

REVIEW ARTICLE

Can pharmacogenomics transform personalized medicine into routine clinical practice? Current evidence and future directions

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Abstract

Personalized medicine (PM) represents a transformative approach in healthcare, particularly in oncology, by tailoring treatments to individual genetic, molecular, and clinical profiles, thereby improving therapeutic specificity and minimizing adverse effects. A key component of this approach is pharmacogenomics (PGx), which examines the influence of genetic variations on drug response to optimize drug selection, dosing, and safety while reducing adverse drug reactions. This review highlights recent advances in molecular biology, PGx, and molecular diagnostics that enable accurate cancer subtype classification and the development of targeted therapies. A structured literature search was conducted to identify studies on genotype-guided therapies, pharmacogenomic biomarkers, molecular diagnostics, and clinical trials evaluating treatment efficacy and interpatient variability across diverse populations. The review emphasizes the role of predictive biomarkers and pharmacogenomic profiling in addressing heterogeneity in treatment response, facilitating individualized therapeutic strategies, and improving clinical outcomes. Despite significant progress in biological therapies and precision oncology, challenges related to biomarker validation, clinical implementation, accessibility, and cost remain. Continued multidisciplinary research and large-scale collaborative studies are essential to strengthen the clinical translation of PGx and PM. The integration of PGx, molecular diagnostics, and targeted therapies is expected to further advance precision oncology and improve patient outcomes worldwide.

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1. Introduction

The landscape of medical treatment is undergoing a profound transformation, evolving from conventional “one-size-fits-all” approaches toward precision-based strategies that account for interindividual variability in genetic, environmental, and lifestyle factors.^{1,2} Within this paradigm, pharmacogenomics (PGx), which examines the influence of genetic variation on drug response, has emerged as an important component of personalized

medicine (PM).^{3–5} By linking genetic information with variability in pharmacokinetics, pharmacodynamics, efficacy, and toxicity, PGx provides a molecular basis for understanding differences in therapeutic response among patients.

The development of the Human Genome Project, high-throughput sequencing, and other genomic technologies has substantially expanded the ability to identify clinically relevant genetic variants.^{6–9} Consequently, PGx has progressed from a primarily research-oriented discipline toward a clinically applicable approach across diverse therapeutic areas, including oncology, psychiatry, neurology, cardiovascular medicine, and infectious diseases.^{10,11} Regulatory recognition of PGx information has further supported this transition, with PGx associations incorporated into drug labeling for numerous therapeutically important medicines, including drugs affected by variants in *CYP2C9*, *VKORC1*, *CYP2C19*, and human leukocyte antigen (HLA) genes.^{12–14} Examples such as genotype-informed use of targeted anticancer therapies and PGx-guided selection of psychiatric medications illustrate the increasing clinical relevance of molecular information in treatment decision-making.^{14–16}

The expanding scope of PGx has also been accompanied by advances in sequencing, gene-editing technologies, single-cell analysis, artificial intelligence (AI), and other data-driven approaches.^{17–20} These developments are broadening the application of genomic information beyond conventional drug-response prediction toward more integrated approaches involving disease risk assessment, molecular characterization, and individualized therapeutic planning.^{21–23} As PGx increasingly intersects with broader PM frameworks, the field is moving toward the integration of genomic, molecular, and clinical information to support more comprehensive patient-specific decision-making.^{24,25}

Against this background, this review examines the evolving role of PGx within PM, with emphasis on its underlying principles, therapeutic applications, genetic testing, technological integration, clinical implementation, and emerging strategies.^{26,27} Particular attention is given to the transition from genetic information to actionable clinical decisions and to the frameworks and technologies that may facilitate the incorporation of PGx into routine healthcare.^{28,29}

This review highlights how PM, driven by PGx, is reshaping healthcare by tailoring treatments to an individual's genetic makeup to improve efficacy and reduce adverse effects. Advances in genomics, bioinformatics, and technologies such as CRISPR and AI are enabling more precise therapies across various fields, including oncology, psychiatry, and infectious diseases.³⁰ Despite great promise,

challenges remain, such as high costs, limited access, ethical concerns, and the need for clinician education and standardized guidelines.³¹

A structured literature search was conducted using PubMed, Scopus, Web of Science, ScienceDirect, and Google Scholar, with emphasis on publications from January 2015 through March 2026. The search strategy employed combinations of keywords, including “personalized medicine,” “precision medicine,” “pharmacogenomics,” “genomics,” “biomarkers,” “molecular diagnostics,” “omics technologies,” “artificial intelligence,” “machine learning,” “targeted therapy,” and “clinical applications,” combined using the Boolean operators AND and OR. Peer-reviewed articles published in English, including original research papers, systematic reviews, meta-analyses, clinical trials, and high-quality review articles relevant to the scope of PM, were included. Seminal earlier studies, relevant clinical guidelines, regulatory resources, and selected publications identified during manuscript revision through August 2026 were also included.

2. Pharmacogenomics

2.1. Understanding pharmacogenomics and drug response

Pharmacogenomics is the study of how genetic variations influence an individual's response to drugs. This field combines pharmacology (the study of drugs) and genomics (the study of genes and their functions) to develop safer and more effective medications tailored to an individual's genetic makeup.^{32,33} The genetic differences in drug-metabolizing enzymes, transporters, and receptors significantly impact how a drug is absorbed, distributed, metabolized, and excreted from the body.³⁴ These variations can determine whether a drug will be effective, require dose adjustments, or cause severe adverse effects in certain patients. For instance, clinically relevant variants in *CYP2D6* can alter the metabolism of drugs such as antidepressants, opioids, and beta-blockers, potentially contributing to therapeutic failure or adverse effects.³⁵ PGx testing helps identify these genetic variations before prescribing medications, thereby supporting precision dosing and reducing trial-and-error prescribing.^{36,37}

2.2. Importance of pharmacogenomics in drug development and precision prescribing

Pharmacogenomics is transforming drug development by providing insights into variability in drug response, which can improve the safety and efficacy of new therapeutics.³² During clinical trials, genetic screening can identify responders and non-responders, reduce the risk of adverse effects, and increase the likelihood of approval by

regulatory agencies such as the United States Food and Drug Administration (FDA) and the European Medicines Agency (EMA).³⁸ Additionally, PGx research has led to the development of companion diagnostics, which are genetic tests that guide the prescription of specific drugs. For example, the FDA-approved human epidermal growth factor receptor 2 (HER2) test determines whether a breast cancer patient will benefit from trastuzumab, a monoclonal antibody therapy targeting HER2-positive tumors.³⁹

Prescribing medications without relevant PGx information may expose patients to avoidable risks, including therapeutic failure, serious adverse drug reactions, inappropriate dosing, and prolonged trial-and-error treatment. For example, *CYP2C9/VKORC1* variants influence warfarin dose requirements, *DPYD* variants increase the risk of severe fluoropyrimidine toxicity, *CYP2D6* polymorphisms affect opioid and antidepressant response, and HLA-associated variants predict severe hypersensitivity reactions to drugs such as carbamazepine and abacavir. Incorporating validated PGx testing before prescribing clinically actionable medications can improve treatment efficacy, enhance patient safety, and reduce healthcare costs associated with preventable drug-related complications.^{40–42}

2.3. Application of pharmacogenomics in cancer therapy

Cancer therapy represents one of the most clinically developed areas for PGx application because genetic and molecular characteristics can influence drug metabolism, toxicity, and response to targeted treatments.⁴³ PGx enables clinically relevant variants to be incorporated into treatment selection and dose optimization. [Figure 1](#) illustrates the broader integration of big data analytics, precision early detection, targeted disease prevention, and advanced therapeutic testing in modern healthcare.

One established example is fluorouracil (5-FU), a

commonly used chemotherapeutic agent in colorectal and breast cancers.⁴⁴ The *DPYD* gene encodes dihydropyrimidine dehydrogenase (DPD), the principal enzyme involved in 5-FU catabolism. Reduced DPD activity associated with clinically relevant *DPYD* variants can result in excessive systemic exposure to 5-FU and an increased risk of severe or potentially life-threatening toxicity. PGx testing can therefore identify patients with relevant *DPYD* variants before treatment, allowing dose modification or consideration of alternative therapy where appropriate.⁴⁵

Other clinically relevant gene–drug pairs further illustrate the role of genotype-informed treatment in oncology. Variants in *TPMT* (thiopurine S-methyltransferase) and *NUDT15* (nudix hydrolase 15) influence thiopurine metabolism and toxicity. Patients with reduced or absent activity of these enzymes have an increased susceptibility to severe myelosuppression following treatment with thiopurines such as mercaptopurine, thioguanine, and azathioprine; consequently, genotype-guided dose adjustment can be considered before therapy.⁴⁶ Similarly, genetic variation in *UGT1A1*, particularly the *UGT1A1**28 allele, can reduce glucuronidation of the active metabolite of irinotecan and increase susceptibility to adverse effects such as neutropenia and diarrhea. Pretreatment assessment of relevant *UGT1A1* variants can therefore provide information for treatment and dose selection.⁴⁷

Pharmacogenomics also contributes to molecularly guided treatment of non-small cell lung cancer (NSCLC). Activating mutations in *EGFR* can support the selection of epidermal growth factor receptor (EGFR) tyrosine kinase inhibitors (TKIs), whereas *ALK* rearrangements can identify patients who may benefit from anaplastic lymphoma kinase (ALK)-targeted therapies.^{48,49} These examples illustrate how molecular and PGx information can be incorporated into treatment selection for genetically defined cancer subgroups.^{50,51}

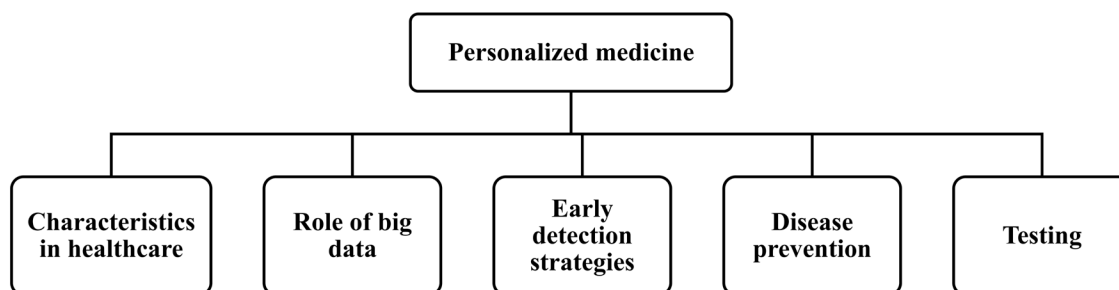


Figure 1. Multidimensional framework of personalized medicine: Interplay of big data analytics, precision early detection, targeted disease prevention, and advanced therapeutic testing in modern healthcare systems. Created using power point.

Beyond oncology, clinically actionable gene–drug associations have also been established in other therapeutic areas. Variants in *SLCO1B1* are associated with an increased risk of statin-associated muscle toxicity, particularly with simvastatin, while *CYP3A5* variation contributes to interindividual differences in tacrolimus metabolism and dose requirements in organ transplantation.⁵² These examples demonstrate that the application of PGx extends beyond cancer therapy and provides a basis for its broader integration into individualized drug selection and dosing across therapeutic areas.⁵³

3. Personalized medicine

3.1. Characteristics of personalized medicine and its impact on healthcare

Personalized medicine represents a broader clinical framework in which patient-specific characteristics are integrated to guide disease prevention, diagnosis, and treatment. The integration of electronic health records (EHRs) with omics technologies, particularly genomics, further facilitates the systematic incorporation of patient-specific information into clinical practice and supports the development of predictive, preventive, personalized, and participatory (P4) medicine.^{54,55}

Pharmacogenomics constitutes an important component of PM, but the two concepts are not synonymous. PGx primarily focuses on the influence of genetic variation on drug response, including differences in therapeutic efficacy, drug metabolism, dosing requirements, and susceptibility to adverse drug reactions. In contrast, PM encompasses a broader range of patient-specific determinants and integrates genetic information with molecular, clinical, environmental, and lifestyle factors to guide healthcare decisions. Thus, PGx can be considered a specific genomic component within the wider PM framework, providing information that can be incorporated into individualized drug selection and dosing decisions.^{56–58}

3.2. Big data analytics and multi-omics integration in personalized medicine

The implementation of PM increasingly requires the integration of diverse and high-dimensional datasets beyond individual genetic variants. In addition to genomics and PGx, relevant data may include transcriptomics, proteomics, metabolomics, epigenomics, microbiomics, clinical phenotypes, EHRs, and, increasingly, information obtained from wearable and digital health technologies. Whereas PGx primarily provides information on genetic determinants of drug response, these complementary datasets capture dynamic biological and clinical processes

that may influence disease phenotype and therapeutic response. Integrating these data layers can therefore provide a more comprehensive representation of individual biological states and support the identification of clinically relevant molecular patterns and biomarkers.^{59,60}

However, the integration of multi-omics and clinical datasets presents substantial data-management challenges. These datasets differ considerably in their **structure**, dimensionality, scale, measurement technologies, and data formats, resulting in considerable heterogeneity. For example, genomic data are generally represented as sequence variants, whereas transcriptomic, proteomic, and metabolomic datasets contain quantitative measurements obtained using different analytical platforms. EHRs provide largely heterogeneous clinical information, including structured laboratory and medication records alongside unstructured clinical notes. Differences in data acquisition methods, preprocessing procedures, reference standards, and metadata further complicate the integration and comparison of datasets generated across studies and healthcare institutions.^{15,59}

Data interoperability and standardization represent additional challenges for PM implementation. Inconsistent terminologies, coding systems, data models, and metadata conventions can limit the ability to combine datasets from different sources. Standardized vocabularies, common data models, ontologies, and interoperable data-exchange frameworks are therefore required to facilitate consistent representation and linkage of genomic, multi-omics, and clinical information. In addition, variations in data quality, missing observations, batch effects, measurement errors, and differences in sampling and experimental conditions can introduce systematic biases and reduce the reliability of integrated analyses.¹⁵

The large volume and complexity of these datasets also create substantial computational requirements. Next-generation sequencing (NGS), high-throughput omics platforms, biobanks, EHR systems, and wearable devices continuously generate biomedical data at a scale that exceeds the capacity of conventional data-processing approaches. High-performance computing, cloud-based infrastructures, distributed computing, and scalable analytical frameworks can facilitate the storage, processing, and integration of these large datasets. Nevertheless, the increasing complexity of data integration requires analytical approaches capable of handling high dimensionality and identifying meaningful relationships across multiple biological and clinical data layers.⁶¹

Artificial intelligence and machine learning (ML) approaches provide computational strategies for addressing some of these challenges by identifying

complex and nonlinear relationships within integrated datasets. When appropriately validated, these methods can assist in extracting relevant patterns from multi-omics and clinical data and support the development of predictive models. However, the usefulness of AI/ML-based approaches depends strongly on the quality, representativeness, standardization, and interoperability of the underlying datasets. Differences between training and clinical populations, incomplete datasets, batch effects, and insufficient external validation may affect model generalizability and reproducibility. Thus, the integration of multi-omics, EHR, and other patient-derived data requires not only advanced computational methods but also robust standards for data quality, harmonization, interoperability, and validation.^{1,62}

3.3. Personalized early detection strategy

Individuals with an elevated risk of disease or disease recurrence may warrant longitudinal monitoring. Risk can be estimated using family history, genetic testing, polygenic risk score (PRS), biomarkers, clinical history, and lifestyle or environmental factors. To establish assertions concerning evidence or indicators of sickness or a pathogenic process, it is presently thought that such monitoring should be done using “personal thresholds” as opposed to “population thresholds.”^{63,64} Population thresholds include, for instance, systolic blood pressure > 140 as an indicator of hypertension, risk of stroke or heart disease, or cholesterol levels > 200 as an indicator of risk of heart disease. Population thresholds are obtained from epidemiologic data and demographic surveys.^{65,66} Personal thresholds are determined by historical values of a measure that have been gathered over time on a person and are used to predict how skewed future values of that measure may be for that individual. Whether or not the values are over a population threshold, clinically meaningful departures from historical or average legacy values are interpreted as an indication of a change in the health state of the population.^{67,68}

3.4. Personalized disease prevention

Personalized medicine has expanded the scope of disease prevention by enabling preventive strategies to be informed by individual genetic and molecular risk profiles. Genetic risk assessment can identify individuals with an increased susceptibility to particular diseases and may support earlier intervention, risk-adapted screening, and targeted preventive strategies before clinical manifestations occur. However, the predictive value of genetic information is not uniform across diseases, and the identification of a genetic risk variant does not necessarily indicate that an individual will develop the corresponding disease. Disease

susceptibility is often determined by the combined effects of multiple genetic variants, molecular characteristics, environmental exposures, lifestyle factors, and other clinical determinants. Therefore, genetic risk information should be interpreted within the broader biological and clinical context rather than considered as an independent predictor of disease development.⁶⁹

Colorectal cancer provides an important example of how genetic information may contribute to individualized prevention and risk-based therapeutic strategies. Although early detection and prevention have substantially improved outcomes, colorectal cancer remains a major cause of cancer-related morbidity and mortality. Genetic and molecular biomarkers can provide information regarding disease susceptibility and may also identify patient subgroups with differential responses to preventive interventions.⁷⁰ For example, patients with colorectal tumors carrying *PIK3CA* mutations experienced improved overall survival and reduced colorectal cancer-specific mortality following postoperative aspirin use compared with patients with *PIK3CA* wild-type tumors. Subsequent investigations have further suggested that the preventive effects of aspirin may vary according to molecular characteristics, supporting the potential role of biomarker-guided prevention.⁷¹

Despite these advances, the translation of genetic and molecular risk information into routine preventive care requires careful consideration of predictive accuracy and clinical utility. A biomarker may demonstrate a statistically significant association with disease risk without providing sufficient predictive performance to alter clinical management. Similarly, the magnitude of an individual's genetic risk may be difficult to interpret when absolute disease risk is influenced by multiple interacting factors. Therefore, candidate biomarkers require rigorous analytical and clinical validation in diverse populations to establish their reproducibility, predictive performance, and relevance to clinically meaningful outcomes. The incremental value of a genetic or molecular marker over established clinical risk factors should also be demonstrated before its incorporation into preventive decision-making.⁷²

Appropriate interpretation and communication of predictive information are equally important. Overestimation of genetic risk may result in unnecessary surveillance or preventive interventions, whereas underestimation may lead to false reassurance and delayed preventive care. Consequently, personalized prevention should incorporate validated risk estimates, relevant clinical and environmental factors, and evidence-based thresholds for intervention. Rather than relying solely on individual genetic variants, future preventive strategies

are likely to benefit from integrated risk models that combine genetic, molecular, phenotypic, and conventional clinical risk factors. Such an approach may improve the clinical relevance of predictive information while reducing inappropriate application of genetic risk assessment in disease prevention.⁷³

3.5. Testing in personalized medicine

The clinical implementation of PM requires accurate, reliable, and clinically validated diagnostic tests capable of identifying actionable genetic and molecular biomarkers. For PGx testing to provide clinically useful information, the selection of relevant genetic variants should be supported by robust evidence linking genotype with drug response or toxicity. In addition, genetic assays should demonstrate adequate analytical validity, while the clinical validity and clinical utility of identified variants should be established before their incorporation into therapeutic decision-making. The timely availability and appropriate interpretation of test results are also important, particularly when genetic information is required before treatment initiation.⁷⁴

Standardized testing procedures, variant nomenclature, reporting formats, and evidence-based interpretation frameworks are necessary to ensure consistency and reproducibility across laboratories and clinical settings. Integration of genetic test results with EHRs and clinical decision support (CDS) systems can further facilitate the incorporation of actionable PGx information into routine clinical workflows, allowing relevant genotype-guided recommendations to be available at the point of prescribing.⁷⁵

3.5.1. Role of genetic testing in advancing personalized medicine

Genetic testing is an important component of PM because it enables the identification of inherited and acquired genetic variations associated with disease susceptibility and therapeutic response. By analyzing an individual's genomic profile, clinicians can obtain molecular information that can complement conventional clinical characteristics and support individualized prevention, diagnosis, and treatment strategies. Genes encode proteins involved in drug absorption, distribution, metabolism, and excretion, cellular signaling, immune responses, and disease pathogenesis. Genetic variations within these genes may alter protein expression or function, thereby contributing to interindividual differences in pharmacokinetics, pharmacodynamics, disease susceptibility, and therapeutic response. The relationship between the genome, transcriptome, and proteome, illustrated in [Figure 2](#), highlights how genetic information can influence the

production and function of proteins involved in disease processes and drug response.^{76–78}

The clinical relevance of genetic testing depends on the strength of evidence linking specific variants to clinically meaningful phenotypes or treatment responses. Accordingly, genetic information can contribute to the identification of individuals with clinically relevant genetic characteristics and support the selection or modification of therapeutic strategies when validated genotype–phenotype relationships are available. As genomic technologies continue to advance, appropriately validated genetic testing can provide an increasingly important molecular basis for individualized clinical decision-making.⁷⁹

3.5.2. Clinical example of pharmacogenomic implementation

One of the well-established clinical applications of pharmacogenomic testing is genotype-guided warfarin therapy. As discussed in [Section 2.2](#), genetic variants in *CYP2C9* and *VKORC1* contribute to interindividual variability in warfarin dose requirements. Consequently, PGx testing enables clinically relevant genetic information to be incorporated into dosing algorithms alongside conventional clinical factors, thereby supporting individualized anticoagulant therapy.^{80,81}

This example illustrates the essential components required for successful clinical implementation of PGx testing, including accurate identification of actionable genetic variants, validated genotype–phenotype associations, standardized interpretation of test results, and integration of genetic information into clinical decision-making. More broadly, it demonstrates how validated genomic investigations can support precision prescribing and improve the safety and effectiveness of drug therapy within the framework of PM.⁸²

3.6. Factors affecting the development of personalized medicine

3.6.1. Patients/consumers

As patients increasingly grow aware of the benefits of PM, including the power of genome sequencing, patients will begin to behave more like consumers, creating demand. Patients will strongly desire more data-driven, personalized care delivery than has been the case in the past, where decisions were often made with intuition and experience of individual physicians.⁸³ Additionally, patients will desire to be more actively engaged in shared decision-making and have visibility into their data and key health indicators. For example, the Accu-ChekView bundle by Roche Diagnostics helps patients with diabetes take control of their disease and prevent type 2 diabetes from forming by helping them

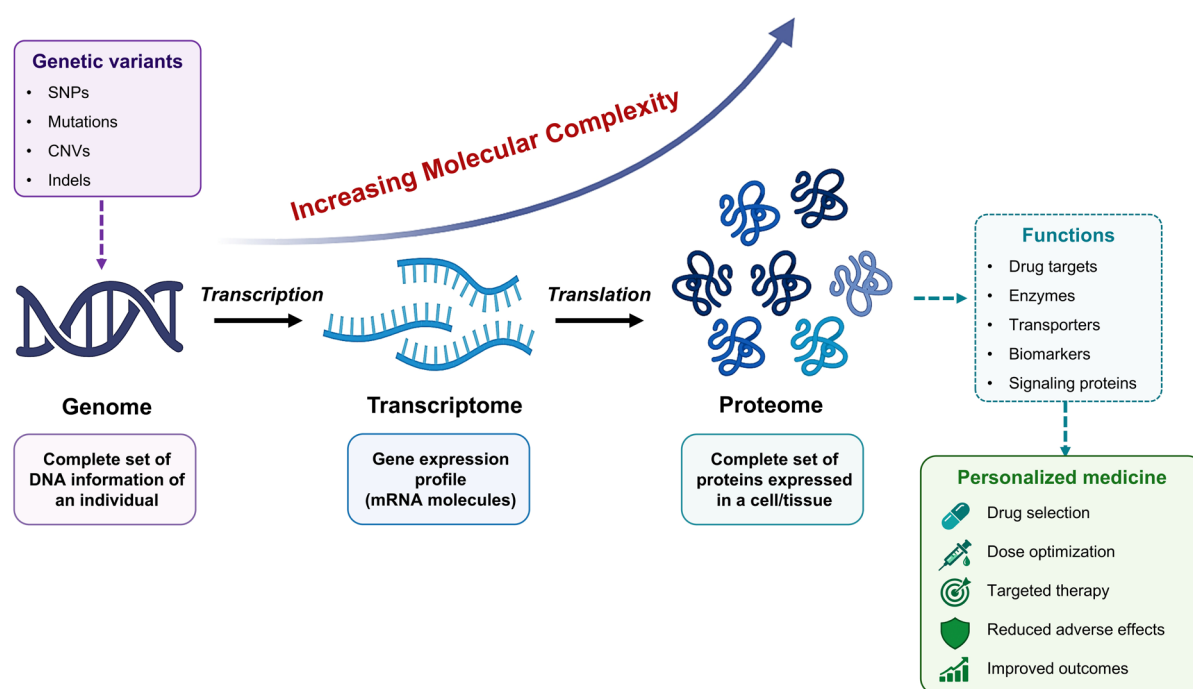


Figure 2. Multi-omics integration: From genomic foundations to transcriptomic and proteomic complexity. Created in BioRender.
Abbreviations: CNVs: Copy number variants; SNPs: Single-nucleotide polymorphisms

to change their lifestyle.⁸⁴ Applications such as the Accu-ChekView bundle, as well as gamification products such as wearable fitness trackers, all help increase the education and understanding of one's health. These change management agents are critical because even if a provider has access to genome sequencing, the best diagnostics, and personalized treatments, the outcome of care greatly depends on a patient's ability to change their behavior when they leave the doctor's office.^{82,85} A physician has no control over the patient's ability to fill a prescription, nor can they control the patient's habits, such as smoking, exercise, or healthy nutrition. Education, gamification, and health trackers are imperative resources for helping patients become partners in their health.⁸⁶

3.6.2. The shift to value-based care

Doctors are increasingly being compensated and measured by the value they provide instead of the number of patients they see or the procedures they perform. To succeed in the era of value-based care, providers and organizations will need to improve the effectiveness and efficiency of their care delivery. This will be a strong force in accelerating the adoption of PM.⁸⁷ In addition to Roche Diagnostics, other companies have also teamed with SAP to create

mobile apps that help facilitate patient engagement in their healthcare. Heidelberg University Hospital released an app that helps pregnant mothers connect with their doctors at all times, enabling the constant exchange of information and the ability to monitor the patient remotely and help catch early signs of prenatal depression.⁸³

3.6.3. Advanced computational infrastructure and data analytics

The successful development and widespread implementation of PM depend heavily on advanced computational infrastructure capable of managing and interpreting the vast amounts of genomic, multi-omics, imaging, and clinical data generated in modern healthcare. Unlike conventional healthcare systems, PM requires the integration of heterogeneous datasets obtained from whole-genome sequencing, transcriptomics, proteomics, metabolomics, EHRs, and wearable health devices. Processing these complex datasets efficiently requires high-performance computing, cloud-based computing platforms, and big data analytics, which together provide the computational capacity necessary for rapid data storage, processing, and interpretation. AI and ML can further support implementation by translating complex

biomedical data into predictive biomarkers, genotype-guided treatment recommendations, dosing support, and adverse drug reaction alerts. Their clinical value, however, depends on validated models, representative datasets, interoperability, and secure data-sharing practices. Continued development of AI-enabled clinical decision support may therefore improve the scalability and routine use of PM.⁸⁸

3.7. Advantages and disadvantages of personalized medicine

The principal advantages and limitations of PM are summarized in Table 1.

Personalized medicine enables more precise disease prevention, diagnosis, and therapy by incorporating patient-specific information, whereas challenges related to clinical validation, implementation, equitable access, cost, and data governance continue to influence its routine clinical adoption. Continued advances in genomic technologies and healthcare infrastructure are expected to address many of these limitations.

3.8. Application of personalized medicine

3.8.1. Oncology: Targeted cancer therapies

Personalized medicine has revolutionized oncology by enabling therapies tailored to a tumor's molecular profile. For HER2-positive breast cancer, trastuzumab (a monoclonal antibody) improves survival by 50% compared to conventional chemotherapy when prescribed based on HER2/neu amplification detected by immunohistochemistry (IHC) or fluorescence *in situ* hybridization (FISH).⁸⁹ Similarly, in NSCLC, patients harboring activating *EGFR* mutations are identified through molecular profiling and selected for osimertinib therapy, enabling individualized treatment based on each patient's tumor genotype rather than empirical treatment decisions. While clinical trials have reported high overall response rates, treatment selection is guided by the

patient's molecular profile, which informs the choice of an appropriate TKI.⁴⁸ Liquid biopsies now allow real-time monitoring of resistance mutations (e.g., *EGFR* T790M), facilitating timely therapy switches (Table 2). Challenges include tumor heterogeneity and cost, but NGS panels (e.g., MSK-IMPACT) are making genomic profiling more accessible.^{90,91}

3.8.2. Cardiology: Genetic insights in anticoagulants

Pharmacogenomics optimizes anticoagulant use, particularly for warfarin, where *CYP2C9* and *VKORC1* variants influence dosing. Patients carrying decreased-function *CYP2C9* alleles, such as *CYP2C9**2 and *CYP2C9**3, generally require lower warfarin doses (1–2 mg/day) and may have an increased risk of bleeding, whereas patients with normal *CYP2C9* function may require higher maintenance doses (5–7 mg/day). Warfarin therapy is individualized according to each patient's *CYP2C9* and *VKORC1* genotype, allowing personalized dose optimization to achieve therapeutic anticoagulation while minimizing bleeding risk.⁹² For clopidogrel, *CYP2C19* genotype-guided therapy, including the use of alternative P2Y12 inhibitors such as ticagrelor in poor metabolizers, has been associated with an approximately 50% reduction in stent thrombosis compared with conventional therapy.⁹³ Despite FDA recommendations, adoption remains limited due to testing delays and inconsistent reimbursement, as shown in Table 3.

3.8.3. Psychiatry: Precision prescribing

Antidepressant selection based on *CYP2D6*/*CYP2C19* genotypes reduces trial-and-error prescribing by 40%.¹⁰ For example, *CYP2D6* poor metabolizers experience severe side effects with paroxetine and benefit from alternative selective serotonin reuptake inhibitors such as sertraline. *HLA-B15:02* screening is recommended before carbamazepine therapy in genetically susceptible populations to prevent Stevens–Johnson syndrome and toxic epidermal necrolysis. In addition, *HLA-A31:01*

Table 1. Advantages and disadvantages of personalized medicine

Advantages	Disadvantages
• Reduce the likelihood of therapy side effects • Knowledge about the population or particular patient treatments utilized. • Interpreting genomic data. • Improving patient care with individualized medication. • Increased diagnostic accuracy and tailored medicine delivery. • Improves the precision of diagnosis.	• Costly and inaccessible • It is not financially feasible to target a tiny patient population. • Unlicensed technology • Concern about data leaks • Providers of services are uncommon. Need to build additional tools.

Table 2. FDA-approved pharmacogenomic biomarker-guided targeted therapies in oncology

Cancer Type	Pharmacogenomic Biomarker	Companion diagnostic/test	Targeted therapy	Efficacy improvement	Key clinical outcome
Breast cancer (HER2-positive)	HER2 amplification/overexpression	IHC and/or FISH	Trastuzumab ⁸⁹	Approximately 50% reduction in recurrence when combined with chemotherapy	Improved disease-free and overall survival in HER2-positive patients
Non-small cell lung cancer (EGFR-mutated)	EGFR exon 19 deletion or L858R mutation	PCR/NGS	Osimertinib ⁴⁸	Approximately 80% objective response rate with significantly prolonged progression-free survival	Superior central nervous system penetration and delayed resistance compared with first-generation EGFR tyrosine kinase inhibitors
Melanoma (BRAF V600E-mutated)	BRAF V600E mutation	PCR/NGS	Vemurafenib ⁹¹	Approximately 60% 1-year overall survival compared with conventional therapy	Rapid tumor regression and prolonged survival in metastatic melanoma

Abbreviations: BRAF: B-Raf proto-oncogene, serine/threonine kinase; EGFR: Epidermal growth factor receptor; FDA: Food and Drug Administration; FISH: Fluorescence in situ hybridization; HER2: Human epidermal growth factor receptor 2; IHC: Immunohistochemistry; NGS: Next-generation sequencing; PCR: Polymerase chain reaction.

Table 3. Pharmacogenomics-guided dosing for cardiovascular drugs

Drug	Pharmacogenomic biomarker	Clinical indication	Genotype-based recommendation	Clinical benefit	Clinical outcome
Warfarin ⁹²	CYP2C9 ^a /VKORC1	Anticoagulation (atrial fibrillation, venous thromboembolism)	Reduced CYP2C9 activity and VKORC1 variants require lower initial doses and careful dose titration	Genotype-guided dosing reduced major bleeding events by approximately 30% compared with conventional dosing	Improved therapeutic INR control and reduced bleeding complications
Clopidogrel ⁹³	CYP2C19 ^b	Prevention of thrombotic events following PCI/acute coronary syndrome	CYP2C19 poor metabolizers should receive alternative P2Y12 inhibitors (prasugrel or ticagrelor)	Genotype-guided therapy reduced stent thrombosis by approximately 50% in poor metabolizers	Improved platelet inhibition and reduced major adverse cardiovascular events

Notes: ^aCYP2C9 is primarily responsible for S-warfarin metabolism; ^bCYP2C19*2 and CYP2C19*3 are no-function alleles, whereas CYP2C19*17 is an increased-function allele. Abbreviations: CYP2C9: Cytochrome P450 family 2 subfamily C member 9; CYP2C19: Cytochrome P450 family 2 subfamily C member 19; INR: International normalized ratio; PCI: Percutaneous coronary intervention; P2Y12: Purinergic receptor P2Y12; VKORC1: Vitamin K epoxide reductase complex subunit 1.

has been associated with carbamazepine-induced hypersensitivity reactions in several populations.⁹⁴ Barriers include clinician unfamiliarity and lack of point-of-care testing, though commercial panels (e.g., GeneSight) are bridging this gap, as summarized in Table 4.⁹⁵

3.8.4. Infectious diseases: Precision treatment

HLA-B*57:01 testing prevents abacavir hypersensitivity in patients with human immunodeficiency virus with 100% efficacy.⁹⁶ In tuberculosis, NAT2 genotyping identifies slow acetylators at risk of isoniazid-induced hepatotoxicity, enabling dose adjustments that reduce liver injury by 70%.⁹⁷ Emerging tools such as whole-genome sequencing are personalizing antibiotic regimens for multidrug-resistant infections, as given in Table 5.

4. Case studies on personalized medicine and pharmacogenomics

The integration of PM and PGx has revolutionized lung cancer treatment by enabling biomarker-driven therapies tailored to individual tumor genetics. Molecular testing now allows clinicians to identify targetable mutations such as EGFR, ALK, and ROS1, guiding the selection of TKIs that significantly improve survival compared to traditional chemotherapy. However, challenges remain in accessibility, resistance mechanisms, and equitable implementation globally. The following case studies demonstrate how precision oncology transforms outcomes in advanced NSCLC, highlighting both the promise of targeted therapies and real-world complexities in molecular testing, treatment selection, and long-term management. These

Table 4. Pharmacogenomics in psychiatry

Drug	Pharmacogenomic biomarker	Clinical indication	Genotype-based recommendation	Clinical benefit	Clinical outcome
Sertraline ¹⁰	CYP2D6 ^a / CYP2C19 ^b	Major depressive disorder; anxiety disorders	CYP2C19 poor metabolizers require dose reduction or an alternative SSRI; ultrarapid metabolizers may require alternative therapy due to reduced exposure	Genotype-guided prescribing improves antidepressant selection and reduces trial-and-error prescribing	Improved treatment response, reduced adverse drug reactions, and better medication adherence
Carbamazepine ^{9a}	HLA-B*15:02	Epilepsy; bipolar disorder; and trigeminal neuralgia	Avoid carbamazepine in HLA-B*15:02-positive individuals	Pretreatment genetic screening markedly reduces the incidence of SJS and TEN	Prevention of life-threatening severe cutaneous adverse reactions
Carbamazepine ^{9a}	HLA-A*31:01	Epilepsy; bipolar disorder	Consider alternative therapy or close monitoring in HLA-A*31:01-positive patients	Genetic screening reduces the risk of carbamazepine-induced hypersensitivity reactions	Reduced incidence of drug-induced hypersensitivity syndrome, maculopapular eruptions, and DRESS

Notes: ^aCYP2D6 metabolizes approximately 20% to 25% of clinically used drugs; ^bCYP2C19 metabolizes approximately 10%. Both enzymes contribute to the metabolism of several antidepressants, including SSRIs. Abbreviations: CYP2C19; Cytochrome P450 family 2 subfamily C member 19; CYP2D6; Cytochrome P450 family 2 subfamily D member 6; DRESS: Drug reaction with eosinophilia and systemic symptoms; HLA: Human leukocyte antigen; SJS: Stevens–Johnson syndrome; SSRI: Selective serotonin reuptake inhibitor; TEN: Toxic epidermal necrolysis.

Table 5. Pharmacogenomics in infectious diseases

Disease	Pharmacogenomic biomarker	Clinical indication	Genotype-based recommendation	Clinical benefit	Clinical outcome
HIV ^{9c}	HLA-B*57:01	Initiation of abacavir-containing antiretroviral therapy	Screen all patients before initiating abacavir; avoid abacavir in HLA-B*57:01-positive individuals	Pretreatment genetic screening virtually eliminates abacavir-induced hypersensitivity reactions (≈100% prevention)	Improved patient safety and avoidance of potentially life-threatening hypersensitivity reactions
TB ^{9f}	NAT2	Isoniazid-based anti-tuberculosis therapy	Slow acetylators require dose adjustment or close liver function monitoring to minimize toxicity	NAT2 genotype-guided dosing reduced isoniazid-induced hepatotoxicity by approximately 70%	Improved treatment safety while maintaining therapeutic efficacy

Abbreviations: HIV: Human immunodeficiency virus; TB: Tuberculosis.

examples underscore the critical role of comprehensive genomic profiling in modern cancer care. The case studies presented in this section are representative examples adapted from previously published clinical studies and landmark clinical trials. They are included for illustrative and educational purposes to demonstrate the practical application of PGx in PM and routine clinical practice. These examples do not represent original patient cases or unpublished clinical data but are synthesized from the cited literature to highlight clinically relevant PGx principles.

4.1. Case study 1: *EGFR*-mutated non-small cell lung cancer treated with osimertinib⁹⁸

4.1.1. Patient profile

- Age/gender/smoking history: 58-year-old female, non-smoker.
- Diagnosis: Stage IV lung adenocarcinoma (NSCLC).
- Symptoms: Persistent cough, chest pain, and weight loss.

4.1.2. Molecular testing & diagnosis

- Biomarker analysis: NGS revealed an *EGFR* exon 19 deletion (a driver mutation).
- Programmed death-ligand 1 (PD-L1) status: Low expression (<1% by IHC), ruling out first-line immunotherapy.

4.1.3. Treatment plan

- First-line therapy: Osimertinib (a third-generation *EGFR* TKI) was selected due to its superior central nervous system (CNS) penetration and efficacy against T790M resistance mutations.
- Rationale: *EGFR* TKIs (e.g., gefitinib, erlotinib) show 80% response rates in *EGFR*-mutated NSCLC vs. 30% with chemotherapy. Osimertinib reduces the risk of CNS metastases by 50% compared to earlier TKIs.

4.1.4. Outcome

- Response: Partial tumor regression within eight weeks (RECIST criteria).
- Progression-free survival (PFS): 18 months (vs. 10 months with chemotherapy).
- Adverse effects: Mild rash and diarrhea (Grade 1), managed with dose adjustment.

4.1.5. Key takeaways

- Precision diagnosis: NGS identified a targetable mutation, avoiding ineffective chemotherapy.
- Superior outcomes: Osimertinib's CNS activity prevented brain metastases, a common failure site in NSCLC.
- Resistance monitoring: Liquid biopsy at progression detected *EGFR* C797S mutation, guiding next-line

trials with brigatinib + cetuximab.

4.2. Case study 2: *ALK*-rearranged non-small cell lung cancer treated with alectinib⁶⁰

4.2.1. Patient profile

- Age/gender/smoking history: 42-year-old male, light smoker (10 pack-years).
- Diagnosis: Stage IIIB *ALK*-positive NSCLC (adenocarcinoma).
- Symptoms: Dyspnea, hemoptysis.

4.2.2. Molecular testing & diagnosis

- Biomarker analysis: FISH confirmed *EML4::ALK* fusion (*ALK* rearrangement).
- PD-L1 status: Negative (0% expression).

4.2.3. Treatment plan

- First-line therapy: Alectinib (second-generation *ALK* inhibitor), chosen for its 75% intracranial response rate.
- Rationale: *ALK* inhibitors outperform chemotherapy (PFS: 34.8 months vs. 10.9 months).
- Alectinib's CNS penetration reduces brain metastasis risk by 85%.

4.2.4. Outcome

- Response: Complete metabolic response on positron emission tomography/computed tomography at six months.
- Progression: 36 months (ongoing).
- Adverse effects: Grade 2 fatigue (managed with lifestyle modifications).

4.2.5. Key takeaways

- Biomarker-driven selection: *ALK* testing identified a rare but highly actionable alteration.
- CNS protection: Alectinib's efficacy in preventing brain metastases improved long-term survival.
- Cost-effectiveness: Despite high drug costs, *ALK* inhibitors reduce hospitalizations and palliative care needs.

5. Summary

Personalized medicine and PGx have progressively shifted therapeutic decision-making from conventional population-based approaches toward patient-specific strategies informed by genetic, molecular, and clinical characteristics. Advances in genomics, biomarker analysis, and computational approaches have expanded their applications across oncology, cardiovascular medicine, psychiatry, and infectious diseases. Genotype-informed approaches, including trastuzumab for HER2-positive breast cancer, osimertinib for *EGFR*-mutated lung cancer, *CYP2C9*- or *VKORC1*-guided warfarin therapy, *CYP2D6*-

and *CYP2C19*-informed psychopharmacology, and *HLA-B*57:01* screening before abacavir therapy, demonstrate the clinical potential of individualized treatment strategies. These applications support more appropriate drug selection, dose optimization, and identification of patients at increased risk of treatment-related toxicity. However, broader clinical implementation remains dependent on robust evidence for clinical utility, appropriate testing and data infrastructure, standardized interpretation and clinical guidance, and equitable access to precision healthcare. Continued integration of genomic information with other molecular and clinical data, together with advances in analytical and computational technologies, may further expand the role of PM and PGx in routine healthcare and facilitate more individualized approaches to disease prevention, diagnosis, and treatment.

6. Clinical implementation of pharmacogenomics

6.1. Clinical practice guidelines: Clinical Pharmacogenetics Implementation Consortium and Dutch Pharmacogenetics Working Group

The successful integration of PGx into routine clinical practice relies on standardized, evidence-based guidelines that assist healthcare professionals in translating genetic information into therapeutic decisions. Two internationally recognized organizations, the Clinical Pharmacogenetics Implementation Consortium (CPIC) and the Dutch Pharmacogenetics Working Group (DPWG), have developed comprehensive recommendations for genotype-guided drug prescribing. CPIC provides peer-reviewed guidelines that help clinicians understand how available genetic test results should be used to optimize drug selection and dose without making recommendations regarding whether genetic testing should be performed. Similarly, DPWG develops evidence-based recommendations that classify gene–drug interactions according to clinical significance and provide practical guidance on dose modification, alternative therapies, and monitoring strategies. Both organizations regularly update their recommendations as new evidence emerges and have established guidelines for numerous clinically actionable gene–drug pairs, including *CYP2C19*–clopidogrel, *CYP2D6*–codeine, *TPMT/NUDT15*–thiopurines, *DPYD*–fluoropyrimidines, *UGT1A1*–irinotecan, and *SLCO1B1*–simvastatin. The widespread adoption of CPIC and DPWG guidelines has improved prescribing accuracy, minimized adverse drug reactions, and enhanced therapeutic efficacy, making them indispensable tools for implementing precision medicine in clinical practice.⁹⁹

6.2. PharmGKB and pharmacogenomic knowledge resources

The increasing volume of PGx information has necessitated the development of comprehensive knowledge bases that facilitate the interpretation and clinical application of genetic data. The Pharmacogenomic Knowledgebase (PharmGKB) is one of the most widely used public repositories that curates PGx information from scientific literature, clinical studies, regulatory agencies, and expert guidelines.¹⁰⁰ PharmGKB provides detailed annotations on gene–drug interactions, genotype-based dosing recommendations, PGx pathways, variant annotations, drug labels, and links to CPIC and DPWG guidelines, thereby serving as an important decision-support resource for clinicians, researchers, and regulatory agencies. In addition to PharmGKB, databases such as the FDA Table of Pharmacogenetic Associations, PharmVar, and ClinVar provide valuable information regarding clinically significant genetic variants and standardized allele nomenclature. These resources collectively facilitate evidence-based prescribing, improve interpretation of PGx test results, and promote the consistent implementation of PM across healthcare systems.^{101,102}

6.3. Preemptive versus reactive pharmacogenomic testing

Pharmacogenomic testing can be implemented using either a reactive or preemptive approach. Reactive testing is performed after a medication has been selected or when a patient experiences therapeutic failure or adverse drug reactions, allowing treatment decisions to be modified based on the patient's genotype. Although useful, this strategy may delay treatment while awaiting genetic test results. In contrast, preemptive PGx testing involves analyzing multiple clinically relevant pharmacogenes before drug therapy is initiated, with results stored in the patient's EHRs for future use.¹¹ This proactive approach enables immediate genotype-guided prescribing whenever medications are prescribed throughout a patient's lifetime. Large implementation programs such as the Mayo Clinic RIGHT Study, Vanderbilt PREDICT Program, and the eMERGE Network have demonstrated the clinical feasibility of preemptive testing and reported improvements in medication safety, prescribing efficiency, and prevention of adverse drug reactions. As the cost of genomic sequencing continues to decline, preemptive PGx testing is expected to become an integral component of routine precision healthcare.¹⁶

6.4. Clinical decision support systems

The successful implementation of PGx in clinical practice depends not only on the availability of genetic information

but also on effective mechanisms for translating this information into actionable prescribing decisions. CDS systems integrated with EHRs have emerged as essential tools for delivering real-time PGx recommendations during prescribing. These systems automatically identify clinically relevant genetic variants and generate alerts when a prescribed medication may require dose adjustment, additional monitoring, or substitution with an alternative therapy according to CPIC or DPWG recommendations.¹⁰³ For example, CDS systems can recommend alternative antiplatelet therapy for patients carrying CYP2C19 loss-of-function alleles prescribed clopidogrel or suggest reduced doses of fluoropyrimidines in patients with DPYD deficiency. Several healthcare institutions, including Vanderbilt University Medical Center, Mayo Clinic, and the eMERGE Network, have successfully incorporated PGx-based CDS into clinical workflows, demonstrating improvements in prescribing accuracy, patient safety, and clinician acceptance. Continued advances in AI, interoperability, and standardized genomic data integration are expected to further enhance the effectiveness of CDS systems in supporting precision medicine.¹⁰⁴

7. Future prospects

The future of PM will be driven by advances in genomic technologies, AI, and multi-omics integration that enhance disease prediction, diagnosis, and individualized therapeutic decision-making. Continued reductions in the cost of genomic sequencing, together with improvements in analytical accuracy and accessibility, are expected to facilitate broader implementation of PM across diverse healthcare settings. The development of PRS may further improve risk prediction for complex diseases, enabling earlier preventive interventions and more personalized disease management strategies. In addition, the integration of EHRs, wearable devices, and other real-world clinical data with AI-based analytical platforms is expected to support more dynamic and evidence-based clinical decision-making.¹⁰⁵

Emerging therapeutic approaches, including CRISPR-based gene editing, mRNA therapeutics, and digital twin technologies, have the potential to expand precision treatment beyond conventional pharmacotherapy by enabling individualized disease modeling and targeted interventions. Furthermore, the integration of multi-omics datasets through systems biology, AI, and ML is anticipated to provide comprehensive patient-specific molecular profiles, thereby improving disease prediction, therapeutic optimization, and clinical decision support beyond the capabilities of genomics alone.¹⁰⁶

The successful future implementation of PM will also depend on the establishment of diverse global genomic databases and continued interdisciplinary collaboration among researchers, clinicians, policymakers, and patients to promote equitable and evidence-based healthcare. Continued technological innovation and international collaboration are expected to accelerate the translation of precision medicine into routine clinical practice while ensuring that its benefits are accessible across different populations.

8. Conclusion

Personalized medicine represents a paradigm shift from the conventional “one-size-fits-all” approach toward individualized healthcare by integrating genetic, molecular, clinical, and environmental information to optimize disease prevention, diagnosis, and treatment. Among its core components, PGx has emerged as one of the most clinically advanced components of personalized medicine, enabling genotype-guided therapeutic decisions that improve drug efficacy, reduce adverse drug reactions, and enhance patient safety. Its successful implementation across oncology, cardiovascular medicine, psychiatry, and infectious diseases underscores its growing role in evidence-based, patient-centered healthcare. Despite these advances, several scientific, technical, economic, and ethical challenges continue to hinder widespread adoption, including limited access to genomic testing, underrepresentation of diverse populations in genomic databases, high implementation costs, fragmented healthcare systems, and concerns about data privacy and equitable access. Overcoming these challenges will require standardized clinical guidelines, robust regulatory frameworks, interdisciplinary collaboration, and sustained investment in genomic infrastructure, workforce training, and implementation research. Importantly, the future of PM extends beyond PGx alone. The integration of multi-omics technologies—including transcriptomics, proteomics, metabolomics, microbiomics, and epigenomics—with AI, ML, systems biology, and digital health platforms will enable a more comprehensive understanding of individual biological variability. Emerging innovations such as PRS, digital twins, and CRISPR-based genome editing further expand opportunities for predictive and precision healthcare. As these technologies mature and become more accessible, PM is poised to transform routine clinical practice by delivering safer, more effective, and truly individualized therapeutic strategies, ultimately improving health outcomes and advancing equitable, patient-centered healthcare worldwide.

List of abbreviations

Abbreviation	Full term
AI	Artificial intelligence
ALK	Anaplastic lymphoma kinase
BRCA	Breast cancer susceptibility gene
CNS	Central nervous system
CYP2C9	Cytochrome P450 2C9
CYP2D6	Cytochrome P450 2D6
DPYD	Dihydropyrimidine dehydrogenase
EGFR	Epidermal growth factor receptor
EHR	Electronic health record
EMA	European Medicines Agency
FDA	Food and Drug Administration
FISH	Fluorescence <i>in situ</i> hybridization
HER2	Human epidermal growth factor receptor 2
IHC	Immunohistochemistry
NGS	Next-generation sequencing
PARP	Poly(ADP-ribose) polymerase
PFS	Progression-free survival
PGx	Pharmacogenomics
PM	Personalized medicine
PRS	Polygenic risk score
RECIST	Response Evaluation Criteria in Solid Tumors
TKI	Tyrosine kinase inhibitor
VKORC1	Vitamin K epoxide reductase complex subunit 1

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