Applications of machine learning and deep learning in precision medicine: Opportunities and challenges in genomics, oncology and clinical integration (Review)

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Abstract. With the advancement of precision medicine, machine learning (ML) and deep learning have increasingly become a pivotal tool for driving medical innovation. Precision medicine, grounded in individual variability, aims to deliver personalized treatment interventions, with ML serving as a critical enabler for achieving this goal. Recent ML-driven progress in genomic analysis, personalized treatment optimization and disease diagnostics have significantly elevated the accuracy and efficacy of medical decision-making processes. However, the widespread adoption of artificial intelligence also faces multifaceted challenges, including data privacy frameworks, cybersecurity risks, ethical considerations and the integration of technology with clinical workflows. The present review seeks to analyze cutting-edge applications of ML within precision medicine domains, examine its challenges, and project future evolutionary pathways, emphasizing the critical need for proactive attention to these issues to ensure tangible benefits for patients and healthcare systems.

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1. Introduction

Precision medicine embodies a paradigm shift in healthcare delivery, centered on the development of personalized treatment strategies that integrate individual genomic profiles, environmental exposures and lifestyle factors. This paradigm shift signifies an evolution from traditional 'one-size-fits-all' protocols to more patient-specific and precise medical interventions. The exponential growth of 'omics' technologies, particularly genomics and metabolomics, highlights the substantial promise of precision medicine across disease prevention, diagnostic frameworks and therapeutic intervention strategies (1).

Machine learning (ML) and algorithms facilitate high-throughput analysis of biological and clinical datasets. Within this framework, the rapid advancement of ML technologies demonstrates strong capabilities for data processing in precision medicine contexts. ML systems can efficiently handle complex biological and medical data, thereby enabling clinicians to gain comprehensive insights into individual patient characteristics and develop more targeted treatment protocols. For instance, in oncology, ML algorithms predict tumor progression risk and therapeutic response by analyzing genomic profiles, supporting personalized treatment planning (2). Furthermore, ML contributes to accelerating drug discovery processes and optimizing clinical trial design, thereby enhancing both operational efficiency and therapeutic effectiveness in medical practice (3).

However, despite the significant potential of ML in precision medicine, multiple challenges remain. Data security remains a paramount concern, requiring robust safeguards to ensure the confidentiality and integrity of medical data. Additionally, ethical dilemmas are emerging as prominent concerns, particularly regarding patient trust in ML-driven decision-making processes and transparency in data usage (4). Furthermore, clinical integration of ML remains impeded by diverse technical barriers, underscoring the necessity to validate ML efficacy and reliability (5). To offer a clearer overview of real-world applications and constraints of ML in precision medicine, the present review introduces a

comparative table. The table systematically compares three core domains-genomics, medical imaging and personalized treatment-along dimensions of data types, key ML methodologies, representative applications and major challenges (Table I).

In summary, while the integration of ML within precision medicine offers transformative potential, it is accompanied by multifaceted challenges. The current narrative review was executed through a systematic literature review framework. PubMed (https://pubmed.ncbi.nlm.nih.gov/), Web of Science (https://clarivate.com/academia-government/scientific-and-academic-research/research-discovery-and-referencing/web-of-science/) and IEEE Xplore (https://ieeexplore.ieee.org/Xplore/home.jsp) were systematically queried for English-language articles published between 2015 and 2025. Search terms encompassed 'artificial intelligence (AI)', 'ML', 'precision medicine', 'genomics', 'deep learning', 'clinical integration' and 'data privacy.' Article selection prioritized relevance to ML applications in precision medicine, with emphasis on studies focusing on algorithm development, medical ethics and data interoperability. Editorials, opinion pieces and redundant publications were excluded. The present study aims to deliver a comprehensive review and critical analysis of the current landscape, opportunities and barriers associated with ML in precision medicine, thereby providing a foundational resource for future research and clinical implementation.

2. ML and precision medicine

Utilization of AI in genomic data analysis

Genomic data mining. Genomic data mining constitutes a critical component of contemporary medical research, particularly within the domain of large-scale data analysis leveraging ML. Advancements in high-throughput sequencing technology have led to an exponential increase in genomic data volume, surpassing the analytical capacity of traditional methods. ML technologies, notably ML, excel in processing and interpreting these complex genomic datasets, yielding critical insights. For instance, deep learning algorithms identify genomic patterns and variations potentially associated with specific diseases (6). Additionally, ML integrates heterogeneous literature-derived data through text mining approaches, enabling researchers to identify candidate biomarkers and therapeutic targets (7). This capacity to process and analyze data not only enhances research efficiency but also accelerates precision medicine progress, expanding genomic applications.

Association of genetic variations with diseases. Elucidating the relationship between genetic diversity and disease susceptibility represents a cornerstone of genomic research. The integration of ML has significantly enhanced both the depth and efficiency of analytical capabilities in this field. Leveraging ML algorithms, researchers can now analyze large-scale genomic datasets to identify genetic variations associated with specific diseases. For instance, ML enables the detection of distinct gene mutations linked to tumor mutation burden, which play pivotal roles in complex disease progression (8). Furthermore, ML demonstrates the capacity to integrate multi-omics data, including genomic, transcriptomic and metabolomic information, thereby facilitating a more comprehensive understanding of disease mechanisms (9). This multi-layered analytical framework not only deepens insights into the genetic basis of diseases but also provides actionable insights for early diagnosis and personalized treatment strategies within clinical settings.

ML and precision diagnosis. ML exhibits remarkable capabilities within precision diagnostics, particularly for complex diseases such as cancer and genetic disorders. ML enables researchers to rapidly and accurately assess disease risks through comprehensive analysis of patients' genomic data. For example, ML reliably identifies ovarian cancer by analyzing epigenomic modifications in circulating free DNA, thereby supporting the development of personalized treatment strategies for individuals (10). Furthermore, ML integration into image analysis enables innovative methodologies in pathology, exemplified by automated analysis of histopathological images, which significantly enhances diagnostic precision for conditions such as gastric cancer (11). Collectively, ML not only augments diagnostic efficiency but also facilitates improved prognostic outcomes and therapeutic responses for patients, marking a transformative era in medical diagnostics.

ML in personalized treatment

Models for predicting drug response. The integration of AI in predicting patient responses to pharmacological interventions is garnering increasing attention. Leveraging ML methodologies, researchers can develop sophisticated predictive models capable of analyzing patients' genomic data, clinical features and historical medication responses. These models play a pivotal role in identifying key factors influencing drug efficacy, thereby enabling healthcare providers to develop personalized treatment plans. For instance, multiple studies have shown that multilayer cell line drug response network models can reliably predict therapeutic responses in breast cancer patients (12). Additionally, predictive models for acute myeloid leukemia drug responses demonstrate significant promise, offering critical insights to inform clinical decision-making (13). By implementing these models, clinicians can gain deeper insights into interpatient variability, ultimately enhancing pharmacological treatment effectiveness. Within precision medicine, ML functions through four sequential processes: Patient data acquisition, ML-driven analysis, personalized treatment implementation and outcome feedback, collectively forming a closed-loop system (Fig. 1).

Precision medicine for oncology. Personalized cancer treatment, synonymous with precision medicine, represents a paradigm-shifting strategy that tailors therapeutic interventions to the unique characteristics of each patient. This approach integrates multidimensional factors-including genetic predisposition, environmental exposures and lifestyle determinants-that collectively influence cancer development and progression. By focusing on these individualized parameters, healthcare providers can identify optimal treatment modalities for specific patients (14). Through analysis of tumor-specific biomarkers and genetic mutations, clinicians can develop tailored therapeutic regimens that not only enhance therapeutic efficacy but also mitigate the risk of adverse effects (15). This patient-centric paradigm not only improves clinical outcomes but also fosters greater patient engagement and adherence to therapeutic regimens. Recent

Table I. ML applications in addressing precision medicine challenges.

Field	Data type	Main ML methods	Main challenges
Genomics	Genomic sequences, gene expression profiles and other multi-omics numbers	ML, deep learning	The problem of imbalance between high-dimensional data and samples makes it difficult to integrate cross-omicsdata. Genetic data is highly sensitive, and there are challenges in data sharing and privacy protection
Medical imaging	Medical imaging and pathological tissue sections, ultrasound images	Deep learning, traditional image processing and radiomics analysis methods	The demand for large-scale labeled data is high, and different devices and imaging protocols bring challenges to model generalization. Challenges in integrating regulatory approval and clinical processes
Precision medicine	Electronic health records (medical records), clinical phenotypes, lifestyle data, multi-omics and drug response data	ML, deep learning, reinforcement learning, network models	The fusion and standardization of heterogeneous data are highly challenging. The explainability of ML results is insufficient. Ethical compliance (such as obtaining consent) and clinical adoption barriers

ML, machine learning.

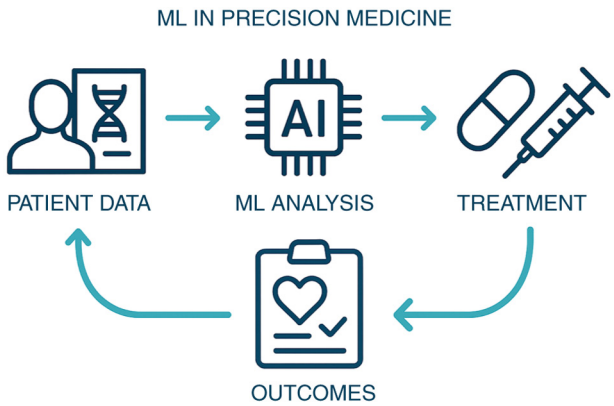


Figure 1. Four sequential processes of generative ML in precision medicine are illustrated: Patient data acquisition, ML-driven analysis, personalized treatment implementation and outcome feedback, forming a closed-loop optimization system. ML, machine learning.

advancements in genomic sequencing and bioinformatics have significantly enhanced the feasibility of implementing personalized treatment protocols. Consequently, oncologists are now better equipped to make evidence-based decisions grounded in a comprehensive understanding of each patient's tumor molecular profile. Ultimately, precision medicine holds substantial promise for improving survival rates and quality of life, marking a transformative shift toward individualized and molecularly targeted therapeutic options in oncology.

Optimizing treatment outcomes and dynamic monitoring. ML plays a critical role in optimizing treatment outcomes and enabling dynamic monitoring. By integrating real-time data analytics with patient-reported feedback, ML demonstrates the capacity to iteratively adjust therapeutic protocols in response to changes in patient conditions. For instance, in diabetes management, the integration of continuous glucose monitoring with ML-driven algorithms allows clinicians to dynamically optimize medication dosing, thereby improving patients' quality of life (16-18). Furthermore, in psychiatry, ML enhances treatment adherence and efficacy through continuous oversight of pharmacotherapy regimens (19). As ML technologies continue to advance, an expanding array of ML-based tools is expected to emerge, enabling healthcare providers to refine treatment strategies and monitor patient health more effectively, ultimately delivering more precise and personalized medical care.

Promise of ML in disease prediction and early diagnosis

Application of advanced image recognition technology in disease screening. The integration of advanced image recognition technology within disease screening is advancing rapidly, particularly in medical imaging analysis. Leveraging deep learning and ML algorithms, ML demonstrates proficiency in extracting complex features from medical images, thereby improving diagnostic accuracy. For example, in ophthalmology, ML has been applied to screen for diabetic retinopathy, with studies indicating that ML systems achieve diagnostic precision comparable to experienced ophthalmologists and, in certain cases, exceed the performance of specialists (20). Furthermore, ML exhibits significant potential in detecting hepatic pathologies, accurately identifying early-stage conditions such as non-alcoholic fatty liver disease through analysis of ultrasonography and computed tomography scans (21). This advanced image recognition technology not only enhances screening efficiency but also diminishes reliance on manual interpretation, thereby facilitating broader access to healthcare services.

Collaboration of biomarkers and ML models. The integration of biomarker identification with ML models has paved the way for novel approaches in early disease diagnosis. By leveraging multi-omics data-including genomics

and proteomics-ML demonstrates proficiency in detecting biomarkers linked to specific disease states (22). For instance, in early-stage cancer screening, ML can evaluate individual cancer risk and develop tailored surveillance protocols through analysis of multiple serum biomarkers. Additionally, ML identifies potential biomarkers by analyzing large-scale datasets, providing critical insights for novel drug development and disease prediction (23). This synergy not only improves the accuracy of biomarker discovery but also establishes a robust foundation for advancing precision medicine.

Enhancing early intervention and prognostic assessment capabilities. ML demonstrates significant potential in enhancing early intervention and prognostic assessment capabilities. Through comprehensive analysis of patient data, ML can identify individuals at elevated risk and develop tailored intervention strategies. For instance, in the clinical management of cardiovascular diseases, ML predicts the probability of cardiac events by analyzing patients' historical data and physiological parameters, enabling timely interventions (24). Furthermore, ML supports prognostic evaluations; by integrating clinical data with imaging results, it provides personalized prognostic assessments, enabling healthcare professionals to develop more effective treatment plans (25). This progress not only enhances patient outcomes but also reduces medical resource waste, thereby enhancing the overall efficiency of healthcare services.

Challenges of ML in the security and privacy of medical data
Data encryption and access control. In the domain of medical data security, the implementation of data encryption and access control is recognized as a critical technical strategy. The rapid advancement of ML has driven transformative changes in the storage and transmission of medical data, thereby posing new challenges to traditional security practices. Data encryption serves as a formidable barrier to prevent unauthorized access and ensure patient confidentiality. For instance, the application of homomorphic encryption enables computations to be performed without necessitating data decryption, thereby effectively safeguarding data privacy (26). Homomorphic encryption is a cryptographic technique that enables data to be analyzed in its encrypted form, facilitating secure analysis without exposing raw patient information. Moreover, the Attribute-Based Access Control model has gained significant traction in the administration of medical big data sharing, enabling flexible governance of access permissions based on user-specific attributes, thereby strengthening data security and adaptability (27). However, despite the potential these technologies offer for protecting medical information, challenges such as implementation complexities must be mitigated. Thus, healthcare organizations are encouraged to perform thorough evaluations when choosing the most suitable technologies.

Regulations of data privacy and ethical compliance. Adherence to data privacy regulations and ethical compliance frameworks is paramount in the deployment of ML technologies. With the enactment of legislation such as the General Data Protection Regulation (GDPR), medical institutions are mandated to ensure that their data processing practices align with legal standards to protect patient privacy rights (28). Furthermore, ethical compliance transcends mere legal obligations, encompassing the imperative to respect and safeguard patient autonomy through informed consent. When leveraging ML for patient data processing, healthcare providers must guarantee transparent communication regarding data utilization purposes and obtain explicit patient consent (29). However, practical implementation reveals significant challenges for healthcare organizations, largely attributable to the complexity and ambiguity inherent in legal frameworks. A critical tension also persists between advancing technological innovation and preserving patient confidentiality (30).

Significance of patient informed consent. Patient informed consent constitutes a cornerstone of ethical practice in healthcare, particularly regarding ML applications. Patients must be granted the autonomy to understand how their personal health information will be utilized, including associated risks and benefits (31). In the context of ML implementation, informed consent transcends mere legal compliance-it serves as a critical mechanism for fostering patient trust and safeguarding rights (32). As ML systems increase in complexity, patients may struggle to comprehend underlying algorithms and data processing methodologies. Healthcare organizations are therefore advised to implement transparent communication strategies that effectively convey technical details in accessible language (33). Ensuring informed patient consent and adhering to data privacy regulations such as GDPR remain essential. Dynamic consent platforms enable patients to control data-sharing preferences; while addressing ML biases requires vigilance-imbalanced training datasets may perpetuate healthcare disparities. Homomorphic encryption and federated learning represent promising solutions for enabling secure, decentralized data analysis.

Limitations of ML applications. Algorithmic bias has emerged as a critical ethical challenge in the application of ML within healthcare. When ML models are trained on datasets lacking demographic diversity or featuring imbalanced representations, they may produce biased outputs, resulting in disparate healthcare outcomes across diverse patient populations. For instance, empirical studies indicate that predictive models frequently exhibit suboptimal performance for racial minority groups or patients with rare diseases, attributable to inadequate representation in training datasets. Such disparities raise significant ethical concerns, as they risk perpetuating existing inequities in care access and diagnostic precision. To address this challenge, developers are advised to implement fairness audits, integrate diverse datasets, and employ bias-detection frameworks throughout the model development and validation lifecycle.

Challenges in technology integration and implementation
Data compatibility among diverse systems. In the modern healthcare ecosystem, ensuring data compatibility across heterogeneous systems is critical for the provision of high-quality medical care. Currently, the healthcare sector faces a multiplicity of data formats and standards, creating significant barriers to information exchange between institutions. A persistent challenge in healthcare has been the standardization of electronic health record systems, with the lack of interoperable data standards impeding the exchange and utilization of critical clinical information (34). To address this challenge, researchers have proposed various strategies, including the adoption of structural and semantic

interoperability frameworks to improve communication among healthcare information systems (33). Furthermore, given the increasing demand for cross-border medical data compatibility, the development of health data warehouse systems has emerged as a pivotal focus of research. This initiative not only enhances the efficiency of data sharing but also facilitates cross-border collaboration in healthcare initiatives (36). Nevertheless, despite continuous technological advancements, concerns regarding data privacy and security remain critical barriers to implementation. The lack of adherence to standardization protocols can give rise to data breaches and potential misuse (37). Consequently, advancing data compatibility across healthcare systems requires collaborative efforts from policymakers, technology developers and healthcare providers. Adopting international standards such as Fast Healthcare Interoperability Resources (FHIR) facilitates data interoperability by defining standardized resource definitions and interfaces. FHIR structures patient, diagnosis and medication information into standardized resources, thereby enabling data exchange through JSON/XML formats and RESTful APIs. This eliminates system barriers, allowing applications across devices or platforms to access unified clinical data.

Recognition of ML by the healthcare insurance system. The integration of ML within the healthcare sector is progressing at a rapid pace; however, the current healthcare insurance system's recognition of these innovations poses a significant barrier to their widespread adoption. Presently, healthcare insurance frameworks often exhibit delays in adapting to the evolving demands of emerging technologies, thereby hindering the implementation of ML applications (38). For instance, many insurance providers maintain skepticism regarding the clinical efficacy and reliability of ML-driven diagnostic tools, creating significant financial burdens for healthcare organizations seeking to deploy ML solutions (39). To enable the scalable adoption of ML technologies, it is imperative to redesign current healthcare insurance frameworks and establish rigorous evaluation systems capable of comprehensively assessing the clinical value and cost-effectiveness of ML interventions (40). Furthermore, regulatory bodies should facilitate collaboration between healthcare providers and technology developers by launching pilot programs to generate empirical evidence of ML's effectiveness, thereby providing a foundation for necessary insurance system reforms (41). Only through enhancing the adaptability of healthcare insurance frameworks can the full potential of ML in medical practice be realized.

Training of medical personnel and acceptance of ML. The professional development of healthcare workers and their readiness to adopt technological innovations are critical factors influencing the success of technological integration. While the adoption of emerging technologies holds potential to enhance healthcare efficiency and quality, limited comprehension and trust among clinical staff regarding these tools frequently lead to implementation resistance (42). Consequently, training programs for healthcare professionals must extend beyond technical proficiency, emphasizing foundational principles to build confidence and foster acceptance of novel technologies (43). For instance, some institutions have implemented simulation-based training and virtual reality platforms to facilitate comprehensive understanding of technological applications, demonstrating significant improvements in staff technological readiness (44). Additionally, organizational leadership should provide enhanced support mechanisms to encourage healthcare professionals to contribute insights on technological implementations, thereby promoting engagement and ownership (45). Only through structured training and open communication can the effective adoption of innovative technologies in clinical practice be guaranteed.

3. Discussion

The integration of ML into precision medicine presents transformative opportunities while simultaneously posing substantial challenges. Across genomics, oncology and clinical integration, ML-driven approaches have demonstrated the capacity to accelerate biomarker discovery, enhance diagnostic accuracy, and enable dynamic, individualized treatment strategies. For instance, in genomics, deep learning models have improved the identification of pathogenic variants and disease-associated patterns beyond the capacity of traditional bioinformatics pipelines. Similarly, in oncology, predictive modeling has facilitated patient-specific drug response forecasting, thereby optimizing therapeutic regimens. However, despite these advances, several systemic limitations remain evident. The reliance on heterogeneous and often imbalanced training datasets heightens the risk of algorithmic bias, potentially exacerbating disparities in diagnostic precision and therapeutic outcomes among underrepresented populations. Furthermore, the limited interoperability of healthcare data infrastructure, coupled with the reluctance of insurance frameworks to reimburse ML-driven interventions, constrains large-scale clinical adoption. Ethical concerns, including patient informed consent and the transparency of decision-support algorithms, also demand continuous oversight. Addressing these limitations requires a multi-stakeholder framework involving clinicians, policymakers and developers to standardize data protocols, mandate bias audits, and ensure rigorous clinical validation. Such coordinated efforts are essential for translating ML-enabled precision medicine from a conceptual paradigm into a scalable, equitable component of clinical care.

4. Conclusion

The integration of ML into precision medicine offers transformative potential, optimizing genomic analysis workflows, enabling personalized treatment planning, and enhancing disease prediction accuracy. These technological advancements not only improve therapeutic outcomes but also provide patients with more tailored healthcare solutions. However, the rapid advancement of ML necessitates addressing several critical challenges.

Primarily, challenges related to data privacy and security have become increasingly prominent. The effectiveness of ML systems relies heavily on large-scale patient datasets; thus, protecting sensitive information from misuse or unauthorized access has emerged as an urgent priority. Additionally, the complexities of technology integration demand careful consideration, as interoperability between heterogeneous ML systems and legacy healthcare infrastructure remains a significant barrier to seamless implementation. Achieving interoperability between diverse ML platforms and their

effective integration into legacy healthcare systems is essential for realizing the full potential of precision medicine. Furthermore, ethical dimensions require rigorous scrutiny, particularly concerning patient informed consent and transparency in algorithmic decision-making processes.

Looking ahead, conducting comprehensive assessments of both the capabilities and risks associated with ML applications in precision medicine is imperative. Balancing technological innovation with ethical practices necessitates multistakeholder collaboration among research institutions, healthcare providers and regulatory authorities. Standardizing data formats, ensuring algorithmic fairness, and engaging clinicians and patients in co-design processes will maximize ML's clinical impact. Through rigorous governance frameworks, ML can realize its potential to deliver personalized, equitable and efficient care. To accelerate clinical translation and equitable adoption, clinicians should strengthen ML literacy through training programs, policymakers should develop unified data standards and insurance reforms to facilitate ML integration, and developers must ensure transparency, fairness and multi-center validation of their models. Coordinated implementation of these measures will be essential for transforming ML from an emerging innovation into a sustainable, equitable and indispensable component of precision medicine.

In summary, while the integration of ML within precision medicine holds significant promise, addressing the introduced multifaceted challenges remains imperative. A multidimensional framework encompassing technological, ethical and security dimensions is essential to ensure the sustainable evolution of precision medicine, ultimately delivering equitable benefits to diverse patient populations.

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Authors' contributions

QZ, GL and KD conceptualized the study and wrote the manuscript. All authors read and approved the final version of the manuscript. Data authentication is not applicable.

Ethics approval and consent to participate

Not applicable.

Patient consent for publication

Not applicable.

Competing interests

The authors declare that they have no competing interests.

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