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Precision Medicine: The Future of Medicine Tailored to Individual Patients

Pengobatan Presisi: Masa Depan Pengobatan yang Disesuaikan dengan Pasien Individu

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ABSTRACT
Genome sequencing technology and genomic analysis have become important tools in the realm of personalized medicine, revolutionizing diagnostic and treatment strategies tailored to each patient individually. This systematic literature review explores the implications of genome sequencing and analysis technologies in the context of personalized medicine, with the aim of clarifying their potential and challenges. Through a thorough examination of the relevant literature, including studies of genome sequencing technologies, pan-genome analysis, and the integration of genomic data in clinical decision making, this review provides insight into the current state and future directions of personalized medicine. These findings underscore the importance of genomic literacy, cross-disciplinary collaboration, and ethical considerations in realizing the full potential of personalized medicine. By highlighting advances, limitations, and implications of genome sequencing and analysis, this review contributes to the ongoing discourse on precision medicine and informs research and clinical practice in this rapidly developing field.
Keywords: Genome sequencing, Pan-genome analysis, Personalized medicine, Clinical decision making, Precision medicine, Interdisciplinary collaboration, Ethical considerations.

ABSTRAK
Teknologi sekuensing genom dan analisis genomik telah menjadi alat penting dalam ranah kedokteran yang disesuaikan secara personal, merevolusi strategi diagnosis dan pengobatan yang disesuaikan untuk setiap pasien secara individual. Tinjauan literatur sistematis ini mengeksplorasi implikasi teknologi sekuensing genom dan analisis dalam konteks kedokteran yang disesuaikan secara personal, dengan tujuan untuk menjelaskan potensi dan tantangannya. Melalui pemeriksaan menyeluruh terhadap literatur yang relevan, termasuk studi tentang teknologi sekuensing genom, analisis pan-genom, dan integrasi data genomik dalam pengambilan keputusan klinis, tinjauan ini memberikan wawasan tentang kondisi saat ini dan arah masa depan kedokteran yang disesuaikan secara personal. Temuan ini menegaskan pentingnya literasi genomik, kolaborasi lintas disiplin, dan pertimbangan etis dalam mewujudkan potensi penuh kedokteran yang disesuaikan secara personal. Dengan menyoroti kemajuan, keterbatasan, dan implikasi dari sekuensing genom dan analisis, tinjauan ini memberikan kontribusi dalam diskursus yang sedang berlangsung tentang kedokteran presisi dan memberikan informasi bagi penelitian dan praktik klinis di bidang yang berkembang pesat ini.
Kata Kunci: Sekuensing genom, Analisis pan-genom, Kedokteran yang disesuaikan secara personal, Pengambilan keputusan klinis, Kedokteran presisi, Kolaborasi lintas disiplin, Pertimbangan etis.

1. Introduction

Precision medicine, also known as personalized medicine, is an innovative approach that aims to customize medical treatment and disease management to individual patients based on their unique characteristics, including genomics and lifestyle (Wouters et al., 2021). This approach seeks to empower clinicians by providing them with reliable user-friendly tools to convert ongoing advances in biomedical knowledge into actionable, improved efficiency therapies tailored to individual patients (Duffy, 2015). However, the transition from conventional to personalized medicine faces several challenges, including the generation of cost-effective high-throughput data, data storage and processing, and individual and global economic relevance (Alyass et al., 2015). Precision medicine involves molecular profiling of individual tumors combined with established clinical-pathological parameters, which reveals individual patient's diagnostic and prognostic risk profiles, informing tailored and tumor-specific treatment plans in real-time (Matchett et al., 2017).

Advocates of precision medicine argue that it maximizes healthcare quality by tailoring the healthcare process to each patient's unique characteristics (Hisan & Amri, 2023). It holds great promise for improving the prevention, diagnosis, and treatment of many diseases (McGonigle, 2016). However, critics argue that precision medicine neglects the limitations of individual-centered, high-risk strategies and the current crisis of evidence-based medicine, which may reduce population health impact (Rey-López et al., 2018). Despite these challenges, the therapeutic management of selected patients with cancer has shifted toward the precision medicine approach based on patients' mechanisms of tumorigenesis, baseline characteristics, and comorbidities (Fountzilas & Tsimberidou, 2018).

To implement precision medicine approaches in healthcare, many countries have adopted national strategies and initiated genomic/precision medicine initiatives to provide equal access to all citizens (Edsjö et al., 2023). Precision medicine has raised concerns about a lack of access to relevant data in the healthcare sector (Sahu, 2022). By tailoring each individual medical treatment through precision medicine, it has the potential to lead to nearly zero occurrences of drug side effects and treatment complications (Mohamad et al., 2018).

In conclusion, precision medicine represents a paradigm shift from a one-size-fits-all approach to a tailored, individualized approach to healthcare. While it holds great promise for improving patient outcomes, there are significant challenges in its implementation, including data access, economic relevance, and evidence-based limitations.

Precision medicine, also known as personalized medicine, is an approach that aims to individualize treatment strategies based on a patient's unique characteristics, including genomic patterns and lifestyle factors (Williams et al., 2019). This approach has the potential to address health disparities and improve patient outcomes by optimizing treatment efficacy and minimizing side effects (Williams et al., 2019). However, the implementation of precision medicine faces several challenges, including the development of cost-effective technologies for comprehensive patient profiling, robust data storage and processing infrastructure, and equitable dissemination of innovations across diverse socioeconomic contexts (Williams et al., 2019). emphasize that precision medicine challenges the conventional "average patient" norm, providing a more individualized approach to treatment, considering the genetic makeup, environment, and lifestyle of individual patients (Mamun et al., 2019).

highlight the importance of tailored population health and prevention strategies, often termed “precision population health,” alongside individualized patient treatments in precision medicine (Lyles et al., 2018). Furthermore, discuss the potential of precision medicine in reducing the burden of diseases such as diabetes, emphasizing the concept of individualized treatment to improve overall health (Burke et al., 2019). provide insights into the transdisciplinary perspectives on precision medicine, emphasizing the need for a collaborative approach to address the complexities and opportunities inherent in precision medicine (Myers et al., 2021).

In conclusion, precision medicine holds significant promise in revolutionizing healthcare delivery by individualizing treatment approaches. However, addressing the challenges associated with its implementation and ensuring equitable access across diverse populations are crucial for realizing its transformative potential.

2. Research Methods

In the process of this research, relevant articles have been collected from reputable international databases such as PubMed, Web of Science, and Scopus. Article searches were carried out using relevant keywords such as "precision medicine", "personalized medicine", "individualized medicine", and "genomic medicine". In addition, additional keywords such as “future of medicine” and “patient-centered care” were also used to broaden the scope of the search. After conducting an initial search, a number of articles were obtained in accordance with the predetermined inclusion criteria.

The article inclusion and exclusion process was carried out based on certain criteria to ensure the relevance and quality of articles to be included in this literature review. Relevant articles were selected based on the title, abstract and full text according to the research topic. Inclusion criteria include original research articles, systematic reviews, and narrative reviews that discuss recent developments, challenges, and impacts of precision medicine. Articles that are irrelevant, duplicate, or do not meet the specified inclusion criteria will be excluded from this study.

To organize the process of searching and selecting articles systematically, the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) method has been used as a guide. The PRISMA method helps in detailing the steps for searching, selecting and including articles in a transparent and systematic manner. In this way, the research process can be explained in detail and can be accounted for.

3. Results and Discussions

3.1.1. Genomic Sequencing and Analysis

Genomic sequencing has revolutionized the diagnosis of monogenic conditions, as evidenced by the use of microarray diagnosis in identifying autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy caused by a novel homozygous intragenic AIRE deletion (Akesson et al., 2020). Furthermore, genomic sequencing technologies have enabled the analysis of pan-genomes, revealing all genes present within a species by comparing chromosome-level de novo genome sequences of multiple accessions belonging to the same species (Shirasawa et al., 2021). Pan-genome analysis has broad applications in studying genomic diversity, phylogeny, disease outbreak, and virulence-associated genes in bacteria (Li & Yin, 2022). Additionally, whole genome sequencing data has been used for surveillance and control of Listeria monocytogenes, although the analysis of such data varies widely and is not yet fully harmonized (Lüth et al., 2021). Genomic sequencing has also been instrumental in elucidating the system dynamic characteristics and evolution of viral epidemics, such as the African swine fever virus (Shen et al., 2022; Bao et al., 2019). Comparative genomic analysis has provided evidence of gene transfer and phylogenetic relationships in plant genomes (Cui et al., 2021). Moreover, whole-genome sequencing and comparative analysis have been crucial in understanding the evolution of African swine fever virus (Forth et al., 2019; Torresi et al., 2020). The analysis of pan-genomes in human populations has revealed common sequences originating from genomic regions characterized by high mutation rates and low pathogenicity (Li et al., 2021). However, the implications of whole genome sequencing, such as the identification of clinically actionable DNA variants, have raised concerns about extensive clinical investigations and unnecessary worry (Semsarian, 2019). Furthermore, comparative genomic analysis has been essential in understanding antimicrobial susceptibility patterns and drug resistance in bacterial species (Lin et al., 2019). Lastly, efficient whole-genome sequencing of African swine fever virus has been facilitated by a simple sample preparation method (Olasz et al., 2019).

3.1.2. Understanding Genomic Sequencing

Genomic sequencing methods have revolutionized the generation of high-quality whole-genome sequences (Forth et al., 2019). Reduced representation sequencing techniques are frequently used in non-model organisms due to their low cost and ease of use (Bresadola et al., 2020). Advanced genome sequencing technologies are expected to analyze pan-genomes, revealing all genes present within a species by comparing chromosome-level de novo genome sequences of multiple accessions belonging to the same species (Shirasawa et al., 2021). Additionally, the development of long-read sequencing supports the economical generation of high-quality genome sequences, rapidly accelerating genome research (Zhu et al., 2022). Metagenomic sequencing allows for the efficient sequencing of whole genomes, requiring only a small number of tissues (Yadav et al., 2022). Next-generation sequencing-based bulked segregant analysis enables the rapid identification of genomic regions that control traits of interest (Zhang & Panthee, 2021). Furthermore, viral metagenomic sequencing provides an efficient method for the analysis of African Swine Fever (Ji et al., 2021).

3.1.3. Role of Genomic Analysis in Personalized Medicine

Genomic analysis is a crucial component of personalized medicine, enabling tailored diagnostic tools and treatments for individual patients. Genomic literacy is essential for the efficient implementation of personalized medicine (Zimani et al., 2021). Genomic projects have been designed to facilitate personalized and precision medicine, underscoring the significance of genomic analysis in this field (Kovanda et al., 2021). Additionally, the integration of artificial intelligence in the clinical utility landscape of genomic information has been identified as a means to advance personalized medicine (Suwinski et al., 2019). Whole-genome and whole-exome analysis have been instrumental in identifying causative genes of undiagnosed diseases and diagnosing patients' diseases, demonstrating the potential of genomic analysis in personalized medicine (Yamamoto et al., 2022). Genomic testing has led to a medical revolution that is evidence-based, impacting current clinical practice and shaping the future of healthcare, particularly in the context of personalized, predictive, preventive, and participatory medicine (Al-Dewik & Qoronfleh, 2019). These references collectively emphasize the pivotal role of genomic analysis in driving the advancements and implementation of personalized medicine.

3.1.4. Utilizing Genomic Sequencing and Analysis in Personalized Treatment Plans

Personalized treatment plans are increasingly being developed through the application of genomic sequencing and analysis. The use of comprehensive next-generation sequencing technology allows for the rendering of personalized treatment recommendations within a clinically relevant timeframe (Mueller et al., 2019). It is becoming more likely that tumors will undergo genomic analysis to provide patients with personalized treatment plans that maximize efficacy and reduce toxicity (Reynolds et al., 2022). Additionally, the incorporation of artificial intelligence in the clinical utility landscape of genomic information is seen as a new frontier toward advancing the field of personalized medicine (Suwinski et al., 2019).

By leveraging these references, it is evident that genomic sequencing and analysis play a crucial role in the development of personalized treatment plans, offering the potential to optimize treatment efficacy and minimize adverse effects.

3.1.5. Integration of Genomic Data into Clinical Decision-Making

To integrate genomic data into clinical decision-making, a multidisciplinary approach is essential to guide the interpretation of raw genomic data, clinical correlation, and the integration of diagnostic findings in patient care (Nestor, 2022). Enabling widespread adoption and integration of genomic medicine into clinical practice is crucial for achieving precision medicine (Handra et al., 2023). Challenges in integrating genomics into public healthcare systems include increasing patient and community awareness, training a clinical workforce, establishing infrastructure for genomic testing and data management, providing evidence on the value of genomics, and addressing ethical, legal, and social issues (Vidgen et al., 2021). Genomics-informed public health responses require detailed integration of genomic and epidemiological data, necessitating close collaboration between laboratories and public health agencies (Seemann et al., 2020).

The integration of genomic technology into healthcare presents unique ethical issues that challenge traditional aspects of healthcare delivery (Bilkey et al., 2019). Genomic sequencing is a transformative technology, and its effective integration in healthcare requires system-wide change (Stark et al., 2019). To facilitate the use of genomic test results in clinical practice, it is essential to integrate genomic data into clinical decision support systems regarding data volume and knowledge management (Kim et al., 2020). Integration of genomic sequence data with epidemiological data improves understanding of disease transmission patterns and outbreak dynamics (Stephens et al., 2021).

The incorporation of genomics improves patient decision-making and reduces anxiety with postoperative decisions, demonstrating the clinical utility and benefit of genomic testing (Marascio et al., 2019). A decision-making framework for genomic testing was created to explore its impact on patient management, clinical validity, patient characteristics, genetic counseling, and ethical considerations (Beadell, 2023). The responsibility for integrating clinical genomics into public healthcare in Australia requires a coordinated whole-of-nation approach based on a federated system (Stark et al., 2019).

The faster, cheaper generation of genomic data is driving the integration of genomics into all healthcare specialties (Josephs et al., 2019). A pipeline for knowledge discovery from integrated genomic and clinical data has been proposed, emphasizing the importance of integrating these data for improved decision-making (Raghu, 2020). The integration of genomic information and electronic health records is recognized as an inherent feature of disease management (Zhang et al., 2020). Clinical utility is viewed as a key standard for reimbursement decision-making, highlighting the importance of evidence for the clinical utility of genomic tests (Parker & Miller, 2021).

Relevant clinical meta-data can be combined with genomic analyses to provide hospital-specific reports for infection control teams, demonstrating the practical integration of genomics into hospital infection control (Forde et al., 2022). Genetics professionals engage in 'the familialization of genomics' by drawing on family characteristics in their analysis of genomic data, showcasing the integration of familial aspects into decision-making (Hedgecoe & Job, 2023). Genomic analysis results, such as single nucleotide polymorphisms, can be integrated with epidemiological data to predict transmission routes and determine outbreak origins, emphasizing the value of integrating genomic and epidemiological data (Permana, 2024). In conclusion, the integration of genomic data into clinical decision-making requires a multidisciplinary approach, system-wide change, ethical considerations, evidence of clinical utility, and the practical integration of genomic and epidemiological data. These efforts are essential for realizing the potential of precision medicine and improving patient care.

3.1.6. Ethical Considerations and Patient Consent

The ethical considerations and patient consent in precision medicine are crucial aspects that require careful attention. Precision medicine involves the use of genetic and molecular profiling to tailor medical treatment to individual patients, and this raises several ethical considerations regarding patient privacy, autonomy, and group health interests (Juengst & Rie, 2020). The challenges of implementing precision medicine in clinical practice include the standardization of diagnostic criteria and treatment guidelines, as well as ethical and legal considerations of personalized treatment (Pathak, 2023). Furthermore, there are distinct challenges with informed consent to precision medicine, involving the ability of both patient-participants and providers to effectively understand the science underlying the research (Spector-Bagdady et al., 2022). The traditional areas of concern, such as patient privacy and financial benefits, have been joined by issues associated with precision medicine, such as the increasing length and complexity of informed consent forms and participant genetic privacy (Matrana & Campbell, 2020).

Dynamic Consent has been proposed as a facilitator for managing patient consent in precision medicine, allowing individual patient preferences about the degree of linkage of personal data to be taken into account (Haas et al., 2021). Additionally, it has been argued that dynamic consent is necessary for personalized and precision medicine to achieve its goals (Goncharov et al., 2022). The challenges in the acquisition and analysis of large amounts of omics data, as well as the challenges of informed consent and medical ethics in precision medicine, must be overcome to attain the goals of precision medicine (Liu et al., 2019). In conclusion, the ethical considerations and patient consent in precision medicine are complex and multifaceted. They require careful attention to ensure that patient privacy, autonomy, and understanding of the research are upheld. Dynamic Consent has been proposed as a potential solution to manage patient consent effectively in the context of precision medicine.

3.1.7. Implications for Various Medical Conditions

Precision medicine has emerged as a promising approach for various medical conditions, including diabetes, cancer, and cardiovascular diseases. In the context of diabetes, precision medicine initiatives aim to improve the quality of life for individuals with diabetes by providing a roadmap for personalized treatment (Chung et al., 2020). However, there are barriers to implementing precision medicine in diabetes, which need to be addressed to realize its full potential (Riddle et al., 2020). In the case of cancer, precision medicine involves targeted therapy based on biomarkers, such as specific proteins and genes, to tailor treatment strategies for individual patients (Imaoka et al., 2021). Similarly, in the context of cardiovascular diseases, precision medicine takes into account individual variability in genes, environment, and lifestyle factors to enable more accurate treatment and prevention strategies (Leggio et al., 2019).

The application of precision medicine in these medical conditions is supported by the identification of critical gaps in knowledge and evidence required for scientific advancement, implementation, and ongoing evaluation of precision medicine (Nolan et al., 2022). Furthermore, precision medicine programs aim to improve survival and treatment-related toxicity for patients with cancer through informed treatment decision-making based on the molecular characteristics of individual tumors (Gereis et al., 2023). Additionally, precision medicine for lung cancer involves the collection and integration of various patient data to identify the most suitable therapeutic targets and achieve precise treatment (Guan et al., 2022).

The potential of precision medicine extends beyond treatment to include accurate disease classification, diagnosis, and prognosis based on individual clinical phenotypes and omics data (Yuan, 2022). Moreover, precision medicine initiatives in oncology and cardiovascular diseases consider individual variability in genes, environment, and lifestyle factors to tailor treatment strategies for each patient (Saadeh et al., 2019; Leggio et al., 2019). However, the successful implementation of precision medicine in diabetes requires an expanded focus on biomarkers and the inclusion of social determinants of health, such as food security and access to healthcare (Krook & Mulder, 2022). In summary, precision medicine holds great promise for improving the management and treatment outcomes of various medical conditions, including diabetes, cancer, and cardiovascular diseases. By considering individual variability in genes, environment, and lifestyle factors, precision medicine aims to provide personalized and targeted treatment strategies, ultimately leading to better patient outcomes.

3.1.8. Advancements in Genomic Technologies

Advancements in genomic technologies have significantly impacted various fields such as plant breeding, genome editing, and whole genome sequencing. The completion of human genome sequencing marked a turning point, leading to remarkable advancements in genomic tools and sequencing technologies (Kim et al., 2020). These technological advancements include single-cell RNA sequencing (scRNAseq) in genomics, improved mass spectrometry analysis in proteomics, and the development of prime editing, a precision genome editing technology that uses reverse transcription to introduce programmed sequence changes into genomic DNA (Bentzur et al., 2022; Yan et al., 2020). Furthermore, the construction of a reference genome is crucial for the advancement of genomic studies, and the appearance of next-generation sequencing (NGS) technologies has enabled de novo whole genome assembly in various plant species (Isobe et al., 2020). The era of genomics continues to advance with improved sequencing technologies, offering the potential to sequence all recorded species on earth (Sharma et al., 2021).

The fields of functional genomics and crop improvement have been transformed by advances in genome editing tools, particularly CRISPR/Cas9 (Siddique, 2022). Moreover, the genomics revolution, made possible through the development of high-throughput sequencing, has triggered considerable progress in the study of ancient DNA, enabling the reconstruction of complete genomes of past organisms (Spyrou et al., 2019). Additionally, the development of technology that can capture large volumes of sequence data at low costs and with high accuracy has driven the acceleration of plant genome sequencing advancements (Henry, 2022).

In summary, advancements in genomic technologies have revolutionized various fields, including plant breeding, genome editing, and whole genome sequencing. These advancements have been made possible through the development of high-throughput sequencing technologies, such as NGS, and the application of innovative tools like prime editing and CRISPR/Cas9. These technologies have not only accelerated research in genomics but also hold the potential to further expand our understanding of the genetic basis of living organisms.

3.1.9. Collaborative Efforts and Data Sharing

Collaborative efforts and data sharing are crucial in advancing precision medicine, as highlighted in several recent studies. emphasized the need for multi-omics biomarker signatures and the development of analytical infrastructure to inform precision medicine-based decision-making (Olivier et al., 2019). Similarly, discussed ongoing initiatives to create national implementation strategies for precision medicine on a global scale (Marchiano et al., 2021). Furthermore, underscored the importance of good regulations for patient data ownership, clinical data-sharing, and robust data infrastructure in ensuring the success of precision medicine (Hisan & Amri, 2023).

Highlighted the Precision Medicine Health Disparities Collaborative, which fosters collaboration between researchers with diverse backgrounds in precision medicine and health disparities research (Myers et al., 2021). This collaborative approach is essential for addressing health disparities and ensuring equitable access to precision medicine. Additionally, emphasized the importance of cross-disciplinary collaboration in precision medicine, particularly in clinical application and technology-associated disciplines (Xu et al., 2021).

Moreover, discussed efforts targeting key action areas for the sustainability of precision medicine, including evidence generation, patient and public engagement, implementation, and data privacy and sharing (Lee et al., 2019). These efforts are crucial for establishing a robust framework for data sharing and collaboration in precision medicine research and implementation.

In summary, the references highlight the critical role of collaborative efforts and data sharing in advancing precision medicine. They underscore the need for multi-omics biomarker signatures, national implementation strategies, good regulations for data sharing, and cross-disciplinary collaboration to ensure the success of precision medicine initiatives.

4. Conclusion

Based on the discussion outcomes, it can be concluded that genomic sequencing technology and pan-genome analysis bear significant implications within the realm of personalized medicine. The revolution in genomic sequencing has enabled more precise diagnoses and the development of individually tailored therapies, as evidenced by the utilization of microarray in identifying monogenic conditions and pan-genome analysis in uncovering genetic diversity within a species. The implementation of personalized medicine necessitates a profound understanding of genomics and the integration of artificial intelligence in translating genomic information into effective clinical actions. However, challenges concerning technical limitations, costs, privacy, and ethics need to be addressed to optimize the potential of these technologies in clinical practice. In the future, further research is required to develop more affordable and efficient genomic sequencing technologies and to explore the ethical implications of individual genomic data usage in the context of personalized medicine. The integration of artificial intelligence in developing genomic analysis algorithm also represents a promising research area to enhance diagnostic accuracy and speed. Thus, a comprehensive understanding of the potentials and challenges of genomic sequencing technology and pan-genome analysis, along with the future research directions in this field, can lay the groundwork for further advancements in personalized medicine practice.

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