

A Review on Integration of Precision Medicine in Oncology Practice

Harini Karimanal*, Dinesh Bandaru

PharmD Intern, Seven Hills College of Pharmacy, Tirupati, Andhra Pradesh, India

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Abstract: Oncology, the medical specialty focused on cancer study, diagnosis, and treatment, has experienced remarkable progress over the years. The introduction of chemotherapy, targeted therapy, and immunotherapy has broadened treatment possibilities, yet the field continues to face significant challenges. Precision medicine, also known as personalized medicine, has emerged as a transformative approach in this complex environment. It offers innovative strategies for disease prevention and treatment by considering individual genetic, environmental, and lifestyle differences. A leading trend in precision oncology is the formulation of personalized treatment plans based on the distinct genetic and molecular characteristics of each patient's tumor. The evolution of precision medicine in oncology has been driven by technological advancements such as genomic sequencing technologies, high-throughput screening, bioinformatics, targeted therapy, liquid biopsy, CRISPR and gene editing, and the application of artificial intelligence and machine learning. These innovations have significantly influenced cancer treatment, enabling more precise and effective interventions. This article examines the critical technological developments propelling precision medicine forward and assesses their impact on oncology. It also explores the essential steps required to integrate precision medicine into clinical practice, addressing the challenges and advantages associated with its adoption. Integrating precision medicine into oncology promises to enhance the accuracy of diagnoses, improve treatment outcomes, and reduce adverse effects by tailoring interventions to individual patient profiles. This review discusses the importance of continued research and technological innovation in realizing the full potential of precision oncology.

Keywords: Precision Medicine; Genomic sequencing; Targeted therapy; High Throughput screening; Liquid biopsy; Artificial Intelligence.

1. Introduction

Over the past few years, cancer treatment has been radically transformed by the discovery of numerous molecular alterations that fuel the development and progression of the disease. Personalized medicine is a growing field where doctors utilize diagnostic tests to pinpoint unique biological markers, frequently genetic, to ascertain the most effective treatments and procedures for individual patients [1]. Personalized oncology refers to evidence-based, individualized treatment strategies that provide the appropriate care to each cancer patient precisely when needed, leading to significant improvements in patient outcomes and a reduction in healthcare costs [2]. Precision medicine, also referred to as personalized medicine, is an innovative approach to disease prevention and treatment that takes into account individual variability in genes, environment, and lifestyle for each person. At the core of precision medicine is the utilization of advanced technologies [3].

In the realm of oncology, precision medicine has transitioned towards tailoring treatments to suit individual patients, taking into account not just their genetic makeup and that of their cancer, but also other distinguishing factors such as lifestyle, microbiome, and diet. Each patient presents a distinct combination of genomic, proteomic, epigenomic, and environmental influences, all of which play a role in cancer development, progression, and treatment response. With cancer encompassing numerous heterogeneous diseases, treatment efficacy varies not only between patients but also within the same individual. As cancer continually evolves mechanisms to resist treatment, a singular cure-all drug remains elusive. Precision oncology now hinges on leveraging the unique attributes of each patient to guide both immunotherapy and targeted treatment strategies [4]. This involves the use of molecular profiling techniques, such as genomic sequencing, to identify specific mutations or alterations in a patient's tumor [3].

One of the key benefits of precision medicine is its ability to improve patient outcomes by providing treatments that are better aligned with each individual's unique biological makeup. By selecting therapies based on a patient's genetic profile and other relevant factors, precision medicine can enhance treatment efficacy, reduce the likelihood of adverse reactions, and ultimately,

* Corresponding author: Harini Karimanal

improve survival rates for many diseases, including cancer, cardiovascular conditions, and rare genetic disorders [5]. Furthermore, precision medicine holds the potential to revolutionize healthcare by enabling early detection and intervention, thereby preventing diseases before they manifest or progress to more advanced stages. [5, 6] This main aim of this review is to study the critical technological developments propelling precision medicine forward and assesses their impact on oncology.

2. Technological advancements driving precision medicine

Precision medicine, a transformative approach to healthcare, tailors treatments to individual patient characteristics. Several key technological advances are driving this field forward, enabling more accurate diagnosis, treatment, and monitoring of diseases. Emerging technologies are swiftly increasing the range of tools available for precision medicine, enabling more accurate diagnostics and personalized treatment plans. These advancements also support the creation of innovative therapies, enhancing the ability to target diseases at the molecular level and improve patient outcomes [5]. There are many technological advances like genomic sequencing, Bioinformatics and Computational technology, Liquid biopsy, Telemedicine. [6, 7]

2.1. Genomic sequencing technologies

Genomic sequencing technologies have significantly advanced the field of oncology, enabling personalized approaches to cancer diagnosis, treatment, and management. Genomic sequencing technologies are revolutionizing precision oncology by providing detailed insights into the genetic underpinnings of cancer. By identifying specific genetic alterations, these technologies enable personalized treatment strategies tailored to the unique genetic profile of each patient's tumor [7]. As these technologies continue to advance, they hold the promise of improving cancer diagnosis, treatment, and patient outcomes. The primary genomic sequencing technologies driving precision medicine in oncology are:

2.1.1. Next-Generation Sequencing (NGS)

Next-Generation Sequencing (NGS) is a cornerstone technology in precision oncology. NGS allows for the rapid sequencing of large amounts of DNA, providing comprehensive genomic information. This technology can identify a wide range of genetic alterations, including single nucleotide variants (SNVs), insertions and deletions, copy number variations (CNVs), and structural variations. NGS is used in various applications, such as whole genome sequencing (WGS), whole exome sequencing (WES), and targeted gene panels, to detect mutations that drive cancer progression and influence treatment response. [8]

2.1.2. Whole Genome Sequencing (WGS)

Whole Genome Sequencing (WGS) involves sequencing the entire genome of a cancer cell. This comprehensive approach provides a complete picture of the genetic changes in cancer, including mutations in coding and non-coding regions. WGS can uncover rare genetic variants and complex structural changes that might be missed by other sequencing methods. It is particularly useful for identifying novel oncogenic mutations and understanding the genetic landscape of different cancer types. [8]

2.1.3. Whole Exome Sequencing (WES)

Whole Exome Sequencing (WES) focuses on sequencing the exome, the protein-coding regions of the genome, which represent about 1-2% of the entire genome but contain approximately 85% of known disease-related mutations. WES is a cost-effective alternative to WGS, providing detailed information about genetic mutations in the coding regions that are most likely to impact cancer development and treatment response. WES is widely used to identify actionable mutations that can be targeted with specific therapies. [8]

2.1.4. RNA Sequencing (RNA-seq)

RNA sequencing (RNA-seq) analyzes the transcriptome, the complete set of RNA transcripts produced by the genome. RNA-seq provides insights into gene expression levels, alternative splicing events, and fusion transcripts that are often involved in cancer. By comparing RNA-seq data from cancerous and normal tissues, researchers can identify dysregulated pathways and potential therapeutic targets. RNA-seq is particularly useful for understanding the functional impact of genetic alterations. [8, 9]

2.1.5. Single-Cell Sequencing

Single cell sequencing technologies allow for the analysis of the genomic and transcriptomic profiles of individual cancer cells. This high-resolution approach uncovers intra-tumor heterogeneity, providing insights into the diverse cellular populations within a tumor and their unique genetic profiles. Single-cell sequencing helps identify resistant subclones, track tumor evolution, and develop strategies to overcome therapeutic resistance. [8, 9]

2.2. Bioinformatics and Computational Biology

Bioinformatics and computational biology are crucial in bridging the gap between biological research and clinical applications, particularly in oncology. They serve as the interface between the clinical development of drug targets and the discovery and development of biomarkers. In oncology, computational biology leverages computational tools and machine learning for data mining to identify potential cancer therapies and understand tumor biology. Bioinformatics employs computing and mathematics to analyze biological datasets, supporting the solution of complex biological problems. Specifically, bioinformatics is essential in analyzing data from various 'omics' platforms, annotating and classifying genes and pathways for cancer target identification and disease association, thereby advancing personalized cancer treatment and improving patient outcomes [10].

2.3. Liquid Biopsy

Most cancer-related deaths result from tumor cells spreading through the bloodstream from the primary site to other parts of the body. Detecting and analyzing circulating tumor cells (CTCs) in blood samples from cancer patients has provided a new method to investigate the dissemination of tumor cells via the blood [2]. Clinically, liquid biopsies—such as circulating tumor cells (CTCs) and circulating tumor DNA (ctDNA) obtained from noninvasive blood samples throughout the disease—can be utilized for various purposes. These include: (i) the early detection of cancer, albeit screening remains challenging, often requiring larger blood volumes; (ii) tumor staging and patient monitoring in localized cancers to distinguish between those at low versus high risk of recurrence; (iii) predicting metastatic progression in advanced cancer patients; (iv) assessing therapy effectiveness and differentiating early responders from nonresponders; and (v) tracking tumor evolution by identifying therapeutic targets and resistance mechanisms. This non-invasive approach provides real-time insights into tumor dynamics, allowing for the early detection of resistance mutations and the adjustment of treatment strategies. Advances in digital PCR and NGS have enhanced the sensitivity and specificity of liquid biopsies, making them a valuable tool in precision oncology. [11]

2.4. CRISPR and Gene Editing

Precision oncology represents a transformative approach in cancer treatment, tailoring interventions to individual genetic profiles, and central to this paradigm is the revolutionary CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) technology. CRISPR is a versatile tool for gene editing that offers unprecedented precision in modifying the genome, fundamentally changing our understanding of cancer biology and therapeutic development. Adapted from a bacterial defense system, CRISPR uses the Cas9 enzyme guided by RNA to identify and cut specific DNA sequences, allowing precise modifications at the target site. Its ease of use and high efficiency make it indispensable in genetic research and therapeutic advancements. In oncology, CRISPR is pivotal for identifying cancer drivers through genome-wide screens, creating accurate cancer models, and elucidating gene functions. This technology also holds promise for directly treating cancers by correcting mutations in oncogenes or tumor suppressor genes, enhancing CAR-T cell therapy, and overcoming drug resistance by identifying and counteracting resistance mechanisms [12].

2.5. Telemedicine

Telemedicine is revolutionizing precision oncology by providing remote consultations, facilitating the management of genomic and molecular data, and enabling real-time patient monitoring through wearable devices. It supports multidisciplinary collaboration via Virtual tumor boards, supported by telemedicine bring together multidisciplinary teams to discuss complex cases, review genomic data, and develop personalized treatment plans. This collaborative approach enhances decision-making and ensures comprehensive care. Additionally, telemedicine expands access to clinical trials, integrates diverse data sources for advanced analytics, and provides platforms for patient education and psychological support. Despite regulatory and reimbursement challenges, telemedicine is poised to significantly enhance personalized cancer treatment, improve access to specialized care, and optimize patient outcomes in precision oncology [12-14]

3. Key trends in Precision oncology

Precision oncology is rapidly evolving, driven by advances in genomics, technology, and data analysis. As our understanding of cancer biology deepens, several key trends are shaping the field, offering new insights and opportunities for improving patient care. They are :

3.1. Immunotherapy

Immunotherapy has emerged as a transformative approach in cancer treatment, harnessing the body's immune system to target and destroy cancer cells. Key advancements include immune checkpoint inhibitors and chimeric antigen receptor (CAR) T-cell therapy. Precision oncology aims to identify predictive biomarkers, such as PD-L1 expression or tumor mutational burden, to select patients who are most likely to benefit from immunotherapy. [12-14]

3.1.1. Immune Checkpoint Inhibitors

Immune checkpoint inhibitors (ICIs) target proteins that regulate immune responses, such as programmed cell death protein 1 (PD-1) and cytotoxic T-lymphocyte-associated protein 4 (CTLA-4). By blocking these checkpoints, ICIs enhance the immune system's ability to recognize and attack cancer cells. Examples include pembrolizumab, nivolumab, and ipilimumab, which have demonstrated efficacy across multiple cancer types, including melanoma, lung cancer, and renal cell carcinoma. [12-14]

3.1.2. CAR T-cell Therapy

Chimeric antigen receptor (CAR) T-cell therapy involves genetically modifying a patient's T cells to express synthetic receptors targeting specific antigens on cancer cells. Upon infusion back into the patient, these engineered T cells can recognize and destroy cancer cells. CAR T-cell therapy has shown remarkable success in treating hematologic malignancies, such as B-cell acute lymphoblastic leukemia (ALL) and certain types of non-Hodgkin lymphoma. Examples include axicabtagene, ciltacabtagene, and tisagenlecleucel. [12-14]

3.1.3. Predictive Biomarkers

Identifying predictive biomarkers is essential for selecting patients who are most likely to benefit from immunotherapy. Biomarkers such as programmed death-ligand 1 (PD-L1) expression, tumor mutational burden (TMB), and microsatellite instability (MSI) status help stratify patients and guide treatment decisions. For example, PD-L1 expression predicts response to PD-1/PD-L1 inhibitors in various cancer types, including non-small cell lung cancer and bladder cancer. [12-14]

3.2. Targeted Therapy

Targeted therapy, a cornerstone of precision oncology, tailors treatment to the molecular features of a patient's tumor. By selectively inhibiting key molecules or pathways involved in cancer growth and progression, targeted therapies offer more precise and effective treatment options compared to traditional chemotherapy. Molecular profiling, facilitated by techniques like next-generation sequencing, identifies actionable genetic alterations driving cancer development. For instance, tyrosine kinase inhibitors (TKIs) such as gefitinib and erlotinib target mutated EGFR in non-small cell lung cancer, while vemurafenib inhibits the BRAF V600E mutation in melanoma. Precision medicine approaches, guided by biomarker testing like HER2 amplification status in breast cancer, ensure optimal patient selection for targeted therapies [11].

3.3 Artificial Intelligence and Machine Learning

In the wake of the recent big data revolution, the biomedical community is now able to apply artificial intelligence (AI) and machine learning algorithms to vast amounts of data thanks to the creation of continuously collected large data sets from various molecular profiling (genetic, genomic, proteomic, epigenomic, and others) of patient samples, wearable medical devices, mobile health applications, and clinical outcome data. The development of technology has opened up new avenues for study in the areas of virtual diagnosis, tailored therapy delivery, precision medicine, predictive diagnostics, and patient monitoring. Researchers in academia, business, and government agencies have all been interested in these developments, which are also giving doctors new resources [8].

4. Implementation of Precision Medicine in Oncology Practice

The implementation of precision medicine in clinical practice represents a transformative shift in the approach to cancer treatment. By tailoring therapies based on the individual genetic and molecular profiles of patients' tumors, precision medicine aims to enhance treatment efficacy and reduce adverse effects [9].

4.1. Steps for Implementation

4.1.1. Comprehensive Genomic Profiling

The foundation of precision medicine is the comprehensive genomic profiling of tumors. This involves using technologies like next-generation sequencing (NGS) to analyze the genetic mutations, alterations, and expression patterns within the tumor. Clinical laboratories and specialized genomic testing companies perform these analyses, providing detailed reports that identify actionable mutations and potential targets for therapy.

4.1.2. Multidisciplinary Tumor Boards

Multidisciplinary tumor boards, comprising oncologists, geneticists, pathologists, bioinformaticians, and other specialists, review genomic data and clinical information to formulate personalized treatment plans. These boards discuss complex cases, interpret genomic findings, and recommend targeted therapies or clinical trials suitable for the patient's unique tumor profile.

4.1.3. Integration of Electronic Health Records (EHR)

Integrating genomic data into electronic health records (EHR) systems ensures that this information is readily accessible to healthcare providers. EHR systems facilitate the documentation of genomic findings, treatment decisions, and patient outcomes, enabling a comprehensive view of the patient's medical history and personalized treatment plan [9].

4.1.4. Utilization of Biomarkers

Biomarkers are critical for selecting appropriate therapies, monitoring treatment response, and detecting recurrence. Implementing precision medicine involves regular biomarker testing using blood, tissue, or liquid biopsy samples. Companion diagnostics, which are tests specifically designed to identify patients likely to benefit from a particular therapy, are increasingly used in clinical practice.

4.1.5. Clinical Decision Support Systems (CDSS)

CDSS are digital tools that assist clinicians in interpreting complex genomic data and making evidence-based treatment decisions. These systems integrate genomic information with clinical guidelines and the latest research, providing recommendations tailored to individual patients' molecular profiles [9].

4.1.6. Patient Education and Consent

Educating patients about the potential benefits, risks, and limitations of precision medicine is essential for informed consent. Oncologists and genetic counselors work together to explain the significance of genomic testing, the implications of the results, and the personalized treatment options available.

5. Challenges in implementation of precision medicine

The key challenges [15, 16] to overcome are:

5.1.1. Cost and Accessibility

The high cost of genomic testing and targeted therapies can be a barrier to widespread adoption. Ensuring that these advanced treatments are accessible to all patients, regardless of their financial status, is a significant challenge. Insurance coverage and reimbursement policies for genomic tests and personalized treatments vary, impacting patients' access to precision medicine.

5.1.2. Standardization and Quality Control

There is a need for standardized protocols and quality control measures to ensure the accuracy and reliability of genomic testing across different laboratories. Variability in testing methodologies and interpretation of results can lead to inconsistencies in patient care.

5.1.3. Data Management and Privacy

The integration of large volumes of genomic data into clinical practice requires robust data management systems and safeguards to protect patient privacy. Ensuring compliance with regulations such as the Health Insurance Portability and Accountability Act (HIPAA) is crucial for maintaining the confidentiality and security of patient information.

5.1.4. Educational and Training Needs

Continuous education and training of healthcare providers are essential to keep pace with the rapid advancements in genomic technologies and precision medicine. Clinicians need to be proficient in interpreting genomic data, understanding its clinical implications, and effectively communicating this information to patients.

The strategies to overcome implementing the precision in healthcare system are represented in Figure 1.

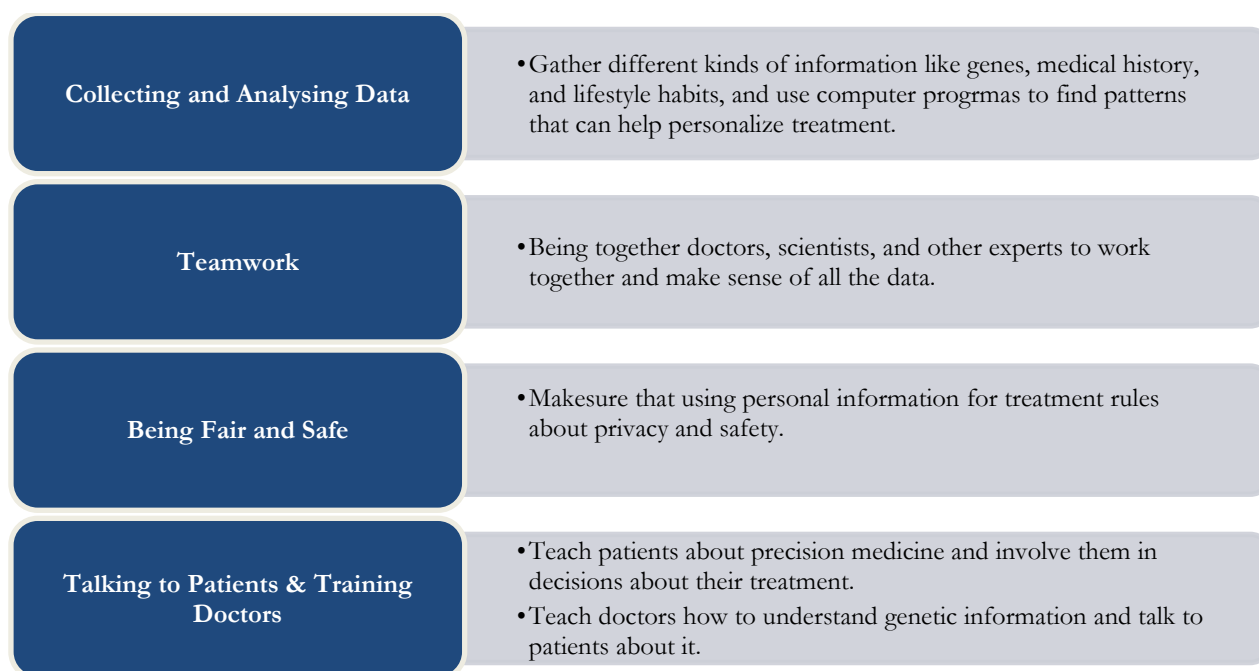


Figure 1. Strategies to overcome implementing the precision in healthcare system

6. Conclusion

Precision medicine has the potential to revolutionize oncology by providing highly individualized treatment plans based on the genetic and molecular characteristics of each patient's tumor. The technological advancements driving this field—genomic sequencing, high-throughput screening, bioinformatics, AI, liquid biopsy, and gene editing—are transforming how cancer is diagnosed, monitored, and treated. These innovations promise to improve treatment outcomes, reduce adverse effects, and enhance the quality of life for cancer patients. However, the integration of precision medicine into clinical practice is not without challenges. As the healthcare system transitions from a traditional, one-size-fits-all approach to a personalized medicine paradigm, it will be necessary to address several critical issues. Strategies involving educational programs, awareness campaigns, and initiatives to empower patients can be implemented now or in the near term to facilitate this shift. Ensuring access to advanced technologies, addressing ethical and legal concerns, and managing tumor heterogeneity are also critical issues that need to be addressed. Continued investment in research and development, along with policy initiatives to support the equitable distribution of these technologies, will be essential for the widespread adoption of precision medicine in oncology. In conclusion, while significant hurdles remain, the ongoing advancements in precision medicine are poised to make a substantial impact on the future of cancer treatment.

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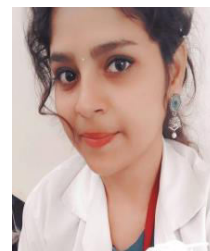
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Author's short biography

Harini Karimanal

Studying Pharm D in Seven Hills College of Pharmacy, Tirupati, Andhrapradesh, India and currently doing internship at Sri Venkateswara Institute of Medical Sciences (SVIMS), Tirupati.



Dinesh Bandaru

Studying Pharm D in Seven Hills College of Pharmacy, Tirupati, Andhrapradesh, India and currently doing internship at Sri Venkateswara Institute of Medical Sciences (SVIMS), Tirupati.

