-limited settings. This narrative review synthesizes current applications of genomic public health integration, including polygenic risk scores (PRS), infectious disease surveillance, and digital health initiatives. It highlights key opportunities for enhancing personalized disease prevention while critically examining persistent methodological, technical, and ethical barriers. Future progress will depend on diverse population inclusion, methodological standardization, interoperable infrastructures, and robust ethical frameworks. By addressing these gaps, genomic insights can move from research settings to scalable, equitable public health impact.

Keywords: Precision public health, Genomic data integration, Personalized disease prevention, Polygenic risk scores, Digital health, Artificial intelligence

INTRODUCTION

The rapid advancement of genomics has profoundly reshaped our understanding of disease mechanisms, shifting prevention strategies from generalized risk factors toward more personalized approaches.¹ Precision medicine, which leverages genetic profiles alongside environmental and lifestyle data, has emerged as a powerful tool for tailoring care to individual patients.²

This paradigm is increasingly extending into public health through concept of precision public health, which seeks to integrate genomic insights with population-level health strategies to enhance risk prediction, facilitate early disease detection and enable more targeted interventions.³

Emerging applications illustrate this potential. Whole-genome sequencing has been employed for outbreak investigation and pathogen surveillance, while PRS are

being explored to refine population screening and stratify disease risk.⁴ The convergence of genomics, digital health technologies, and artificial intelligence (AI) further expands opportunities for transforming disease prevention and public health practice.⁵ By harnessing biomedical big data, public health systems could move beyond one-size-fits-all interventions toward more effective, tailored strategies.^{6,7}

Despite these promising developments, integration of genomic data into public health practice remains limited and fragmented.⁸ Key barriers include data interoperability challenges, methodological weaknesses such as outcome heterogeneity and ancestry bias, and ethical concerns surrounding privacy, equity, and informed consent.^{9,10}

Resource constraints in many public health systems, especially in low- and middle-income countries, further complicate large-scale implementation.¹¹ Without addressing these challenges, there is a risk of widening health disparities and undermining public trust in genomic technologies.¹²

This narrative review synthesizes current knowledge on the integration of genomic and public health data for personalized disease prevention. It highlights both opportunities and limitations in existing approaches and discusses future directions for ensuring that genomic innovations are translated into equitable and scalable public health benefits.

GENOMICS IN PUBLIC HEALTH: CURRENT APPLICATIONS

The integration of genomics into public health has expanded considerably in the last decade. Key areas of application include:

PRS

PRS are being developed for conditions such as CVD (cardiovascular disease), diabetes, and cancer. They enable population stratification and more personalized screening, though their accuracy remains uneven across ancestries.^{13,14}

Infectious disease surveillance

Whole-genome sequencing has been applied to track transmission patterns of pathogens such as *Staphylococcus aureus*, influenza, and SARS-CoV-2, enhancing outbreak detection and response.^{15,16}

Oncology and chronic disease management

Integration of genomic profiling into cancer screening programs and pharmacogenomic clinics demonstrates the potential for tailoring interventions at both individual and population levels.¹⁷⁻²¹

Population screening initiatives

Pilot programs in North America and Europe, such as eMERGE and IGNITE, have tested large-scale integration of genomic data with electronic health records, providing models for future implementation.¹⁹

OPPORTUNITIES FOR INTEGRATING GENOMICS INTO PUBLIC HEALTH

Improved risk stratification with PRS

PRS are transforming population-level risk prediction by combining the cumulative effect of thousands of common variants into a single predictive model. Studies from large biobanks have demonstrated that PRS can significantly improve the prediction of cardiovascular traits such as systolic and diastolic blood pressure compared to traditional models.²² Resources like the polygenic score catalogue further enhance reproducibility and enable systematic evaluation across diseases.²³ PRS are already being tested in real-world clinical programs, where they support targeted interventions and prevention strategies.^{13,14,17} For example, their use in coronary artery disease and hypertension screening has shown promise in identifying individual years before disease onset.²⁴ From a public health perspective, embedding PRS into screening and surveillance frameworks could support more efficient allocation of resources, reducing healthcare costs and enhancing equity in disease prevention.^{19,25}

Early disease detection through genomic surveillance

Integration of genomics into public health surveillance offers an unprecedented opportunity for early disease detection and outbreak control. A clear example comes from pandemic where global genomic sequencing networks enabled rapid detection and characterization of SARS-CoV-2 variants of concern. South Africa's early identification of Omicron variant exemplifies how genomic surveillance can provide public health systems with critical lead time for intervention.²⁶ Genomic profiling is also proving valuable in oncology and chronic disease management, where precision medicine programs influence payer decisions and optimize treatment strategies.^{17,27} Additionally, tools such as KidneyIntelX test for early diabetic kidney disease show that genomic integration can change clinical management at population level.²⁷ By strengthening genomic surveillance infrastructure and embedding it into public health systems, policymakers can enhance preparedness and reduce the global burden of emerging diseases.

Integration of family history and electronic medical records

The combined use of genomic data with family history and electronic medical records (EMR) is another opportunity to improve disease prevention strategies.¹⁹

Studies show that family history assessments substantially increase predictive value of genomic testing, particularly when identifying individuals who may benefit from early screening.¹⁹ Meanwhile, integrating PRS and genomic results into the EMR platforms has been piloted successfully in large trials, such as within the EMR and genomics (eMERGE) network, with “which demonstrated feasibility and provided key lessons for the scaling precision medicine across the healthcare systems.”^{13,28}

These combined approaches allow for dynamic updating of risk profiles, ensuring that prevention strategies are both personalized and adaptive. For public health systems, such integration offers the potential to shift prevention from reactive treatment to proactive, continuous monitoring at the population level.

AI and big data analytics

AI and big data analytics offer powerful tools for extracting actionable insights from the vast volume of genomic and health-related data.²⁹⁻³¹ Biomedical big data technologies can improve disease modelling, identify novel biomarkers, and refine population-based prevention programs.¹² AI-driven algorithms, when applied to genomic datasets, enable the discovery of subtle interactions between genetic, environmental, and behavioural factors that influence disease risk. In addition, federated learning models allow collaborative use of multi-institutional data while preserving patient privacy, addressing one of the main ethical barriers to data sharing.³² For public health practice, these innovations support the design of precision interventions that are scalable, adaptive, and equitable, ensuring that populations benefit from cutting-edge analytics without compromising data confidentiality.

Strengthening global health innovation and equity

Genomics has the potential to reduce global health inequities when innovation is extended beyond high-income countries. Programs such as the human heredity and health in Africa (H3Africa) initiative demonstrate how regional capacity-building efforts can empower low- and middle-income countries (LMICs) to generate their own genomic data and develop local expertise.³³ Similarly, the NHLBI TOPMed program has sequenced diverse genomes, creating opportunities to broaden global reference datasets for improved transferability.³⁴

The IGNITE network is another example of how implementation research can connect genomics with real-world health system practice.³⁵ By investing in equitable genomic infrastructure and training, public health systems can ensure that the benefits of genomic integration are accessible globally, preventing a widening gap between high- and low-resource settings. This presents not just an

opportunity for scientific advancement but also for achieving fairer health outcomes across populations.

THEMATIC OVERVIEW OF THE LITERATURE

A synthesis of the literature reveals both progress and limitations in genomic public health integration.

Study quality and design

Cohort studies dominate evidence base, with many conducted in high-income countries. While findings are promising, limited population diversity raises concerns about global applicability.^{22,34}

Sources of bias and variability

Common issues include ancestry bias, inadequate confounder adjustment, and heterogeneity in outcome definitions. These methodological weaknesses hinder comparability and reproducibility.^{36,37}

Emerging strengths

High-quality studies demonstrate the feasibility of integrating genomics into practice, particularly when standardized frameworks, long-term follow-up, and validated outcome measures are used. Initiatives such as eMERGE and IGNITE highlight how genomic medicine can be embedded within health systems, though scalability remains a challenge.^{17,35}

METHODOLOGICAL AND TECHNICAL CHALLENGES

To visually summarize the methodological and technical challenges encountered in this study, an infographic was developed (Figure 1). Figure highlights key limitations such as ancestry bias, study design weaknesses, outcome heterogeneity, and data interoperability issues.

ETHICAL, LEGAL, AND SOCIAL CONSIDERATIONS

These are central to genomic public health integration:

Privacy and consent

Large-scale genomic data collection raises concerns about confidentiality and informed consent.³²

Algorithmic bias

Unequal representation in genomic datasets risks perpetuating disparities through biased predictive models.³⁷⁻⁴⁰

Equity of access

Without deliberate strategies, genomic innovations may disproportionately benefit high-income countries and privileged populations.

Governance frameworks

Transparent policies are needed to regulate data sharing, protect participants and ensure equitable benefit distribution.^{39,40}

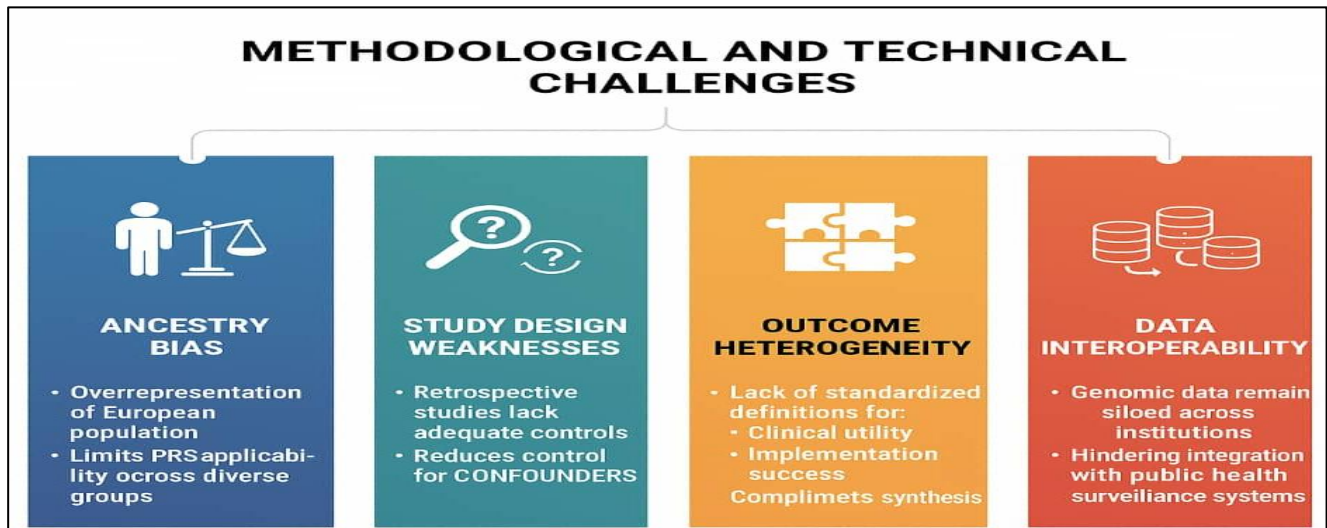


Figure 1: Methodological and technical challenges identified in the study.

FUTURE DIRECTIONS AND IMPLEMENTATION ROADMAP

To unlock the promise of precision public health, several steps are essential:

Methodological rigor

Prospective, multicentre studies with diverse populations are needed to improve generalizability.

Standardization

Adoption of frameworks such as STROBE for observational studies and RE-AIM for implementation outcomes will enhance comparability.^{32,41}

Digital transformation

Interoperability standards (e.g., FHIR, OMOP) and privacy-preserving technologies such as federated learning can enable secure collaboration across health systems.^{15,42}

Policy and governance

Clear regulatory frameworks should balance innovation with protection of privacy and equity.¹⁵

Capacity building in LMICs

Investments in infrastructure and workforce training are necessary to prevent widening global disparities.

CONCLUSION

The integration of genomic and public health data holds transformative potential for advancing personalized disease prevention. By combining genomic insights with population health strategies, precision public health can shift prevention from a reactive model to one that is proactive, targeted, and equitable. However, this promise will remain unfulfilled without deliberate action. Overcoming ancestry bias, fragmented data systems, and inconsistent methodologies requires global collaboration, methodological rigor, and digital transformation. Equally important are ethical and equity frameworks that safeguard privacy while ensuring broad access to innovation. Future progress depends on moving beyond isolated pilot studies toward scalable, interoperable, and ethically governed systems. With coordinated investment and inclusive strategies, genomic science can deliver not just medical breakthroughs, but population-wide improvements in health equity and disease prevention.

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