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## PRECISION MEDICINE – AN OVERVIEW

**BOLLA S<sup>1</sup>, DHOMAKONDA B<sup>2</sup> AND BOGGULA N<sup>2\*</sup>**

**1:** Department of Statistical Programming, Fortrea, USA

**2:** CMR College of Pharmacy, Kandlakoya, Hyderabad, Telangana, India

**3:** Omega College of Pharmacy, Edulabad, Ghatkesar, Hyderabad, Telangana, India

**\*Corresponding Author: Dr. Narender Boggula: E Mail: [drnarenderboggula@gmail.com](mailto:drnarenderboggula@gmail.com)**

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### ABSTRACT

Precision Medicine represents a new way of thinking about medicine. Given the ongoing drive in the healthcare field to better understand the patient experience and involve patients in their treatment, it is critical to provide patients with opportunities to learn about precision medicine. Precision medicine aims to improve healthcare quality by tailoring the treatment process to each patient's unique changing health state. Public health policy and decision-making must adapt to this new frontier of healthcare delivery in order to meet the overarching public health goals of reducing healthcare inequalities and enhancing population health through effective and fair healthcare funding allocation. Precision medicine seeks to distinguish illnesses using a variety of data sources, ranging from genomes to digital health indicators, in order to make individualised but "evidence-based" decisions on diagnostic and treatment techniques. Therapeutics can be tailored to patients depending on their molecular presentation, rather than categorising them with a "one size fits all" approach. To fully realise precision medicine's disruptive potential, a comprehensive scientific, clinical, and policy agenda will be required. This review article is aimed to provide a broad overview of the advent and emergence of precision medicine in view of its current implications.

**Keywords: Precision medicine, preventive medicine, health care**

## INTRODUCTION

Precision medicine makes use of genome sequencing to uncover biomarkers that can help patients understand the nature of their condition. Biomarkers can be prognostic, meaning they provide information about overall outcomes, or predictive, meaning they provide information about the probability of treatment response, which helps optimise therapeutic decisions. For example, precision medicine can assist oncologists in selecting the most effective treatment while avoiding inefficient medicines with negative side effects. In primary care, precision medicine could be used to inform decisions regarding treatments, such as smoking cessation by examining a patient's speed of nicotine metabolism, or inform decisions regarding drinking reduction for alcohol abusers by identifying patients who would respond well to topiramate, a drug that can be used to aid in alcohol abstinence [1, 2].

Precision medicine advancement and integration into ordinary clinical treatment require patient buy-in, as well as an understanding of the related risks and benefits. Multiple studies have found that people struggle to understand precision medicine. In one poll, individuals with advanced cancer were asked about their understanding and opinions regarding genetic testing. The findings indicate that patients were unable to distinguish between

germline sequencing (which analyses the patient's genetic code) and somatic sequencing (which examines the tumor's genetic alterations). Another poll confirmed this conclusion, suggesting that cancer patients considering precision medicine are prone to misinterpret the difference between somatic and germline sequencing [3, 4].

### **Precision and personalized medicine: A definition that implicitly reflects one of the two main models of health**

Throughout history, two primary models of health representation have emerged: the biomedical or biological model and the biopsychosocial model introduced in the 1970s. The biomedical model seeks to define health based on the individual's biology, with health itself being determined by various biological factors such as genetics, molecular composition, cellular structure, histology, and anatomy. This individual-centric representation is complemented by the consideration of environmental factors, encompassing everything outside of the individual. To encompass health in its entirety, a gene  $\times$  environment interaction model is used, providing a broad yet not explicitly defined perspective. Initially, this model is deeply reductionist and materialistic in nature. Precision medicine, as discussed earlier, is largely based on this model of health,

focusing on characterizing individuals through their biological attributes.

The biopsychosocial model considers individual health by incorporating the biological, psychological, and social dimensions. This approach aligns with the WHO's definition of health as a "state of complete physical, mental, and social wellbeing, not merely the absence of disease or infirmity" (World Health Organization, 2013). Unlike the pathology model, the biopsychosocial model does not solely focus on biological determinants of health, but also includes environmental, socio-economic, and psychological factors. By embracing a multidimensional perspective, this model presents a more holistic, detailed, and personalized view of health. In essence, the biopsychosocial model reflects a more personalized approach to medicine [5-8].

#### **Why it is important to develop a personalized medicine and health based on a broader vision of health**

There is substantial evidence indicating the impact of various external exposures on biological functioning at different omics levels in animals and humans, such as gene expression influenced by epigenetic changes. While physicochemical exposures and health-related behaviour have been extensively studied, the influence of the social environment is often overlooked, as recent literature on the exposome has revealed. Despite calls for their inclusion in

research, social environment, and psychosocial exposures have been shown to affect biological functioning, even at the omics level. This underscores the importance of integrating these factors to enhance our understanding of biological functioning, health, and disease. Horton has proposed the concept of a syndemic rather than a pandemic about the COVID-19 epidemic, emphasizing how considering both social and biological aspects can enhance our comprehension and management of the disease.

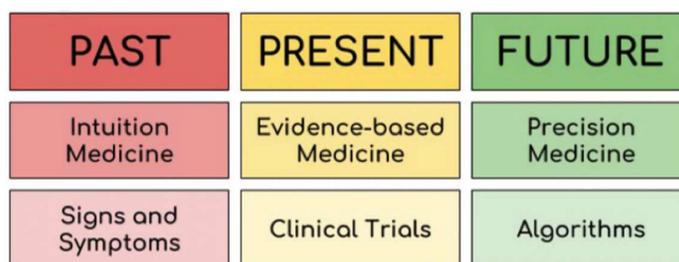
One of the main sources of information for patients is their provider; research shows that precision medicine is unfamiliar and that patients are unlikely to seek information on their own. Unfortunately, clinicians are ill-prepared to provide patients with the type of information that is needed to understand precision medicine, as clinicians with the genetic expertise to provide patients with a robust understanding of precision medicine are limited. With the increased frequency of precision medicine, the demand for these clinicians is greater than the current supply, meaning that there is an increased likelihood that clinicians with limited understanding of genetics concepts will be required to educate patients about them. Research demonstrates that patients are likely to misinterpret risk levels associated with genetic findings and that patients are likely to overestimate the

usefulness of precision medicine findings [9-14].

Due to the complexity of precision medicine, it is important to verify that patient-facing materials are effectively communicating the necessary information. As such, the objective of this review is to identify and report on studies that test the effectiveness of patient-facing precision medicine materials, including educational materials and precision medicine testing results. In response to growing interest from the healthcare community to better understand and improve patient experience, this research identifies components of precision medicine patient-facing materials that improve patient understanding of these complex concepts.

Standing at the peak of healthcare evolution, one might wonder if this increase in life expectancy will continue. Moreover,

within the current limits of the lifespan, we are already paying the price for living longer. Currently, we spend approximately 50% of the budget dedicated to healthcare on treating terminal illnesses such as cancer, heart failure, and other degenerative diseases. The single factor common to all of these diseases is aging. To better target the issues related to this increase in life expectancy, medicine in the 21st century must focus on attaining the 4 Ps stated by Dr. Leroy E. Hood: prediction, prevention, personalization, and participation. We must now transition from traditional reactive medicine based on symptoms, diagnosis, and treatment to a system that targets the disease before it occurs and, if it cannot be avoided, treats the disease in a personalized manner [15-18]. This evolution in medicine is summarized in **Figure 1**.



**Figure 1: The evolution of medicine. In the past, medicine was practiced according to the signs and symptoms presented by the patient and was solely based on the individual knowledge of the physician and thus was called intuition medicine. Currently, medicine is based on the evidence produced by scientific research, including clinical trials, which is designated as evidence-based medicine. In the future, medicine will be practiced according to algorithms that will take into consideration the patient's characteristics, such as their genome, epigenetics, and lifestyle, constituting precision medicine**

### Benefits [19, 20]

Precision medicine, also known as personalized medicine, is an innovative

approach to medical treatment and healthcare that takes into account individual differences in genetics, environment, and

lifestyle. The primary goal of precision medicine is to tailor medical care to each patient's unique characteristics, thereby improving the effectiveness, safety, and efficiency of healthcare. Here are some key benefits of precision medicine:

- **Improved Treatment Effectiveness:** Precision medicine allows healthcare providers to identify the most effective treatments for individual patients based on their genetic makeup, biomarkers, and other specific factors. This can lead to more successful outcomes and reduced trial-and-error in treatment selection.
- **Customized Therapies:** Precision medicine enables the development of targeted therapies that are designed to address the specific molecular mechanisms underlying a patient's disease. This can result in better treatment response and fewer side effects compared to traditional one-size-fits-all approaches.
- **Enhanced Patient Safety:** By tailoring treatment plans to individual patients, precision medicine can help reduce the risk of adverse reactions and side effects. Patients are less likely to be prescribed medications that may be ineffective or harmful based on their genetic predispositions.
- **Early Disease Detection and Prevention:** Precision medicine facilitates the identification of genetic and biomarker signatures associated with disease susceptibility. This enables early detection and intervention, potentially preventing diseases or detecting them at a more treatable stage.
- **Reduced Healthcare Costs:** While the initial investment in precision medicine technologies and research can be substantial, in the long run, the approach has the potential to lower healthcare costs. By avoiding ineffective treatments and adverse events, unnecessary hospitalizations and treatments can be minimized.
- **Accelerated Drug Development:** Precision medicine approaches can streamline the drug development process by identifying patient subgroups most likely to benefit from a new therapy. This can lead to faster drug approvals and a higher success rate in clinical trials.
- **Patient Empowerment and Engagement:** Precision medicine encourages patients to take an active role in their healthcare decisions. Patients may become more engaged in managing their health, making informed choices based on their genetic information and personal data.
- **Better Management of Chronic Diseases:** Precision medicine can

provide insights into the underlying causes of chronic diseases, helping healthcare providers develop targeted treatment plans that address the root causes and provide more effective disease management.

- **Personalized Risk Assessment:** By analyzing genetic and environmental factors, precision medicine can provide individuals with personalized risk assessments for various diseases. This information can guide lifestyle changes and interventions to reduce disease risk.
- **Advancements in Research and Knowledge:** The adoption of precision medicine generates vast amounts of data that can be used to advance scientific understanding of diseases and their genetic underpinnings. This knowledge can lead to the discovery of new therapeutic targets and approaches.

Overall, precision medicine has the potential to revolutionize healthcare by offering more tailored and effective treatments, improving patient outcomes, and contributing to a deeper understanding of human health and diseases.

### **Challenges [3, 21]**

While precision medicine offers numerous benefits, its implementation also comes with several challenges that need to be addressed.

These challenges include:

- **Data Privacy and Security:** Precision medicine relies heavily on the collection and analysis of patient's genetic and personal health information. Ensuring the privacy and security of this sensitive data is crucial to maintain patient trust and prevent potential breaches.
- **Data Quality and Standardization:** The accuracy and consistency of genetic and clinical data are essential for effective precision medicine. Variability in data quality and lack of standardized formats can hinder accurate analysis and interpretation.
- **Ethical and Social Considerations:** Precision medicine raises ethical questions about data ownership, consent, and potential uses of genetic information. It also highlights concerns about disparities in access to advanced treatments, as well as the potential for discrimination based on genetic traits.
- **Healthcare Disparities:** The benefits of precision medicine may not be equally accessible to all populations. Variations in healthcare infrastructure, socioeconomic factors, and geographic location can lead to disparities in access to advanced diagnostic tools and treatments.

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- **Complexity of Data Analysis:** Analyzing vast amounts of genetic and molecular data requires advanced computational and analytical methods. Integrating diverse datasets and extracting meaningful insights can be challenging, requiring expertise in bioinformatics and data science.
  - **Limited Biomarker Discovery:** Identifying relevant biomarkers for specific diseases can be complex and time-consuming. Many diseases have multifactorial causes, making it difficult to pinpoint accurate biomarkers for diagnosis, prognosis, and treatment selection.
  - **Regulatory and Legal Frameworks:** Regulatory agencies must adapt to the unique challenges posed by precision medicine, such as the approval process for targeted therapies and diagnostic tests. Navigating evolving regulatory landscapes can impact the pace of innovation and clinical implementation.
  - **Clinical Validation and Validation Studies:** Before precision medicine approaches can be widely adopted, rigorous clinical validation and validation studies are necessary to demonstrate the clinical utility and effectiveness of new tests and treatments.
  - **Integration with Healthcare Systems:** Integrating precision medicine into existing healthcare systems and workflows can be complex. Healthcare providers need tools and training to effectively interpret and use genetic information in patient care.
  - **Cost and Accessibility:** The development and implementation of precision medicine technologies can be expensive. The cost-effectiveness of these approaches needs to be carefully evaluated, and efforts must be made to ensure that they are accessible to a broad range of patients.
  - **Lack of Longitudinal Data:** Many precision medicine initiatives require long-term data collection and follow-up to understand the impact of interventions over time. Lack of comprehensive longitudinal data can hinder the assessment of treatment outcomes and disease progression.
  - **Patient Education and Consent:** Patients may not fully understand the implications of genetic testing and precision medicine interventions. Ensuring informed consent and providing clear education about
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potential risks and benefits are essential.

Addressing these challenges requires collaboration among researchers, healthcare professionals, policymakers, ethicists, and patient advocates. As precision medicine continues to advance, efforts to overcome these obstacles will contribute to its successful integration into routine clinical practice.

### **Strategy [2, 22]**

Developing and implementing a successful precision medicine strategy requires a comprehensive approach that encompasses various aspects of healthcare, research, technology, and patient engagement. Here is a strategic framework for precision medicine:

#### 1. Data Collection and Integration:

- Establish infrastructure for collecting diverse data types, including genomic, clinical, lifestyle, and environmental information.
- Develop standardized protocols for data collection, storage, and sharing to ensure data quality and interoperability.
- Implement electronic health records (EHRs) and other data systems that facilitate seamless data integration and access.

#### 2. Genomic Sequencing and Analysis:

- Utilize next-generation sequencing technologies to analyze the genetic makeup of individuals.
- Develop robust bioinformatics pipelines for accurate and efficient analysis of genomic data.
- Identify relevant biomarkers and genetic variants associated with diseases and treatment responses.

#### 3. Biomarker Discovery and Validation:

- Research to discover and validate biomarkers for diagnosis, prognosis, and treatment selection.
- Design clinical trials and validation studies to assess the clinical utility of identified biomarkers.
- Collaborate with academic institutions, industry partners, and regulatory agencies for validation efforts.

#### 4. Targeted Therapies and Treatment Development:

- Leverage insights from genomic and biomarker data to design targeted therapies.
- Partner with pharmaceutical companies and research organizations to develop

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- precision medicine drugs and interventions.
  - Implement clinical trials to test the efficacy and safety of targeted treatments in specific patient populations.
5. Clinical Decision Support:
- Integrate genomic and biomarker information into clinical workflows to support treatment decisions.
  - Develop decision support tools and algorithms that provide actionable insights to healthcare providers.
  - Train healthcare professionals to interpret and utilize genetic information effectively in patient care.
6. Patient Engagement and Education:
- Educate patients about the benefits, risks, and implications of precision medicine.
  - Provide clear and understandable information about genetic testing and its potential impact on treatment options.
  - Encourage patients to actively participate in their healthcare decisions and provide informed consent for genetic testing.
7. Ethical and Legal Considerations:
- Establish ethical guidelines and policies for the responsible use of genetic and personal health data.
  - Address issues related to data privacy, security, informed consent, and potential discrimination.
  - Collaborate with legal experts and policymakers to navigate regulatory and legal challenges.
8. Healthcare Integration and Infrastructure:
- Integrate precision medicine into existing healthcare systems and workflows.
  - Ensure interoperability between different healthcare technologies and data platforms.
  - Provide training and support for healthcare providers to effectively use precision medicine tools.
9. Research and Innovation:
- Foster collaboration between academia, industry, and government to drive research and innovation in precision medicine.
  - Support research initiatives focused on understanding disease mechanisms, genetics, and treatment responses.
  - Invest in the development of novel technologies and analytical
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methods to advance precision medicine.

## CONCLUSION

Healthcare systems are gradually shifting towards new care models that emphasize integrated care processes involving multiple providers and empower patients to take an active role in their own care. As precision medicine continues to gain momentum, there will be a growing need for patients to comprehend how it impacts their treatment. Precision medicine is expanding its reach from primary care to oncology, requiring a broader population of patients to engage with precision medicine findings. By reducing healthcare costs, drug development expenses, and time, precision medicine has the potential to enhance health outcomes. It can also have a positive impact on the overall healthcare system. In the future, personalized medicine will enable individuals to receive their complete genomic information from the day they are born, which will be stored in their individual medical records. This valuable data will enable physicians and clinicians to implement more effective healthcare strategies based on each patient's susceptibility to various diseases.

## Conflicts of interest

None.

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