

Sustainable and equal integration of genomics into healthcare

Mónika Nogel

Deák Ferenc Faculty of Law and Political Sciences, Széchenyi István University, 9026 Győr, Hungary; nogel.monika@ga.sze.hu

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Abstract: Technological advancements in genetic research are crucial for nations aiming to uplift their population's quality of life and ensure a sustainable economy. Genomic information and biotechnology can enhance healthcare quality, outcomes, and affordability. The "P4 medicine approach"—predictive, preventive, personalized, and participatory—aligns with objectives like promoting long-term well-being, optimizing resources, and reducing environmental impacts, all vital for sustainable healthcare. This paper highlights the importance of adopting the P4 approach extensively. It emphasizes the need to enhance healthcare operations in real-time and integrate cutting-edge genomic technologies. Ecofriendly designs can significantly reduce the environmental impact of healthcare. Additionally, addressing health disparities is crucial for successful healthcare reforms.

Keywords: sustainability; genomics; health policy; P4; precision medicine

1. Introduction

New genetic technologies have enabled advances in genomic research, identification of disease-causing mutations, and personalized medicine approaches. Genomic medicine is a developing clinical field that incorporates an individual's genomic data into their medical treatment. This can encompass processes such as diagnosing conditions (Vogt et al., 2024), determining suitable treatments or interventions (Mesaki et al., 2024), and tailoring medications to address specific genomic variations (D'Gama and Agrawal, 2024). Its potential lies in transforming clinical practices by addressing the root causes of diseases rather than focusing only on the affected organ where symptoms appear (Zhang et al., 2019). This approach is poised to revolutionize healthcare, benefiting a wide range of stakeholders, including patients, providers, researchers, insurers, and policymakers (Fraser and Pai, 2014). One key advancement of the field is the rise of Next-Generation Sequencing (NGS) also known as Massively Parallel Sequencing (MPS), technology that enables rapid and cost-effective sequencing of large amounts of DNA or RNA. NGS has significantly transformed the landscape of healthcare in general, by offering more personalized and targeted approaches to disease prevention, diagnosis, and treatment. New insights gained in the field of biology, combined with the outcomes of the digital revolution, have led to a paradigm shift in healthcare known as P4-medicine. The four P-s are: personalized, predictive, preventative and participatory (Hood, 2013). This approach promotes the intersection of systems medicine, the digital revolution, and healthcare driven by consumer preferences (Pack, 2016).

Unlike traditional approaches, P4 medicine is proactive. It emphasizes disease prevention, addresses both individual and population health, and prioritizes wellness over illness. This framework relies on data-driven methods such as personalized "data clouds", extensive phenotyping, and the integration of physical, mental, and cognitive health (Horgan et al., 2014; Hood, 2019). Predictive, preventive, personalized, and participatory strategies are exemplified through interventions like vaccinations (Hsieh et al., 2024), COVID-19 genomic surveillance (Boffetta and Collatuzzo, 2022; Chiang et al., 2024), and innovative treatments for allergies (Innomata et al., 2020). Similarly, targeted therapies for cancer, including advancements in immunotherapy and precision oncology, further illustrate the impact of P4 medicine (Mahdi-Esferizi et al., 2023; Tsimberidou et al., 2024). A key real-world application is the UK's 100,000 Genomes Project, which integrates genomic data into healthcare to improve the diagnosis and treatment of rare diseases and cancer (100,000 Genomes Pilot Investigators, 2021). By sequencing the genomes of over 100,000 participants, this initiative has enabled clinicians to identify genetic variations linked to specific diseases, providing a foundation for more accurate diagnoses and personalized treatment plans. The project also fosters public trust by involving patients in decisions about how their genomic data is used, reflecting the participatory aspect of P4 framework. However, challenges remain, such as managing the vast amount of data generated and ensuring equitable access to the benefits of the project across diverse populations. Another groundbreaking application is the use of Chimeric Antigen Receptor T cells (CAR-T cell) therapy in cancer treatment (Dabas and Danda, 2023; Mitra et al., 2023). This personalized cancer treatment involves engineering a patient's immune cells to target and destroy cancer cells. It has shown remarkable success in treating refractory cancers like leukemia and lymphoma (Dabas and Danda, 2023; Mitra et al., 2023). CAR-T therapy exemplifies the predictive and personalized aspects of P4 medicine by tailoring treatments to an individual's genetic and immune profile (Zhang et al., 2023). Another example, gene therapy using Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR) technology represents a revolutionary approach to treating hemoglobinopathies, such as sickle cell disease (SCD) and β-thalassemia, which are among the most prevalent genetic blood disorders globally. Beyond its clinical efficacy, CRISPR-Cas9 technology holds several advantages over conventional gene therapy approaches. Unlike viral vector-based therapies, which carry risks of insertional mutagenesis, CRISPR-Cas9 provides a safer and more precise method of gene correction. Moreover, the adaptability of CRISPR technology allows for customization to address a broad spectrum of genetic mutations associated with hemoglobinopathies. As the first CRISPR-based therapies for hemoglobinopathies, such as CASGEVY (recently approved both by the U.S. Food and Drug Administration and European Medicines Agency for sickle cell disease and β-thalassemia), move closer to routine clinical use, they pave the way for a new era of precision medicine (Parums, 2024; Singh et al., 2024).

With increased adoption of predictive, preventive, personalized, and participatory approaches, innovations in data storage and sharing will further facilitate genomics integration into healthcare (Kwon et al., 2018). However, challenges like disparities in access, limited diversity in genomic datasets, and ethical concerns remain barriers to achieving equitable and sustainable healthcare systems.

2. Sustainable healthcare

The concept of sustainable healthcare is becoming increasingly important. This growth is driven by the recognition that healthcare activities are closely linked to environmental, social, and economic factors. However, there is no universally agreedupon definition of 'sustainable healthcare.' Similarly, healthcare organizations lack a comprehensive framework or standardized guidelines to pursue sustainability. As a result, the term 'sustainability' is used in varied ways within the healthcare field. This diversity creates challenges and uncertainty for managers and organizations. Alliance for Natural Health defines 'sustainable healthcare' as "a complex system of interacting approaches to the restoration, management and optimization of human health that has an ecological base, that is environmentally, economically and socially viable indefinitely, that functions harmoniously both with the human body and the nonhuman environment, and which does not result in unfair or disproportionate impacts on any significant contributory element of the healthcare system" (Alliance for Natural Health in 2018). Sustainable healthcare is commonly known also as eco-friendly healthcare or green healthcare, signifying the delivery of health services in a manner that prioritizes the environment, aiming to enhance well-being and benefiting the community (WHO, 2017). This paper argues that defining sustainable healthcare is not essential. Instead, its components can be identified by determining its key prerequisites (**Table 1**).

Table 1. Key elements of sustainable healthcare**.**

3. Future of genetics and genomics in healthcare

In the last couple of years, the COVID-19 pandemic has revealed deficiencies in the ability to respond adequately to public health emergencies. In order to address these shortcomings, several health policy concepts have been formulated. Certain ones align with the objective of involving healthcare systems in the benefits of the new genomic era. The Global Genomic Medicine Collaborative policy working group for instance has used several strategies to elucidate the approaches that different countries were taking to move genomics from research into routine clinical care. Between 2017 and 2020, 65 initiatives have been identified, and a catalogue of these initiatives is publicly available at genomicspolicy.org. These initiatives range in scale, from small studies to large national and global cohorts involving 1 to 5 million participants. These undertakings employ diverse testing methods, encompassing copy number variation, microarrays, gene panels, whole-exome sequencing, whole-genome sequencing, RNA sequencing, and mitochondrial DNA sequencing. The scope of diseases studied is broad, covering rare, common, and infectious conditions, as well as pharmacogenomics research in both adult and pediatric populations. The initiative can be considered appropriate, because analyzing factors like geographic locations, initiative types, and policy interests reveals several insights into the implementation of genomic medicine (Belcher et al., 2020) and of P4 medicine in general.

To address challenges comprehensively, it is vital to consider the broader framework of sustainable development. Genomic medicine aligns closely with global sustainability goals, particularly in enhancing health outcomes while minimizing environmental and social disparities. Personalized approaches, such as tailoring treatments to genetic profiles, optimize resource utilization and reduce waste, contributing to environmental sustainability. Furthermore, genomic advancements support global health goals, such as the United Nations' Sustainable Development Goal (SDG) 3, which emphasizes ensuring healthy lives and promoting well-being for all. By addressing non-communicable diseases, rare genetic disorders, and disparities in healthcare access, genomics-driven medicine plays a crucial role in advancing equity. Similarly, investments in genomic research and infrastructure stimulate innovation and economic growth, aligning with SDG 8, which focuses on sustainable economic development and job creation. By embedding genomic medicine within the sustainable development framework, healthcare systems can ensure that advancements contribute to a healthier, more equitable, and environmentally sustainable future.

Expectations suggest that continued scientific progress will lead to unprecedented growth in genetics and genomics. Most importantly, this growth will entail fostering collaborations between physicians, scientists, genetic counselors, and clinician-scientists to bridge the clinical context and content. Genetic tests will become more accurate, results will be available sooner, and interpretation will be simplified through the utilization of artificial intelligence.

The generation of extensive genetic and genomic data will persist at a remarkable rate. Ancestry will likely play a critical role in understanding disease origins and the principles of aging. This knowledge will refine risk assessments and improve medical management. Additionally, the ease and affordability of monitoring personal vital signs in real-time and longitudinally will likely result in integrating these comprehensive phenotyping endeavors into each patient's electronic medical record (EMR) (Yehia and Eng, 2019). Pharmacogenomics and genomics-driven treatments will enable personalized drug selection and dosing before starting a regimen, optimizing therapeutic outcomes. Alongside these advances, research into gut and organ-specific microbiomes will improve predictions of disease outcomes and treatment responses (Asnicar et al., 2021; Gupta et al., 2020). It is important to highlight that data sharing appears as a precondition of the success of this goal (Le Texier et al., 2019). Nevertheless, although there is a global increase in scientific output concerning data sharing in P4 framework (Guérin et al., 2021), there are many differences in quantity and quality of the data. A shift in approach is needed to fully leverage this data. Collaborative networks involving researchers from diverse disciplines such as engineering, computer science, and industry are critical to ensure reliable insights from Big Data. Continuous updates to infrastructure and ethical regulations are necessary. Crucially, this must be achieved without implementing stringent measures that could jeopardize the essential access required for progress to thrive (Hulsen, 2019). The integration of genomics into clinical care faces several barriers. These include challenges in managing and interpreting data, workforce training, public acceptance, government involvement, and limited evidence of clinical utility and cost-efficiency (Hoenders et al., 2024; Manolio et al., 2015). Additionally, ethical and social concerns, such as genetic privacy, informed consent, and equitable access, remain critical. Expectations for data-driven personalized medicine in Europe are high, but numerous structural and legal challenges limit access to valuable data for research. Key legal challenges include intellectual property rights, confidentiality, and data protection, which create tensions between transparency and the protection of proprietary information. Optimizing regulatory tools in complex, data-driven research ecosystems requires policy measures that align stakeholder goals and ensure effective participation (Rajam, 2020). The legal, ethical, and social challenges of P4 medicine in general encompass navigating issues related to genetic privacy, informed consent, equitable access, and the potential for exacerbating health disparities (Flores et al., 2013). One of the most important expectations is that genetic medicine should be equally accessible. This is significant not only due to the guarantee of human rights but also because of the accuracy of data. Examining genotype-phenotype relationships across populations enhances understanding of health and disease, aiding in the identification of at-risk groups. However, diversity in genetic testing remains a significant issue. One indicator of this is the fact, that according to the Genome-Wide Association Studies (GWAS) Diversity Monitor's data, that aims to improve the equitability and diversity of participants across all published GWAS, in October of 2024 more than 94% of total participants in GWAS studies were of European ancestry, other groups are highly underrepresented (Mills and Rahal, 2020). This underrepresentation highlights the pressing need to involve diverse populations extensively in genetic testing to improve our understanding of health and disease across all groups. Naturally, in healthcare, it's not exclusively GWAS data that holds relevance, but it doesn't seem overly bold to conclude that even in the realm of genetically narrowed tests for specific markers, the composition of participants might not be significantly more diverse. The reasons for the situation could be different. In

recent years, numerous studies have been published on the attitudes towards genetic tests and the level of trust in genetic testing within the general population. The results indicate that several factors influence attitudes toward genetic tests and the level of trust in genetic testing within the general population: transparency, accuracy, privacy and data security, regulation and ethics, personal experience, access and equity, etc. (Hanneman, 2013). Looking ahead, ensuring equitable access to genetic testing for healthcare purposes is vital. Populations excluded from genetic testing risk being left behind in the distribution of scientific advancements, potentially deepening healthcare disparities across different groups and regions. Health inequalities can lead to poorer health outcomes, restricted access to care, and broader social and economic burdens. By addressing these disparities, healthcare systems can align with global goals of inclusivity and resilience, paving the way for a more just and effective distribution of genomic innovations.

The emergence of direct-to-consumer genetic testing (DTC-GT), which allows individuals to undergo genetic testing without the involvement of healthcare professionals, offers consumers a convenient and cost-effective way to access and take ownership of their genetic data analysis results. There are two contrasting views on direct-to-consumer genetic testing (DTC-GT). One perspective highlights the potential health benefits, such as insights into disease risk and drug sensitivity, and the convenience and affordability of obtaining genetic information. The other focuses on concerns like privacy, accuracy, and the ability of consumers to interpret the genetic data. Effects of DTC-GT on health outcomes are largely influenced by consumer characteristics such as education, familiarity with healthcare management, and trust in genetic testing (Jeong, 2018).

The WHO Traditional Medicine Strategy 2014–2023 emphasizes the importance of integrating traditional and complementary medicine (T&CM) into national health systems. It highlights the need for countries to understand the use, benefits, and risks of T&CM to develop informed and effective policies. The strategy also stresses the significance of ensuring the safety, quality, and efficacy of T&CM practices and products through regulatory frameworks and practitioner qualification standards. Furthermore, it advocates for equitable access to T&CM as part of universal health coverage, aligning with broader global health goals, including personalized and predictive care. By encouraging sustainable resource use and recognizing the value of diverse healthcare practices, the strategy supports efforts to enhance cultural sensitivity and environmental sustainability in healthcare. Its framework for integrating T&CM into mainstream health systems provides a model for incorporating innovative approaches, such as genomic medicine, into healthcare delivery (WHO Traditional Medicine Strategy 2014–2023).

To address the challenges related to genomics-driven medicine mentioned above, in 2022, WHO Science Council made 15 recommendations for WHO and for consideration by multiple sectors within its Member States (WHO, 2022). The recommendations are grouped under four themes (**Table 2**).

The healthcare sector plays a significant role in contributing to pollution and greenhouse gas emissions, which are major drivers of global warming. It has recently been highlighted that the healthcare industries of countries like the United States, Australia, England, and Canada together produce an annual total of 748 million metric tons of carbon dioxide equivalents (Sherman et al., 2019). Several initiatives have been launched by academics, hospital systems, industry partners, non-governmental organizations and governmental bodies to address the sustainability in the healthcare agenda (https://research-and-innovation.ec.europa.eu/research-area/health/environment -and-health en). As the healthcare construction industry begins to resume (from its relative pause during the COVID-19 pandemic), there is also a need—and opportunity—to apply green design principles that could help significantly reduce the healthcare footprint. Research emphasizes that applying findings from implementation science, pursuing interdisciplinary strategies, and integrating environmental economics are key measures for advancing sustainability objectives in healthcare (Hu et al., 2022). To address the current and future challenges in healthcare, such as the rise of non-communicable diseases, emerging epidemics, and the impact of climate change and pollution, coordinated global actions and sustainable development are crucial. One notable advancement is the development of energy-efficient healthcare facilities. These leverage renewable energy sources, advanced insulation technologies, and energy-saving designs to minimize their carbon footprint (Vosshenrich and Heye, 2023). Similarly, green building certifications such as Leadership in Energy and Environmental Design (LEED) and Building Research Establishment Environmental Assessment Method (BREEAM) have been adopted globally to ensure that hospitals and clinics adhere to sustainable construction and operation standards (Kar et al., 2024). Waste reduction programs, particularly in surgical and pharmaceutical contexts, also demonstrate measurable environmental benefits. By engaging suppliers to prioritize sustainable practices, healthcare facilities can reduce their carbon

footprints and promote the growth of eco-friendly industries (Kar et al., 2024). 3D printing technology in pharmaceuticals exemplifies innovation driving both personalization and sustainability. This technology allows for the on-demand production of personalized medications, tailored in dosage, size, and release profiles. Unlike traditional manufacturing methods that require large-scale production and generate significant waste, 3D printing enables pharmacies to produce medications locally, reducing the need for transportation and storage. Research highlights that 3D printing could revolutionize medication delivery systems, offering a sustainable solution to meet patient-specific requirements (Aquino et al., 2018). Another example is the use of personalized medication dosing enabled by molecular medicine. By tailoring prescriptions to individual genetic profiles, this approach reduces overmedication and minimizes pharmaceutical waste. Studies have shown that such precision dosing can cut down on the production of unused or ineffective medications, contributing to both economic and environmental sustainability.

Additionally, digital platforms play also a significant role in shaping public health behaviors. While these platforms can be valuable tools for spreading accurate health information and promoting positive health practices, they also have the potential to misinform the public. This misinformation can lead to negative outcomes, such as poor adherence to long-term treatments or increased vaccine hesitancy, both of which can undermine public health efforts (Singer and Redekop, 2020).

Achieving the full potential of precision medicine requires a holistic understanding of its broader context. This includes addressing technical, organizational, and policy-related factors that influence its implementation. While high expectations for precision medicine can drive significant investment, successful adoption depends on effective coordination across various disciplines. A clear understanding of the system's complexity is essential for integrating new technologies into healthcare. Developers must consider all aspects of the healthcare ecosystem, including infrastructure, workforce capabilities, and patient access, when evaluating the real-world impact of these innovations (Redekop et al., 2018).

4. Policy implications

Successfully integrating genomics into healthcare systems requires well-crafted policies addressing technical, ethical, and social aspects (Özdemir, 2024). Policymakers play a crucial role in ensuring equitable, sustainable, and effective implementation of genomic advancements. Key actions include increasing funding for research that promotes diversity in genomic datasets and fostering interdisciplinary collaborations to translate research into practical healthcare solutions (Dharani and Kamaraj, 2024). Public-private partnerships can drive innovation by pooling resources and applying genomics to clinical and research purposes. Additionally, governance frameworks must align genomic advancements with environmental, ethical, and social sustainability goals, emphasizing international collaboration and robust infrastructure development (Cinti et al., 2024).

Ensuring that advancements in genomics benefit all populations requires addressing structural and social barriers that worsen inequality. Policies that mandate diversity in research funding can encourage the inclusion of underrepresented groups.

To make these efforts effective, community-based recruitment strategies and partnerships with local organizations can increase participation while ensuring studies are culturally sensitive and inclusive. However, promoting diversity in research is only one aspect of achieving equity. Equitable access to genomics-driven care also requires overcoming financial barriers. Subsidizing genomic testing and treatments for lowincome and underserved populations is essential to ensure these advancements are not limited to wealthier groups. Governments and healthcare systems should establish funding mechanisms to reduce out-of-pocket costs, particularly for patients in rural or economically disadvantaged areas. In addition to financial subsidies, investments in expanding genomic services in regions with limited infrastructure are necessary to address broader systemic inequities. Yet, access alone is insufficient without fostering trust and understanding among the populations that stand to benefit. Public education and engagement are key to bridging the gap between genomic advancements and the communities they serve. Many populations harbor mistrust of genetic research due to historical injustices or a lack of familiarity with its benefits. Transparent communication about the goals, risks, and potential outcomes of genomic initiatives is critical for building trust and fostering participation. Educational campaigns should use accessible language and culturally relevant examples to resonate with diverse audiences. Furthermore, engaging community leaders and advocates can amplify the credibility and reach of these initiatives. Public trust, however, depends not just on education but also on strong ethical safeguards. Ethical and social considerations are central to addressing health disparities in genomic medicine. Policies must protect genetic privacy and prevent discrimination, particularly in employment and insurance contexts. Strengthening informed consent processes ensures individuals fully understand the implications of participating in genomic studies or receiving genomicbased care. Establishing oversight bodies to monitor these initiatives is essential to identifying and addressing unintended consequences that may exacerbate inequities. Ethical safeguards and transparency go hand in hand with building collaborative frameworks. (Bilkey et al., 2019; Curtin et al., 2022; Equils et al., 2023; Khouri et al., 2022)

Building public trust is critical for the success of genomic initiatives. Transparent communication about the goals, risks, and benefits of genomic medicine can dispel misconceptions and foster acceptance. Educational campaigns must use accessible language and involve community leaders to ensure diverse perspectives are represented (Acosta et al., 2022; Therianou et al., 2022; Xu and Wu, 2024). Public engagement campaigns should not only inform but also invite input on policies that affect communities, strengthening their relevance and credibility.

Genomic medicine also raises complex ethical, legal, and social challenges. Safeguarding genetic privacy, preventing discrimination, and ensuring informed consent are fundamental to its equitable adoption. Oversight committees should monitor ethical issues and ensure alignment with societal values. These measures are crucial to addressing challenges related to the use and sharing of genomic data, particularly in employment and insurance contexts (Equils et al., 2023). International cooperation is indispensable for addressing cross-border ethical and legal challenges, such as data-sharing agreements and disparities in regulation (Babu et al., 2024).

Effective integration requires secure and ethical data-sharing mechanisms. Comprehensive governance frameworks must balance privacy with transparency, enabling standardized data collection and interoperability across institutions and countries. Open-access initiatives should be incentivized to extend the benefits of genomic advancements globally (Roberts et al., 2024). Policymakers must also consider technical challenges, such as developing IT systems and biobanks to support data-sharing and storage, while ensuring the data is managed responsibly (Bilkey et al., 2019).

By aligning strategies at national and international levels, governments, researchers, and healthcare organizations can harmonize policies to reduce health disparities. Sharing best practices, pooling resources, and coordinating efforts can strengthen equity in both research and clinical applications (Boffa et al., 2024). Policymakers must prioritize research, support data-sharing, ensure equity, foster public trust, and address ethical concerns to unlock the full potential of genomicsdriven medicine. A comprehensive approach will enable healthcare systems to harness genomics for sustainable, inclusive, and patient-centered care worldwide.

5. Conclusions

Merging sustainability and genomics presents a complex and multifaceted concept. As a result, implementing genomics into healthcare in a sustainable manner requires a thoughtful and comprehensive approach. It is necessary to consider that stakeholders will need to increase data sharing in order to leverage the full potential of genomic medicine. Promoting public awareness and engagement in genomic science involves various strategies and communication efforts. The whole process should be driven by ethical principles. Furthermore, preventing the exacerbation of health inequalities due to new biotechnologies and information communication technologies requires careful planning and considering their impact on health systems' sustainability. By targeting genetic predispositions and individual variations, genomics-driven medicine can lead to better patient outcomes, minimizing the environmental burden associated with ineffective interventions. This aligns with the principles of sustainability by promoting efficient resource allocation and reducing medical waste. The combination of green healthcare and genomic based healthcare is crucial for the progress of sustainable healthcare systems. P4 medicine, with its potential to tailor treatments, can contribute to greener healthcare by optimizing resource utilization. By blending eco-friendly practices with the integration of predictive, preventive, personalized, and participatory approaches, there is potential for effective, patient-centric healthcare that enhances health results while reducing environmental effects, ultimately contributing to a more sustainable healthcare environment.

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