Large-Scale Genomics Applications in Public Health and Precision Medicine Using Big Data and Predictive Analytics

Debarghya Biswas[1], Ankita Tiwari[2]

[1]*Assistant Professor, Department of CS & IT, Kalinga University, Raipur, India*
[2]*Research Scholar, Department of CS & IT, Kalinga University, Raipur, India*

KEYWORDS
Genomics Applications, Precision Medicine, Public Health, Big Data

ABSTRACT

After gaining experience from large population sequencing projects, there is a significant gap between the promise of genomics in Precision Medicine (PM) and the existing state of affairs. To fully exploit this potential, Public Healthcare (PH) must adopt procedures, regulations, and technology that are unfamiliar to them. The development needs to be improved by several historical procedures and regulatory hurdles. In the future, PM will include making genetic big data easily accessible at the point of treatment and implementing tools to govern its use effectively. Significant modifications need to be made in billing, payment, and monitoring to accomplish this goal. New systemic and technological structures must also be developed inside the healthcare sector. The responsibilities of clinical geneticists will expand to include overseeing PM systems. Genetic counselors will guide and assist PM by establishing and upholding PM structures. The trajectory of the research is now hindered by several impediments, resulting in avoidable fatalities. Revamping medical facilities to accommodate genomics has the potential to influence the treatment of patients significantly and enable the achievement of long-awaited advancements in PM.

1. Introduction

Precision Medicine (PM) aims to transform healthcare by integrating specific person-level attributes (such as genetic, lifestyle, and environmental variables) into medical interventions and preventive measures to deliver appropriate care to each individual at the optimal moment [1]. This technique enables the implementation of preventative or therapeutic medical treatments that might be more efficient than generic approaches, enhancing the standard of care and minimizing wasteful diagnostic tests and medicines [2].

PM is a methodology that combines precision and population-based tactics to deliver the right measure to the correct population at the optimal moment [3]. This encompasses expanding PM efforts and applying essential science findings to the broader public. PM is characterized by its ability to create more precise techniques for identifying multi-level risk variables and their effect on Public Health (PH). This knowledge can establish particular programs and policies promoting health [4, 5].

Including human genomics studies in precision, PH efforts have helped develop more accurate strategies for preventing and controlling diseases [6, 8]. This includes activities like newborn screenings and targeted prevention measures, such as increased cancer detection for individuals with harmful gene mutations [7]. The use of human genetic research in implementing PM has been restricted. The several core areas of health care, including biostatistics, ecological health, epidemiology, medical policy and offerings, and social and psychological health, have complemented prospects for enhancing PM applicability via human genomics [9]. During the deployment process, the research encountered significant obstacles that hindered the complete implementation of PM, as seen by the substantial shortcomings. This viewpoint offers a clear vision for using PM in everyday clinical practice and recommendations to accelerate its adoption. This perspective is derived from the involvement of genomics in the United States medical system. Some of these obstacles arise in medical systems in other countries.

Background

In the future, genetic testing will be simplified to a single comprehensive test utilized for all reasons during a patient's lifetime, eliminating the need for many panels of tests. The big data obtained from

this test could be easily accessible at the location where medical treatment is provided for a wide range of applications, including, but not restricted to:

- Pharmacogenomics involves prescribing pharmaceuticals based on a person's genetic pattern to ensure the most appropriate prescription and dose are used for the best outcomes, considering their genetic traits [10].
- Gene-based medicines include identifying individuals with diseases with genetic variations that can be effectively treated with gene therapy or biologics and then commencing those specific treatments.
- Disease prevention—Identifying and implementing measures to avoid disease for people who carry harmful genetic variations in genes is called increasing the risk of preventable diseases, as the American College of Medical Genetics (ACMG) suggested [12].
- Disease risk estimation involves using scoring systems to forecast the likelihood of a patient developing a disease and facilitate preventive measures [13]. These models involve polygenic risk ratings and more intricate models that integrate genetic big data with additional patient characteristics and environmental facts to forecast and avert illness [11].
- Real-time genetic diagnostics refers to diagnosing genetic conditions in real-time when individuals present symptoms and seek medical treatment at hospitals or clinics—facilitating the use of expedited and more efficient therapeutic alternatives due to enhanced precision in diagnosis.
- PM genomics refers to medical facilities' use of genomic information to tailor illness management techniques to the genetic disorders of the people they serve [15].
- Reproductive decision-making—Utilizing genomic big data to assist persons contemplating parenthood in comprehending carrier concerns and making well-informed reproductive choices, which might involve in vitro fertilization and preimplantation genetic diagnostics.

The genetic information will be sent with the patient to several medical facilities located in different states or nations. The practical use of this big data would mainly depend on authorized bioinformatics technologies that do not need human evaluation and approval by medical supervisors. This allows for the cost-effective and prompt exploitation of genetic big data.

Advanced Clinical Decision Support Schemes (CDSS) utilize genomic information to enhance physician decision-making, guiding through specialized care pathways based on genetic information [16]. These systems adapt prescriptions based on a patient's anticipated medication response and schedule preventive treatment and disease observation. Portable and wearable technologies implement similar systems to track and direct PH outside the professional environment. This allows patients to engage in their treatment actively. Advanced Artificial Intelligence (AI) systems using genomes analyze patient trajectories in a sophisticated manner and enable adjustments to help patients achieve their ideal health goals.

Medical geneticists are increasingly taking on administrative positions supervising the implementation of genomes and PM in medical facilities. Medical geneticists could be restricted in diagnosing and treating uncommon illnesses since many genetic problems would be assigned to other medical disciplines. Similar to nurses, genetic professionals would be widely available to support the implementation and administration of PM systems.

Integrating Human Genomics Research and PH

The research outlines the methods by which the five areas of PH supported by the Association of Universities and Programs of PH have included the study of human genetics into precision PH study. The five fields include biostatistics, ecological health, epidemiology, health policies and services studies, and social and psychological health.

Biostatistics

Biostatistics is the field that focuses on the examination of ideas and methodologies for gathering, analyzing, and decoding big data that is pertinent to PH. The extensive magnitude and scope of the databases utilized for PM studies need sophisticated statistical approaches with improved techniques for minimizing interference and improving the identification of signals. These datasets contain multidimensional genomic biomarker information collected from many people, with multiple measurements taken throughout time. It will be necessary to develop sophisticated statistical techniques to estimate the connections between individual-level variables (such as genetics and behavior) and macro-level variables (such as environmental encounters and regulations) and detect hidden connections using AI.

Environmental Health

Environmental healthcare is concerned with the interaction between humans and their environment to encourage good health. The discipline of ecological healthcare has extensively observed that individual-level elements, such as genetics, communicate with macro-level ecological influences to influence the health of populations [14]. Integrating human genomic research into environmental health studies can show how individual-level variables, such as -omics information and macro-level factors, communicate to impact health. This can lead to a deeper understanding of the processes that drive complex gene-environment conversations. Further research is needed to comprehend the underlying processes of intricate ecological exposure and how they relate to individual-level characteristics. This research paves the way for environmental treatments targeting groups with a significant likelihood of adverse effects.

Epidemiology

Epidemiology employs analytical techniques to evaluate the distribution and etiology of PH conditions, including individual-level attributes (such as genetics) and macro-level variables (such as neighborhood and communal features). PM care utilizes epidemiological techniques, such as population-level monitoring, to enhance understanding of various health hazards at different levels. This information can be used to develop targeted assessment, therapy, and preventive strategies.

PH policy and services research

Healthcare policy and medical services study focuses on evaluating the quality, affordability, and accessibility of healthcare for the general population. Collecting this big data is crucial for comprehending PM interventions' financial and health consequences, especially their potential to promote PH equality. Genomic testing advancements in prevention and therapy need healthcare studies to assess the medical usefulness, cost-effectiveness, and patient-reported results.

Social and Behavioral Research

The analysis examines how behaviors and the social factors that influence everyday living (social healthcare variables) impact health throughout a person's life. Precision PH studies have mainly examined the ethical, authorized, and societal consequences of human genomics studies. Research can enhance public confidence and willingness to use treatments targeting their genetic vulnerabilities.

Challenges

Challenging the academic approach

Identifying novel mutations that induce pathological conditions in a well-characterized gene receives little scholarly focus. Some of the peers have claimed that securing financing for big data functional investigations is challenging due to losing their originality. Due to the incapacity to disseminate such discoveries, there needs to be more motivation to enhance public awareness of gene variation. As they are being created, they should consider the motivations of healthcare professionals, safeguards for

patient confidentiality, and the regulations set by Institutional Review Boards (IRBs) to reduce obstacles to publishing. Most doctors will wait to draft an IRB procedure to present these results formally.

Technical architecture requirement

Previous research has shown the shortcomings of technology processes in supporting PH, particularly where laboratories and the clinic intersect. These steps are crucial for the successful implementation of scaling PM. There is still a considerable amount of work that has to be done in this field, especially in the field of implementation research.

Prioritization and management of technical systems

With genetic details' increasing availability and importance in medical treatment, medical facilities that have yet to adopt such designs will face challenges in handling big data and its clinical consequences. This led to less-than-ideal medical treatment and perhaps legal responsibility. Medical geneticists are now doing these functions in medical facilities that have advanced in this field without official acknowledgment or designation.

Reimbursement challenges

Genetic analysis and medical genetics interactions have received inadequate reimbursement, resulting in significant financial losses for many medical genetics divisions. To become a clinical geneticist, one must do an extra two years of study after completing their primary specialization. This profession's financial remuneration is often lower than in the prerequisite area. As a result, a group of people are mainly motivated by scientific curiosity and the desire to assist patients, with financial remuneration being a secondary consideration. Instead of imposing restrictions on the number of genes examined or disclosed to patients, the priority should be establishing systems to handle this big data and assisting patients and healthcare professionals in navigating treatment plans. This dilemma will become more challenging to address as the understanding of genomics expands.

Regulation

Although a certain level of regulatory monitoring is necessary as PM becomes more prevalent in the medical field, big data regulation can impede advancements. While rules are often intended to safeguard patients, they might inadvertently favor large industry leaders by imposing substantial obstacles to entering the marketplace, excluding more minor, more inventive competitors. This hampers innovation by diminishing competitiveness. Regulating is essential and provides safeguards for patients, but it needs to be implemented cautiously to avoid inadvertently hindering the availability of extremely helpful products.

2. Conclusion and future scope

The healthcare sector must remain active in the face of evolving global dynamics. By reengineering several PH system components, the research can effectively use genomics to accelerate the progress toward fully realizing the benefits of PM. The researchers are now facing a crucial decision: whether to follow the easier route, which would result in avoidable deaths, or choose a road of transformation that would decrease mortality and illness rates while enhancing the quality of life using big data. The research must enact significant modifications to expedite progress in this domain, as it has immense potential to influence individuals and their healthcare. This perspective represents the viewpoint influenced by the involvement of the most extensive PH sequencing initiatives inside the United States medical system. Some ideas outside of other medical facilities and other experts in this sector have different viewpoints. The goal is to promote a discussion that stimulates diverse viewpoints and promotes cooperation to expedite the use of genomes to improve the area of PM.

Reference

[1] Johnson, K. B., Wei, W. Q., Weeraratne, D., Frisse, M. E., Misulis, K., Rhee, K., ... & Snowdon, J. L. (2021). Precision medicine, AI, and the future of personalized health care. Clinical and translational science, 14(1), 86-93.

[2] Bobir, A.O., Askariy, M., Otabek, Y.Y., Nodir, R.K., Rakhima, A., Zukhra, Z.Y., Sherzod, A.A. (2024). Utilizing Deep Learning and the Internet of Things to Monitor the Health of Aquatic Ecosystems to Conserve Biodiversity. Natural and Engineering Sciences, 9(1), 72-83

[3] Naithani, N., Sinha, S., Misra, P., Vasudevan, B., & Sahu, R. (2021). Precision medicine: Concept and tools. medical journal armed forces india, 77(3), 249-257.

[4] Muralidharan, J. (2020). Design Of High Precision and Frequency Full Wave Rectifier. Journal of VLSI Circuits and Systems, 2(2), 15-17

[5] S. Neelima, Manoj Govindaraj, Dr.K. Subramani, Ahmed ALkhayyat, & Dr. Chippy Mohan. (2024). Factors Influencing Data Utilization and Performance of Health Management Information Systems: A Case Study. Indian Journal of Information Sources and Services, 14(2), 146–152. https://doi.org/10.51983/ijiss-2024.14.2.21

[6] Pan, X., Ojcius, D. M., Gao, T., Li, Z., Pan, C., & Pan, C. (2020). Lessons learned from the 2019-nCoV epidemic on prevention of future infectious diseases. Microbes and infection, 22(2), 86-91.

[7] Nussinov, R., Tsai, C. J., & Jang, H. (2022). How can same-gene mutations promote both cancer and developmental disorders? Science advances, 8(2), eabm2059.

[8] Alamer, L., Alqahtani, I. M., & Shadadi, E. (2023). Intelligent Health Risk and Disease Prediction Using Optimized Naive Bayes Classifier. Journal of Internet Services and Information Security, 13(1), 01-10.

[9] Sherman, R. M., & Salzberg, S. L. (2020). Pan-genomics in the human genome era. Nature Reviews Genetics, 21(4), 243-254.

[10] Pirmohamed, M. (2023). Pharmacogenomics: current status and future perspectives. Nature Reviews Genetics, 24(6), 350-362.

[11] Radmanović, S., Đorđević, A., & Nikolić, N. (2018). Humus Composition of Rendzina Soils in Different Environmental Conditions of Serbia. Archives for Technical Sciences, 2(19), 57–64.

[12] Rocklöv, J., & Dubrow, R. (2020). Climate change: an enduring challenge for vector-borne disease prevention and control. Nature Immunology, 21(5), 479-483.

[13] Mizumoto, K., & Chowell, G. (2020). Estimating risk for death from coronavirus disease, China, January–February 2020. Emerging infectious diseases, 26(6), 1251.

[14] Malathi, K., Shruthi, S.N., Madhumitha, N., Sreelakshmi, S., Sathya, U., & Sangeetha, P.M. (2024). Medical Data Integration and Interoperability through Remote Monitoring of Healthcare Devices. Journal of Wireless Mobile Networks, Ubiquitous Computing, and Dependable Applications (JoWUA), 15(2), 60-72. https://doi.org/10.58346/JOWUA.2024.I2.005

[15] Padmanabhan, S., & Dominiczak, A. F. (2021). Genomics of hypertension: the road to precision medicine. Nature Reviews Cardiology, 18(4), 235-250.

[16] Sutton, R. T., Pincock, D., Baumgart, D. C., Sadowski, D. C., Fedorak, R. N., & Kroeker, K. I. (2020). An overview of clinical decision support systems: benefits, risks, and strategies for success. NPJ digital medicine, 3(1), 17.