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Precision Medicine Using Artificial Intelligence and Genomics: A Comprehensive Review

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Abstract - Precision medicine is an emerging healthcare paradigm that tailors prevention, diagnosis, and treatment to individual patients using genetic, clinical, environmental, and lifestyle information. Advances in next-generation sequencing, bioinformatics, and Artificial Intelligence (AI) now allow clinicians to analyse genomic variants, stratify disease risk, and select therapies at a level of individual detail that population-averaged medicine cannot provide. This paper reviews AI-enabled precision medicine, synthesising landmark and recent peer-reviewed literature published between 2004 and 2026, and tracing the field from early blood-group stratification through the Human Genome Project to national precision medicine programmes and the multimodal AI of the present day. A conceptual framework is proposed that integrates electronic health records, genomic sequencing, laboratory tests, and wearable-device data in a secure platform where AI models identify biomarkers, predict risk, and recommend treatment for clinician validation; it is synthesised from literature-reported practices rather than experimentally validated. Applications in oncology, diabetes, cardiovascular disease, rare genetic disorders, pharmacogenomics, and remote monitoring are examined, followed by the main challenges, including data privacy, algorithmic bias, unrepresentative genomic datasets, cost, interpretability, and ethics, together with mitigation strategies. Emerging directions, including digital twins, federated learning, multimodal AI, and CRISPR-assisted therapies, are discussed. This review highlights the role of AI and genomics in moving healthcare from reactive to predictive and personalised, and identifies priorities for future research.

Keywords: Precision Medicine, Artificial Intelligence, Machine Learning, Genomics, Bioinformatics, Diabetes, Cancer, Digital Twins, Personalized Healthcare

1. INTRODUCTION

Conventional medicine has largely followed a one-size-fits-all model, in which treatments are chosen for the average patient with a given diagnosis. Precision medicine departs from this by customising prevention, diagnosis, and treatment using genomic, clinical, environmental, and lifestyle information [1], [2]. The shift has been driven by the falling cost of DNA sequencing, large cohort and biobank programmes, and the maturing of AI algorithms able to find patterns in these data [2], [3]. Within this landscape, AI has become the main tool for turning high-volume molecular and clinical data into decisions a clinician can act on.

Traditional care still relies on reactive, population-level guidance: a drug is prescribed because it works on average, and its failure or side effect is discovered after the fact. Precision medicine reverses this logic. By linking a patient's genome and clinical profile to evidence on how similar patients respond, clinicians can anticipate which therapy is likely to work before treatment begins. Inherited variation in about 20 genes affecting about 80 medications is already actionable in the clinic [4], and specific EGFR mutations in lung tumours identify patients likely to respond to gefitinib [5].

A growing body of literature has examined this field from many angles: policy initiatives, genomic technologies, machine learning methods, and disease-specific applications. Many studies focus on a single disease or a single technology layer, and few connect the genomic, AI, and clinical-workflow perspectives in one structured account for engineering students and early-career researchers. This paper addresses that gap by bringing together landmark studies and recent reviews through a conceptual framework, comparison tables, and disease-wise analysis, with emphasis on cancer and diabetes. As a narrative review, it does not follow a formal systematic-review protocol.

1.1 Evolution of Precision Medicine

The idea of matching treatment to patient subgroups is not new: blood typing has been used for more than a century to stratify patients [1]. The modern field took shape after the Human Genome Project was completed in 2003 [6], and the steep fall in sequencing cost brought by next-generation technologies made it feasible to sequence individual patients [2]. In January 2015 the United States announced a Precision Medicine Initiative with a near-term focus on cancers and a longer-term aim of generating knowledge applicable to all of health and disease [1]. Its cohort programme, later named All of Us, opened for enrolment in May 2018, and by July 2019 more than 175,000 participants had contributed biospecimens, over 80% of them from groups historically underrepresented in biomedical research [7].

From about 2018, deep learning entered clinical genomics and pathology, for example in variant calling [8] and in predicting tumour molecular status from routine histology [9]. Since roughly 2020, federated learning [10], multimodal AI [11], and digital twin concepts [12], [13] have extended the field towards privacy-preserving, data-integrated, and continuously updated models of individual patients. Figure 1 summarises this timeline.

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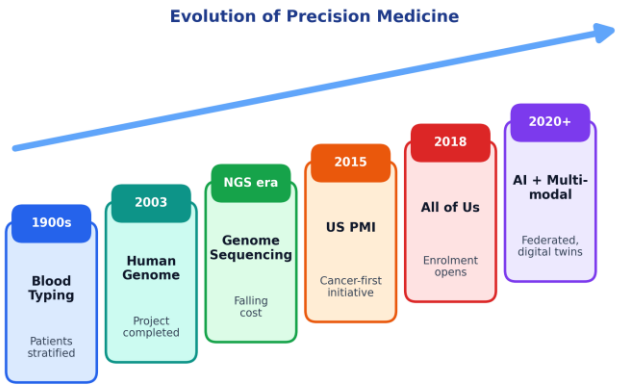

Figure 1. Evolution of Precision Medicine

1.2 Core Components

Regardless of disease, AI-enabled precision medicine implementations decompose into recurring building blocks. On the data side, patient information is gathered from electronic health records (EHRs), laboratory tests, imaging, genomic sequencing, and wearable devices and biosensors. Together with lower-cost genome and microbiome sequencing, these sources have set the stage for multimodal AI that captures the complexity of health and disease [11].

Bioinformatics pipelines then align sequence reads, call variants, and annotate them; deep convolutional networks such as DeepVariant have been shown to outperform existing state-of-the-art variant callers [8]. The resulting variant, biomarker, and clinical features feed machine learning models that predict disease risk, response to therapy, and prognosis [14], [3]. A decision-support layer presents recommendations to the clinician, who validates them before any change in care, and outcomes are fed back to improve the models. Chung et al. describe six categories of precision diabetes medicine, namely diagnosis, therapeutics, prevention, treatment, prognosis, and monitoring [15], which map naturally onto this loop. Figure 2 illustrates a representative architecture, and Table 1 summarises how several publications have described precision medicine.

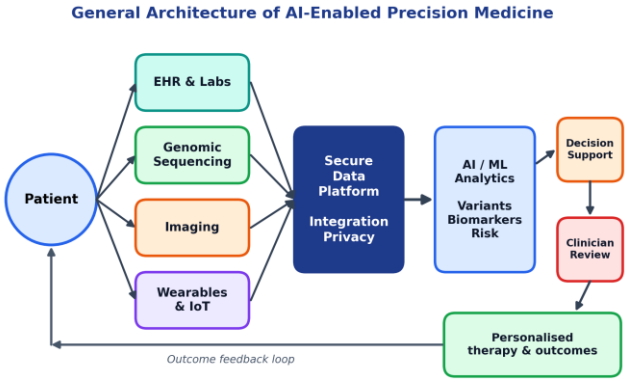

Figure 2. General Architecture of AI-Enabled Precision Medicine

Table 1. Comparison of Precision Medicine Definitions

Author	Year	Definition / Perspective	Main Contribution
Collins and Varmus [1]	2015	A new era of medicine pursued through a national research initiative, starting with cancer and extending to all of health and disease.	Framed the US Precision Medicine Initiative and its cancer-first agenda.
Ashley [2]	2016	Understanding disease at a deeper level so that it can be targeted more precisely.	Surveyed current applications such as precision oncology and argued the field must also become more accurate.
Topol [3]	2019	Convergence of human and artificial intelligence, with deep learning as the enabling method.	Described AI's impact on clinicians, health systems, and patients, and its limits in bias, privacy, and transparency.
Chung et al. [15]	2020	An approach that integrates multidimensional data to optimise diagnosis, prediction, prevention, or treatment of diabetes while accounting for individual differences.	Consensus framework with six categories: diagnosis, therapeutics, prevention, treatment, prognosis, monitoring.
Acosta et al. [11]	2022	Multimodal AI that combines biobank, EHR, imaging, biosensor, and sequencing data to model health and disease.	Outlined applications (personalised medicine, digital trials, remote monitoring, digital twins) and open challenges.

2. RELATED WORK

A considerable number of reviews and empirical studies have examined AI and genomics in precision medicine. Collins and Varmus [1] set out the case for the US Precision Medicine Initiative, beginning with cancer, and Ashley [2] reviews how falling sequencing costs and genome-driven decision making moved the field towards clinical use. Topol [3] and Rajkomar et al. [14] survey AI and machine learning in medicine, covering diagnosis, prognosis, treatment, and the need for high-quality data. Acosta et al. [11] review multimodal AI that merges biobank, EHR, imaging, biosensor, and sequencing data. Zeng et al. [16] present a narrative review of AI across precision medicine, drawing on literature searched up to April 2025, and highlight an uneven evidence base, limited model generalisability, and ethical concerns. Khan et al. [6] focus on integrating genomic, imaging, and EHR data, with implications for oncology, neurology, and cardiovascular medicine.

At the level of individual contributions, Poplin et al. [8] show that a deep convolutional network can call genetic variants from sequencing data and outperform existing state-of-the-art tools. Kather et al. [9] predict microsatellite instability, which influences response to immunotherapy, directly from routine histology slides in gastrointestinal cancer. Khera et al. [17] develop genome-wide polygenic scores that identify 8.0% of the population as having more than threefold risk of coronary artery disease, a group about 20 times larger than the carriers of rare monogenic mutations conferring comparable risk.

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Zeevi et al. [18] combine continuous glucose monitoring in 800 people with clinical, dietary, and gut microbiota data to predict personal post-meal glucose responses, and then lower them with a dietary intervention. Clark et al. [19] pair rapid whole-genome sequencing with automated phenotyping to diagnose genetic disease in seriously ill children, with a median of about 20 hours from sequencing to interpretation.

Equity and bias are recurring themes. Popejoy and Fullerton [20] show that genomic research is heavily skewed towards European-ancestry populations, leaving others behind on the road to precision medicine. Obermeyer et al. [21] find that a widely used commercial risk algorithm, which predicted health costs rather than illness, assigned the same risk score to Black patients who were considerably sicker than White patients; correcting this would raise the share of Black patients flagged for extra care from 17.7% to 46.5%. Rieke et al. [10] argue that federated learning allows models to be trained collaboratively without exchanging patient data, easing privacy and data-silo barriers. At programme level, All of Us [7] deliberately enrols participants from historically underrepresented groups, and the WHO Science Council [22] recommends fifteen actions to accelerate access to genomics worldwide, with particular attention to low- and middle-income countries.

Digital twins are a newer thread. Katsoulakis et al. [12] review digital twins for health and find the field still at an early stage, while Laubenbacher et al. [13] describe medical digital twins as potentially vital for personalised medicine. Together these studies show a field that is technically diverse but shares a common architecture of data integration, AI-based prediction, and clinician-validated decision support, and a common set of barriers. Table 2 summarises nine representative studies, their focus, method, strengths, and weaknesses.

Table 2. Comparison of Literature

Ref.	Focus / Domain	Method	Strength	Weakness
[3]	AI in medicine	Narrative review	Frames AI impact at clinician, health-system, and patient levels	Narrative format; no pooled quantitative comparison
[8]	Variant calling	Deep CNN on read-pileup images	Outperforms existing state-of-the-art callers	Limited to SNPs and small indels
[21]	Algorithmic bias	Empirical audit of a widely used risk algorithm	Shows cost used as a proxy for need causes racial bias	Audits a single commercial algorithm
[20]	Genomic diversity	Analysis of ancestry representation in genomic studies	Shows research is skewed to European ancestry	Remedies need sustained community engagement and investment
[15]	Diabetes	Expert consensus report	Defines six categories; monogenic diabetes care is a precision standard	Expert-opinion based rather than trial evidence
[10]	Privacy	Perspective on federated learning	Training across sites without exchanging patient data	Technical and governance challenges remain open
[18]	Personalised nutrition	800-person glucose-monitoring cohort, ML, dietary intervention	Predicts personal glucose response; validated in an independent 100-person cohort	Interventions were short-term
[17]	Polygenic risk	Genome-wide scores validated in UK Biobank	Flags 8.0% of people at over threefold risk of coronary artery disease	Evaluated within a single biobank
[19]	Rare disease	Rapid whole-genome sequencing with automated phenotyping	97% recall and 99% precision against expert review	Only seven intensive-care infants tested prospectively

3. PRECISION MEDICINE FRAMEWORK

Drawing on recurring architectural elements identified in the reviewed literature, this section proposes a conceptual framework for AI-enabled precision medicine. The framework synthesises reported practices rather than a system that has been experimentally validated, giving students a coherent mental model of how the technologies in Sections 1 and 2 fit together.

The framework, illustrated in Figure 3, begins with the Patient. Data are acquired from EHRs, laboratory tests, genomic sequencing, and wearable devices, and are integrated in a Secure Data Platform that handles consent, encryption, de-identification, and standardisation of formats. Where institutions cannot pool data, federated learning allows models to be trained across sites without exchanging patient-level records [10]. A pre-processing and feature-engineering layer then cleans the data and aligns and annotates sequence data, using variant callers such as DeepVariant [8].

The analytics layer hosts three cooperating engines. A Variant Interpretation engine ranks genomic variants by likely clinical significance, as in automated pipelines that pair genome interpretation with phenotypes extracted from health records [19]. A Biomarker Discovery engine looks for molecular signatures of disease subtype and treatment response, including image-derived markers such as microsatellite instability predicted from histology [9]. A Risk Prediction engine estimates disease risk and trajectory, for example through polygenic scores [17] or glucose-response models [18]. Their outputs converge in a Treatment Recommendation engine that matches the patient's profile to targeted therapies, drug choice, and dosing guidance [5], [4].

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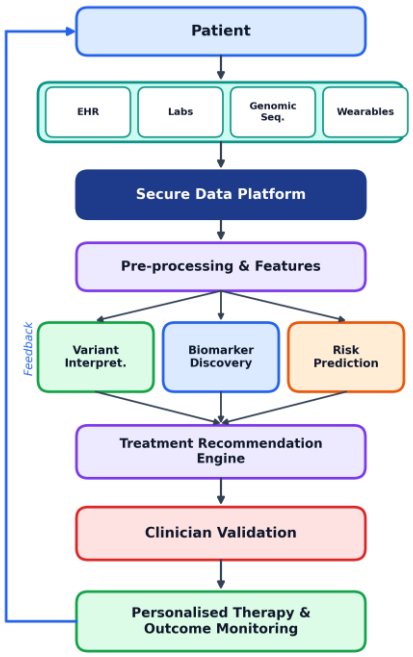

Figure 3. Proposed Framework for AI-Enabled Precision Medicine

Recommendations pass to a Clinician Validation step in which a doctor reviews, accepts, or overrides them, since AI is best viewed as a tool that supports, rather than replaces, human judgement [16]. The chosen Personalised Therapy is delivered, and Outcome Monitoring, using clinical follow-up and wearable data, feeds results back to the data platform, closing the loop.

This closed loop is best understood alongside the broader precision medicine lifecycle in Figure 4, spanning eight stages: data collection, sequencing, interpretation, risk prediction, diagnosis, treatment, monitoring, and learning. A model built at an early stage can be refined with real-world outcome data at every later stage, so the same data that support a diagnosis later inform prognosis and follow-up [15], [11].

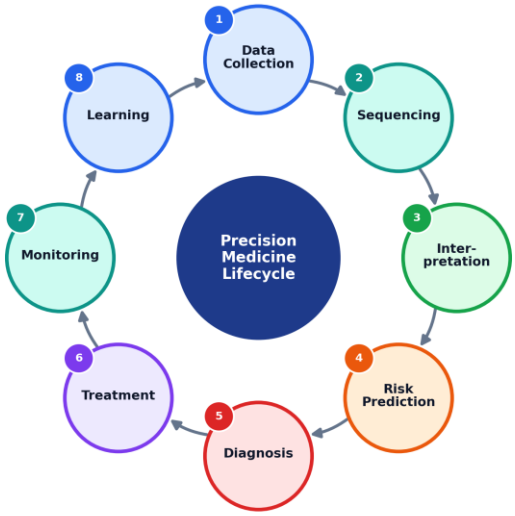

Figure 4. Precision Medicine Lifecycle

The framework also depends on a wider ecosystem around the core acquire-analyse-decide-act loop. As Figure 5 shows, AI-enabled precision medicine sits at the centre of a constellation that includes genomics and sequencing, bioinformatics, big-data analytics, EHRs and imaging, wearables and IoT, cloud computing, federated learning, and explainable AI with ethical oversight [11], [16]. Table 3 compares the proposed approach with three simpler classes of care, to show why integrating genomic and clinical data with clinician validation is needed for individualised care.

Figure 5. Precision Medicine Ecosystem

Table 3. Comparison of Care Approaches

Approach	Data Used	Key Features	Limitations
Traditional (one-size-fits-all)	Population-level trial evidence	Standard guidelines and doses for all patients with a diagnosis	Individual response varies; failures and side effects found after the fact
Stratified medicine	A single biomarker or clinical subgroup	Groups patients, for example by blood type or receptor status	Coarse groups that ignore multi-factor individual variation

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Approach	Data Used	Key Features	Limitations
Clinical-data AI (no genomics)	EHR, laboratory, and imaging data	Risk prediction and decision support	Misses inherited and tumour-molecular drivers; biased if data are unrepresentative [21]
AI + genomics precision medicine (proposed)	Genomic, clinical, imaging, and wearable data	Biomarker discovery, individual risk and treatment prediction, clinician validation	Sequencing cost, privacy, need for diverse data, and clinical validation

4. CLINICAL APPLICATIONS

Precision medicine has moved beyond its oncology origins into many clinical areas. This section summarises six application domains drawn from the reviewed literature, with emphasis on cancer and diabetes. Table 4 compares them.

4.1 Oncology

Cancer was the first major focus of precision medicine, and the 2015 US initiative gave it a near-term emphasis on cancers [1]. Tumour sequencing identifies actionable mutations that guide targeted therapy: specific EGFR mutations in non-small-cell lung cancer correlate with clinical responsiveness to gefitinib, and screening for them can identify patients likely to respond [5]. AI extends this in two ways. Deep learning variant callers improve the accuracy of mutation detection [8], and deep residual networks can predict microsatellite instability, a determinant of response to immunotherapy, directly from routine histology slides, which could widen access to such testing in gastrointestinal cancer [9]. Figure 6 illustrates a typical AI-assisted workflow.

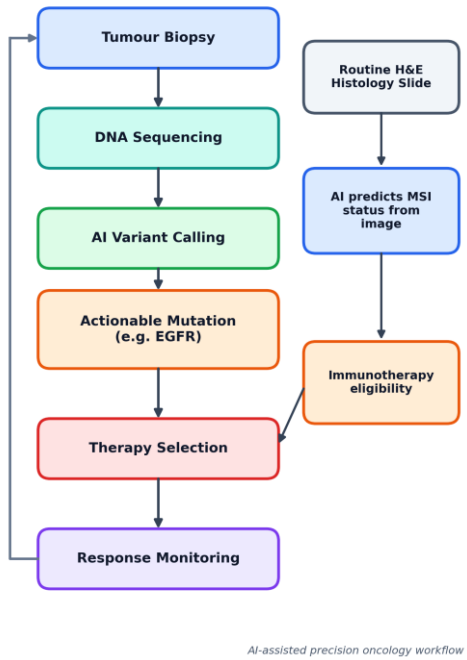

Figure 6. AI-Assisted Precision Oncology Workflow

4.2 Diabetes

Diabetes is a second flagship domain. The ADA and EASD consensus report defines precision diabetes medicine as an approach that integrates multidimensional data to optimise diagnosis, prediction, prevention, or treatment, organised into six categories [15]. Monogenic forms, such as neonatal diabetes and maturity-onset diabetes of the young, account for only 2–3% of people with diabetes, yet for them genetic diagnosis already guides standard care [15].

For the much larger group with common diabetes, data-driven approaches focus on prediction and management. Zeevi et al. [18] monitored glucose continuously in 800 people, recorded responses to 46,898 meals, and found high variability in the response to identical meals, suggesting that universal dietary advice has limited utility. Their machine learning model, integrating blood parameters, diet, anthropometrics, physical activity, and gut microbiota, predicted individual responses and was validated in an independent 100-person cohort. Polygenic scores can also flag people at elevated risk, with 3.5% of the population at more than threefold risk of type 2 diabetes in one study [17]. Figure 7 shows an integrated precision diabetes pathway.

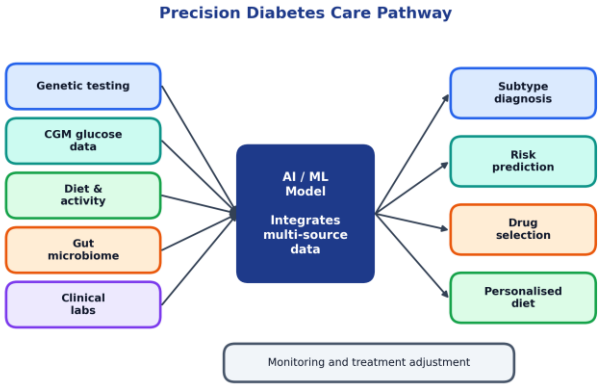

Figure 7. Precision Diabetes Care Pathway

4.3 Cardiovascular Disease

In cardiovascular disease, individualised prevention begins with risk. Khera et al. [17] show that genome-wide polygenic scores identify 8.0% of the population at more than threefold risk of coronary artery disease and 6.1% for atrial fibrillation, and propose that polygenic risk prediction be considered for clinical care. AI-powered predictive models have also been used to assess individual cardiovascular risk factors and recommend personalised interventions [6].

4.4 Rare Genetic Disorders

Rare genetic diseases are a natural target for genomic diagnosis. Clark et al. [19] built a platform that combines rapid whole-genome sequencing with automated phenotyping from health records and automated interpretation. Retrospectively, in 95 children with 97 genetic diseases, automated diagnoses agreed with expert manual interpretation (97% recall, 99% precision). Prospectively, the platform correctly diagnosed three of seven seriously ill intensive-care infants, in each case with an effect on treatment, and the median time from sequencing to interpretation was about 20 hours.

4.5 Pharmacogenomics

Pharmacogenomics matches drugs and doses to inherited variation. After decades of discovery, inherited variation in about 20 genes affecting about 80 medications has been identified as actionable in the clinic, and somatic variants guide the choice of some targeted anticancer drugs;

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implementation, rather than discovery, is now the main task [4]. This offers a route to safer and more effective prescribing than trial-and-error.

4.6 Lifestyle and Remote Monitoring

Wearable devices and biosensors make it possible to track physiology continuously outside the clinic. Multimodal AI combines these streams with genomic, imaging, and EHR data for personalised medicine, remote monitoring and care, and digital clinical trials [11]. AI integrated with wearables and telemedicine platforms supports full-cycle health monitoring, although data standardisation, privacy, and accuracy across diverse populations remain open issues [16].

Taken together, the literature points to recurring advantages: improved diagnostic accuracy, personalised therapies, earlier risk stratification, and better use of medications [3], [4], [17], [19]. These benefits are unevenly supported by evidence, however: the evidence base differs across specialties, and technical performance in controlled studies does not automatically translate into better patient outcomes in routine practice [16].

Table 4. Comparison of Clinical Application Domains

Domain	Representative Applications	Key Benefits	Main Challenges
Oncology	Tumour sequencing, targeted therapy selection, histology-based biomarker prediction	Therapy matched to tumour molecular profile	Cost of sequencing; need for validated, actionable biomarkers
Diabetes	Monogenic diagnosis, glucose-response prediction, personalised diet, risk scores	Better subtype diagnosis and individual glycaemic management	Evidence largely short-term; most patients do not have monogenic forms
Cardiovascular	Polygenic risk scores, wearable-based monitoring, AI risk models	Earlier identification of high-risk individuals	Scores validated in limited cohorts; ancestry bias
Rare genetic disorders	Rapid whole-genome sequencing with automated interpretation	Faster diagnosis (median about 20 hours) with treatment impact	Small prospective samples; infrastructure and cost
Pharmacogenomics	Genotype-guided drug and dose selection	Safer and more effective prescribing	Integration into routine clinical workflows
Lifestyle and remote monitoring	Wearables, continuous glucose monitoring, multimodal AI	Continuous tracking outside the clinic	Data standardisation, privacy, accuracy across populations

5. CHALLENGES AND MITIGATION STRATEGIES

Despite clear benefits, AI-enabled precision medicine faces persistent challenges that recur across the reviewed studies. Data privacy and security are central: health data are highly sensitive and tightly regulated, and much of it sits in separate institutional silos, which limits the data available to train models [10]. Algorithmic bias is a second major concern. Obermeyer et al. [21] show how using cost as a proxy for need produced racial bias in a widely used algorithm, and Zeng et al. [16] identify fairness, interpretability, and generalisability as key barriers to clinical translation.

Unrepresentative genomic datasets compound these problems. Genomic research has been heavily skewed towards European-ancestry populations [20], so findings, risk scores, and variant interpretation may transfer poorly to other groups. Programmes such as All of Us, in which over 80% of participants contributing biospecimens by July 2019 came from historically underrepresented groups, attempt to correct this [7]. Cost and access are closely linked: the WHO Science Council argues that it is neither ethically nor scientifically justifiable for less-resourced countries to gain access to genomic technologies long after wealthier ones, and calls for addressing shortfalls in financing, laboratory infrastructure, and trained personnel [22].

Data integration and evidence quality are further limits. Combining heterogeneous data from EHRs, imaging, sequencing, and biosensors is itself a technical and analytical challenge [11], models can generalise poorly beyond the settings in which they were built [16], and a lack of transparency can make clinicians reluctant to act on AI output [3]. Table 5 summarises the main challenges, their impact, and possible mitigation strategies.

Table 5. Challenges and Mitigation Strategies

Challenge	Impact	Possible Mitigation
Data privacy and security	Sensitive health data restrict sharing and risk breaches	Federated learning, encryption, de-identification, consent and governance
Algorithmic bias	Unequal care across patient groups	Avoid cost-proxy labels, audit models for fairness, include diverse validation data
Unrepresentative genomic data	Findings and risk scores transfer poorly to underrepresented ancestries	Inclusive cohorts such as All of Us; diverse recruitment
Cost and infrastructure	Limited access, especially in low- and middle-income countries	Financing, shared laboratory capacity, equitable technology transfer
Data integration	Fragmented records make multimodal analysis hard	Common data standards and multimodal pipelines

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Challenge	Impact	Possible Mitigation
Model generalisability	Performance may drop at new sites or in new populations	External validation and multi-centre studies
Interpretability and trust	Clinicians hesitate to act on opaque outputs	Explainable AI and a clinician validation step
Ethical, legal, and social issues	Gaps in consent, accountability, and governance	Oversight frameworks and international guidance
Skills shortage	Too few people trained across genomics, data science, and clinical practice	Training programmes and interdisciplinary teams

6. FUTURE RESEARCH DIRECTIONS

The literature points towards several directions likely to shape the next generation of precision medicine. Digital twins, personalised virtual patient models that are updated with new data and used for prediction, are among the most discussed: Laubenbacher et al. [13] describe medical digital twins as potentially vital for personalised medicine, while Katsoulakis et al. [12] conclude that the field is still at an early stage. Multimodal AI that combines genomic, imaging, EHR, and wearable data is expected to capture the complexity of health and disease more fully [11], [6].

Federated learning is gaining traction for training shared models across hospitals without centralising patient data, directly addressing the privacy and data-silo problems of Section 5 [10]. Explainable and human-centred AI designs aim to make recommendations understandable and trusted, keeping clinicians in the loop [16]. Real-time clinical decision support, in which streaming wearable and EHR data update risk estimates at the point of care, is a natural extension of the remote monitoring work in Section 4.6.

CRISPR-assisted therapies, which use genome editing to correct disease-causing variants, represent a longer-term direction in which genomic diagnosis could lead directly to individualised treatment; this remains an emerging area and lies outside the clinical evidence reviewed here. Finally, equitable genomics, through more diverse cohorts, fair algorithms, and wider global access, is a precondition for benefits to reach all populations [20], [7], [22]. Figure 8 summarises these directions.

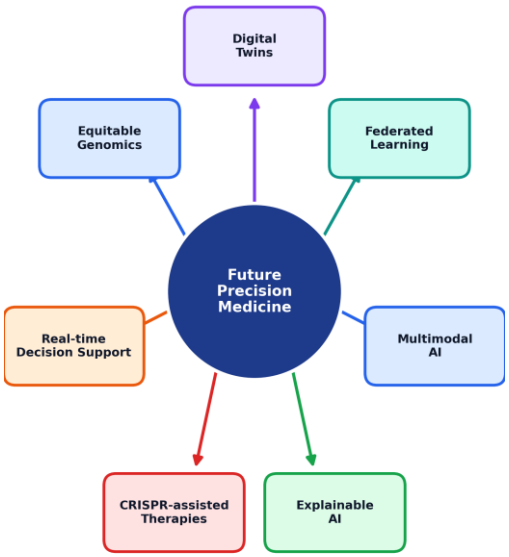

Figure 8. Future Directions in Precision Medicine

7. CONCLUSION

Precision medicine has moved from a concept to a clinical reality in selected areas. Genomic sequencing identifies the molecular basis of disease, and AI turns large, heterogeneous data into predictions and recommendations that clinicians can use. This review has traced the evolution of the field, compared how it has been defined and studied, proposed a conceptual framework linking data, AI analytics, clinician validation, and feedback, and examined applications in oncology, diabetes, cardiovascular disease, rare disorders, pharmacogenomics, and remote monitoring.

The evidence also shows that the benefits are uneven. Strong examples, such as targeted cancer therapy, monogenic diabetes, and rapid genomic diagnosis, sit alongside areas where evidence is early or short-term. Privacy, algorithmic bias, unrepresentative data, cost, and interpretability remain the main barriers, and they are as much social and organisational as technical. Mitigation strategies such as federated learning, fairness auditing, inclusive cohorts, explainable AI, and clinician-in-the-loop design are therefore as important as model accuracy.

The main limitations of this work are that it is a narrative review without a systematic search protocol, and that the proposed framework is conceptual and has not been validated experimentally. Future research should test such frameworks in prospective clinical settings, develop privacy-preserving and fair models, and widen access to genomic medicine globally. Pursued responsibly, the combination of AI and genomics can move healthcare from reactive treatment towards prediction, prevention, and care tailored to each individual.

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