

Review Article

Pharmacogenomics Applications in Clinical Practice: Revolutionizing Patient Care

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Abstract:

Background: Personalised medicine through pharmacogenomics is revolutionizing healthcare delivery by encouraging individualized therapy that takes into consideration an individual's genetic profile, environment and lifestyle. Pharmacogenomics is an aspect of pharmacy that studies the relationship between genetic profile and response to therapeutic agents. However, the application of the concepts of pharmacogenomics in healthcare helps in achieving more effective and safe responses from therapy. This study evaluates the application and benefits of pharmacogenomics in clinical practice based on evidence from current practices in various medical fields.

Methods: In carrying out this review, PubMed database was the primary literature source and we analyzed and synthesized findings from the included literature thematically as it relates to pharmacogenomics applications, benefits and challenges as well as safety and ethical concerns.

Results: Pharmacogenomics has been widely applied in various aspects of healthcare such as in dosing, choice of treatment, reducing and management of adverse reactions, individualization of therapy, optimizing efficacy of therapy. Despite its numerous applications, its adoption faces challenges such as limited clinical evidence, lack of specialized training among healthcare professionals, cost and complexity of genetic mapping as well as ethical concerns.

Conclusion: With ongoing advances in genomic technologies, pharmacogenomics is becoming an integral aspect of individualization therapy in clinical practice and more widely applied in different healthcare sectors.

Keywords: Pharmacogenomic; Clinical practice; Personalized medicine; Gene testing; Precision medicine.

Introduction

Personalised medicine is an evolutionary approach in healthcare that takes into account an individual's genetic makeup, lifestyle, and environment in tackling healthcare related needs of patients such as therapeutic management, optimizing therapeutic outcomes as well as improving or maintaining quality of life [1]. On the other hand, pharmacogenomics, which is the cornerstone of personalized medicine, has to do with the study of an individual's response to treatment regimen in relation to the person's genetic profile. It features studies on drug interaction, gene structure and function at a molecular level for application in drug development [2].

Understanding how genetic factors influence drug absorption, distribution, metabolism, and excretion enables healthcare professionals to determine individuals who may benefit from a medication and those who may experience Adverse Drug Reactions (ADRs). Patients exhibit variable response to prescribed medication, this highlights the significance of personalized medicine in healthcare. For example, some drugs are highly effective in certain patients, but others do not experience the same effectiveness. Instead, they are faced with debilitating ADRs from the same drugs. These variations in responses majorly arise from significant differences in genetic makeup of individuals [3].

In the past decade, several studies on pharmacogenomics have uncovered the genetic variations that influence differences in drug response. Advances in genomic technology such as next-generation sequencing and microarray-based comparative genomic hybridization have played a key role in enabling the Integration of pharmacogenomics

into medical practice by making genetic testing more accessible and affordable [4]

Moreover, platforms like the Clinical Pharmacogenetics Implementation Consortium (CPIC) and the Pharmacogenomics Knowledge Base (PharmGKB) have encouraged the clinical practice of pharmacogenomics by providing guidelines that allow clinicians to apply pharmacogenomics in healthcare settings [5]. However, several challenging factors limit the full implementation of pharmacogenomics in clinical practice some of which include insufficient clinical evidence, inadequate education of healthcare providers in pharmacogenomics, as well as lack of equipment and infrastructure for genomic testing and data storage. Nevertheless, the application of pharmacogenomics in personalized medicine not only holds the potential for more effective treatment, but promises to minimize ADRs encountered by patients leading to cost-effective healthcare [6].

Nevertheless, in carrying out this review, it was hypothesized that the application of pharmacogenomics in clinical practice will significantly improve patient outcomes. However, this review aims to synthesize evidence supporting the applications of pharmacogenomics in clinical practice, as well as explore the benefits of such application and challenges hindering application.

Ultimately, this paper is essential for healthcare professionals, as it provides a comprehensive overview of applications of pharmacogenomics in clinical practice and its potential to improve clinical outcomes and guide decision-making. By addressing the barriers to its implementation, the study aims to guide clinicians in adopting pharmacogenomics.

The Science of Pharmacogenomics

Pharmacogenomic studies are based on the understanding of the basic principles of drug responses in a biological system, which is essentially the pharmacokinetics and pharmacodynamics of the drug. The combination of this understanding with the knowledge of genetic differences that lead to varying phenotypes provides insight on what drugs will be safe and effective for certain kinds of people and at what dose [7]. Pharmacokinetics describes the body's interaction with administered drugs, encompassing factors such as absorption, distribution, metabolism, and excretion, while pharmacodynamics describes factors that affect the drug's action on the target protein,

including receptors, intracellular enzymes, and cell membrane channels [8].

The genetic involvement in the pharmacokinetics and pharmacodynamics can be explained with the evaluation of one of the most studied families of enzymes driving drug metabolism (cytochrome P450). Cytochrome P450 genes such as CYP2C9, CYP2C19, and CYP2D6 can be produced from varying DNA nitrogenous base sequences. Varying cytochrome P450 enzymes with differences in amounts, degree of enzymatic activity, and therefore varying abilities to metabolize drugs are subsequently produced from these gene variants. As a consequence,

individuals with certain variants of cytochrome P450 enzymes may metabolize drugs more slowly or more rapidly, impacting efficacy and the risk of side effects [9, 10, 11].

Furthermore, the majority of the genes coding for ATP-binding cassette transporters, which facilitate movement of several drugs through cells, exist in many different versions (alleles) within populations, contributing to variations in how well the proteins transport drugs and hence biological responses, which may be to increasing or decreasing degrees [12] [13]. These variations impact the effectiveness of the drugs and the risk of serious adverse events.

The impact of genetic variations on drug response is evident in the diverse areas of therapy. In oncology, the efficacy and safety of thiopurine drugs used in the treatment of leukaemia are influenced by the genetic variation of the thiopurine S-

methyltransferase (TPMT) gene encoding for the TPMT enzyme required for the metabolism of thiopurine drugs [14, 15]. These variations may lead to adverse drug reactions in certain patients, necessitating dose adjustments based on genetic testing. In cardiology, the effectiveness of clopidogrel in patients with acute myocardial events is impacted by its conversion to an active metabolite by cytochrome P450 2C19 (CYP2C19) enzyme. The presence of two copies of the CYP2C19 gene in an individual leads to poor enzymatic activity and an inability to benefit from the antiplatelet function of the drug [16]. However, in psychiatry, genetic polymorphisms of CYP2C19 and CYP2D6 enzymes used in phase 1 metabolism of most antidepressants and antipsychotics, and genetic variations in neurotransmitter receptors contribute to suboptimal drug exposure and minimal tolerability of these drugs, necessitating prescriber reliance on pharmacogenomics drug label recommendations [17, 18].

Application of Pharmacogenomics in Clinical Practice

Incorporation of pharmacogenomics in clinical practice has significantly transformed disease treatment and prevention in diverse therapeutic areas. In cardiology, clopidogrel has been widely used as antiplatelet, however, considerable concern has been raised due to the significant variability in response to the P2Y₁₂ receptor antagonist, largely dependent on the presence of the CYP2C19 gene variant [19]. According to Cavallari & Pratt, (2022) [20], CYP2C19 testing in line with pharmacogenomics concept can help predict patients unlikely to have maximum benefit from clopidogrel in whom alternative therapy may be initiated [20].

Additionally, understanding which Vitamin K Epoxide Reductase Complex subunit 1 gene (VKORC1) and CYP2C9 alleles are possessed by a patient informs personalized warfarin dosing during the stabilization phase of anticoagulant therapy [21]. In oncology, personalized cancer therapy can be achieved through genetic profiling of tumors, enabling the selection of targeted therapies that are more effective and have fewer side effects [22]. Classification of human epidermal growth factor receptor 2 (HER2) protein expressions through pharmacogenomics has informed targeted therapy using Trastuzumab, a monoclonal antibody in the treatment of breast cancers [23].

In neuropsychiatry, personalized treatment using different antidepressants and antipsychotics can be achieved by the application of pharmacogenomics in the classification of patients as poor metabolizers, intermediate metabolizers, normal metabolizers, and rapid or ultrarapid metabolizers based on the expression of cytochrome P450 variants responsible for metabolism, leading to the selection of therapy with maximum efficacy and minimum toxicity [24]. Moreover epileptic medications such as carbamazepine require Human Leukocyte Antigen B1502 (HLA-B1502) testing to prevent life-threatening skin diseases like Stevens-Johnson syndrome in positive individuals [25].

In infectious diseases therapy, pharmacogenomics guides the selection of antiviral and antimicrobial therapy by determining genetic variants related to disease susceptibility and response to antimicrobial therapy [26]. For example, in the management of HIV, Human Leukocyte Antigen B5701 (HLA-B5701) allele testing is conducted to determine the presence of HLA-B*57:01 before administering Abacavir to patients so as to prevent hypersensitivity reactions in positive alleles [27].

Table 1 summarizes various drugs and the relevant genetic tests that guide their application in clinical practice in line with pharmacogenomic principles.

Table 1: Some genetic tests required in the application of pharmacogenomics in clinical practice

Condition	Drug	Gene Tested	Impact of Gene
HIV Management	Abacavir	HLA-B5701	High risk of hypersensitivity reaction in positive allele
Epilepsy/Neuropathic pain	Carbamazepine	HLA-B1502	Increased risk of severe skin reactions in certain populations
Antiplatelet Therapy	Clopidogrel	HLA-B1502	Variants affect drug activation and risk of stent thrombosis
Breast Cancer	Trastuzumab/T-DM1	HER2/NEU	Ensures efficacy for HER2-positive breast cancer patients
Breast Cancer	Tamoxifen	CYP2D6	Poor metabolizers may need alternative therapies or doses
Pain Relief	Codeine	CYP2D6	Poor or ultra-rapid metabolizers affect drug efficacy
Anticoagulant	Warfarin	CYP2C9, VKORC1	Guides dose adjustment to prevent bleeding or clotting
Chemotherapy	Irinotecan	UGT1A1 (UDP Glucuronosyltransferase 1A1)	Influences dosing to avoid severe side effects
Immunosuppression	Azathioprine/6-mercaptopurine	TPMT	Increases risk of severe toxicity
Colorectal Cancer	Cetuximab/Panitumumab	KRAS (Kirsten Rat Sarcoma Viral Oncogene Homolog)	Mutation status determines drug efficacy in colorectal cancer

Benefits and Challenges

Pharmacogenomics holds benefits capable of transforming healthcare approaches and enhancing treatment outcomes. However, the application of pharmacogenomics is accompanied by a number of challenges.

Potential benefits of pharmacogenomics in healthcare

I. Reduced Adverse Drug Reactions

One of the most significant benefits of pharmacogenomics is that it reduces incidences of adverse drug reactions (ADRs). ADRs have diverse implications for the health of individuals worldwide, some of which may be life threatening, largely resulting from a one-drug-fits-all approach or polypharmacy. Diverse genes influence an individual's response (negative or positive) to a drug. By understanding the genetic variants possessed by a patient and how they influence drug response, treatment can be initiated with interventions that do not elicit negative responses [28].

This impacts on health outcomes by ensuring that drugs are safe and only positive responses are obtained from a drug.

For example, Pharmacogenomic testing for abacavir and carbamazepine has prevented significant adverse drug reactions (ADRs) that could have been life threatening. Genetic testing before initiating carbamazepine and abacavir therapies helps to identify at-risk patients, allowing clinicians to prescribe safer options and avert fatal side effects. The extensive use of pharmacogenomic testing for these medications has reduced ADR hospitalizations, healthcare costs, and promoted patient safety and outcomes from therapy.

II. More Efficacious Drugs

Pharmacogenomics impacts informed research and drug development tailored to specific genes associated with effective responses and their encoded proteins, enzymes, and RNA molecules. Discovery of therapy can then be supported by findings from genetic testing, leading to the development of drugs that target specific diseases [29]. With this approach pharmaceutical

companies are able to synthesize therapeutic substances that are more potent.

For example, Pharmacogenomic testing for trastuzumab, T-DM1, and irinotecan has improved cancer treatment outcomes by ensuring patients receive the most effective medications. Trastuzumab and T-DM1 are targeted therapy for HER2-positive breast cancer (with overexpressed Human Epidermal Growth Factor Receptor 2 (HER2/NEU) gene). Testing for HER2/NEU expression helps to evaluate if these medications are right for a patient, ensuring that only those who will benefit from it receive the therapy. This targeted and personalized treatment approach helps to improve survival rates and reduce healthcare costs [30, 31].

III. Increased Accuracy in Dosage Determination

Pharmacogenomics provides a more accurate method of dosage determination. Personalising drug dosage based on the individual's unique genetic profile instead of age or weight will ensure more accuracy [32, 33]. This will ensure that maximum value is obtained from the therapy and reduce the likelihood of underdose or overdose.

For example, CYP2C9 and VKORC1 genetic variants alter warfarin metabolism, affecting efficacy and risk of bleeding or clotting. Before administering warfarin, doctors can carry out genetic testing to evaluate the patient's genetic profile to ensure proper dosage. Haemorrhages, which require expensive medical procedures, blood transfusions, and longer hospital stays, can be avoided with precise dosing. Therefore, pharmacogenomic testing helps to reduce healthcare costs by eliminating the trial-and-error approach to warfarin dosing [34, 35].

IV. Overall Reduction in Cost of Healthcare

therapy in other therapeutic areas [37]. This lack of strong clinical evidence makes it difficult for pharmacogenomics to be applied in routine healthcare.

II. Cost and complexity of genetic testing

The cost and complexity of genetic testing significantly impact the seamless adoption of pharmacogenomics. Though the cost of genomic sequencing has reduced considerably in high-income countries, low- and middle-income countries still face severe financial constraints limiting the adoption of pharmacogenomics in these areas [38]. Moreover, genetic testing is a complex process, and the results may

Applying pharmacogenomics potentially leads to a reduction in the financial burden of healthcare by decreasing the occurrence of adverse drug reactions, minimizing patients' duration of therapy, decreasing the number of medications that have to be prescribed in the trial-and-error method, and increasing the number of effective drugs produced by pharmaceutical companies [36]. Figure 1 below summarizes the benefits of applying pharmacogenomics in healthcare.

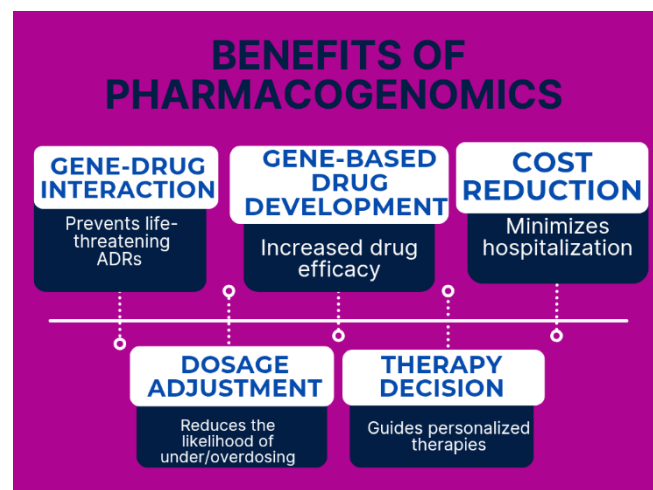


Figure 1: Benefits of Pharmacogenomics in Clinical Practice

Challenges and limitations of implementing pharmacogenomics

The challenges that have to be overcome before pharmacogenomics can be fully adopted include:

I. Minimum evidence from large-scale studies

Though data from clinical studies exist which serve as evidence to support the application of pharmacogenomics in some areas of therapy such as psychiatry and cardiology, there is limited research data to support its application of

not be available until after several hours or even days, after which the patient would have to be contacted before therapy is initiated or not [38].

III. Policy regulation and ethical issues

The implementation of pharmacogenomics requires regulatory guidelines that define the use of genetic profile data. Genetic information is highly confidential; therefore, large-scale collection of genetic information and clinical data raises privacy issues since this information can be put to the wrong use. Policy that establishes ethical use of genetic data is largely lacking [39].

IV. Inadequate training of healthcare professionals

Inadequate training and education of healthcare professionals in pharmacogenomics has become a hindrance to its execution, especially in low- and middle-income countries [40]. Low- and middle-

income areas like Africa are lacking in healthcare professionals, most of whom lack specialty training, making education in pharmacogenomics even more challenging.

Ethical and Societal Implications

Ethical Considerations in Pharmacogenomics

The use of genetic information in healthcare is accompanied by several ethical concerns. One of these concerns is the social discrimination that may be encountered by some people due to their genetic predisposition [41]. Though developed countries like the United States have instituted a legislative framework like the Genetic Information Nondiscrimination Act (GINA) to protect individuals from such discrimination, many other countries do not have such established legislation [42]. Another concern is the need for privacy and confidentiality. Genetic information is deeply personal and may be accessed by unauthorized individuals if not properly protected, leading to misuse [40]. A major ethical issue is informed consent. Before genetic testing is conducted, individuals should be made to fully understand what they are agreeing to, including the nature of data being collected, how it would be used, and the possible implications, including incidental findings like susceptibility to unrelated genetic conditions [43]. Finally, data protection is an issue as digitalization of healthcare may increase unauthorized access to genetic information leading to identity theft and misuse [41].

Potential implications for patients, healthcare professionals, and society

Pharmacogenomics is a revolutionary approach that has a wide range of implications for individuals at different levels. For patients, it promises to offer more effective and safer treatment strategies to ensure improved health outcomes. However, despite the benefits of personalized medicine through pharmacogenomics, the gaps in access to genomic testing due to limited access in developing regions may lead to unequal health outcomes [44].

For healthcare professionals, pharmacogenomics is a new area that is gradually being incorporated into healthcare and requires new skills and competencies. As personalized medicine is becoming the new order of practice, ongoing education and training is necessary for health care professionals to accurately apply genomic testing results to clinical decision-making processes and guide patients accordingly [45].

Universal adoption of pharmacogenomics at the societal level has the potential to transform healthcare delivery, public health intervention and monitoring, as well as drug development [46]. Governing bodies in different regions will also be required to institute policies that ensure adequate data protection and security.

Implementation and Integration

Strategies for Implementing Pharmacogenomics in Clinical Practice

Successfully implementing pharmacogenomics requires a multiple-solution strategy. One of the key strategies is education and training of healthcare professionals [47]. This would enable frontline medical practitioners to understand genetic information and utilize genomic data in clinical decision-making and prescribing; hence, educational institutions should integrate pharmacogenomics into medical curricula to prepare clinicians for its application in practice.

Also, integrating pharmacogenomic testing into Electronic Health Records, along with other smart clinical support tools, can enable clinicians to rapidly and effectively use genetic information in the treatment of patients at the point of care. This strategy is already being implemented by a number of institutions and has demonstrated improved efficiency and clinical outcomes [48].

Another key strategy is developing clear, evidence-based clinical guidelines to enable clinicians to understand and use patients' genetic test results. These guidelines, such as those provided by CPIC and

the Dutch Pharmacogenetics Working Group, would transform complex genetic test results into practical steps guiding clinicians when treating patients [59].

Integrating Pharmacogenomics into Healthcare Systems

For healthcare systems to successfully offer pharmacogenomic services, key infrastructure and policies must be adopted. Healthcare systems must have access to well-equipped laboratories capable of carrying out genetic testing, and establish data-sharing infrastructure like functional computational systems to ensure that different departments of the system, such as laboratory, pharmacy, and clinics, freely access, share, and use genetic data smoothly.

Additionally, policies which ensure genetic tests used are accurate, reliable, and approved by relevant bodies (e.g., Food and Drug Administration (FDA) or local health regulatory bodies), patients' privacy and data protection, as well as ethical

guidelines which prevent genetic discrimination and ensure patients give informed consent, must be adopted. Sustainable funding systems are also necessary for the smooth running of genetic testing and pharmacogenomic technologies within healthcare systems.

Collaborations between government (e.g., health ministries or national research institutes) and private groups (e.g., biotech companies, pharmaceutical firms, or healthcare providers) would facilitate the development and improvement of pharmacogenomics tools and also promote the widespread use of these technologies within healthcare institutions.

Furthermore, actively involving patients and the general public through education to enlighten individuals on the implications of pharmacogenomics for their health outcomes, together with advocacy for the use of pharmacogenomics, will help build confidence and enhance acceptance.

Current Status and Future Prospects

Current state of pharmacogenomics and personalized medicine

Advances in pharmacogenomics have increased its recognition as a standard component of contemporary health care. The extensive integration of pharmacogenomics information into the US Food and Drug Administration (FDA)-approved drug labels for over 300 drugs shows that several pharmacogenomics tests identifying drug-gene pairs have been approved by the regulatory body and have been widely accepted in mainstream healthcare [50]. Also, in a bid to strengthen the integration of pharmacogenomics in routine clinical practice and enable clinicians interpret pharmacogenomics data in clinical practice, freely accessible gene/drug regulatory guidelines have been developed by institutions like the Clinical Pharmacogenetics Implementation Consortium (CPIC) [51].

Application of pharmacogenomics in personalised medicine has commenced in major areas of clinical practice like oncology (where targeted cancer treatment is guided by pharmacogenomics testing for HER2, Kirsten rat sarcoma viral oncogene homolog (KRAS), and Epidermal Growth Factor Receptor (EGFR) mutations) [52, 53], psychiatry and cardiology. Additionally, preemptive pharmacogenomics testing is routinely conducted in some healthcare institutions,

which involves testing patients for a collection of genes related to drug response and storing the results in their electronic health records. These records are used to predict drug response and alert clinicians when a prescribed drug is unsuitable for a patient [54].

Emerging trends and future directions

One of the latest trends which has emerged in the pharmacogenomics field is the utilisation of artificial intelligence and machine learning in pharmacogenomics studies to aid the analysis of large-scale genomic data, thereby improving the prediction of drug response and treatment outcomes in clinical settings [55]. Pharmacogenomics have been integrated with other powerful biological study approaches like proteomics, metabolomics, and transcriptomics. These omics technologies offer a more comprehensive understanding of the reasons for variation in drug responses among individuals. Combining this data with pharmacogenomics holds promise to enhance drug response prediction in personalised medicine [56].

Also, emerging compact and easy-to-use technological devices like Genedrive® System, ID Now™, and Spartan RX CYP2C19, capable of analyzing specific genes within an hour, and other advances in point-of-care technologies are capable of shifting pharmacogenomics from specialized lab environments to the bedside [57, 58].

Future directions necessary for improved adoption of pharmacogenomics in clinical practice include:

- More robust integration of pharmacogenomics in routine clinical practice.
- Expansion of research to include more gene candidates that are involved in drug response regarding other diseases aside from cardiovascular disease, mental health disorders, and cancer.
- Conducting preemptive testing before illnesses emerge or before treatments commence to
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ensure that drug choices are immediately available when needed.

- Establishing the use of machine learning and deep learning for the analysis of large genomic data sets to realize more accurate prediction of drug response.
- Developing standardized regulatory guidelines and insurance policies to encourage the adoption of pharmacogenomics in clinical practice.

Conclusion

Evidently, personalised medicine piloted by pharmacogenomics signifies the emergence of a new era in healthcare. Pharmacogenomics facilitates individualisation of drug therapy by harnessing patients' genetic profiles, leading to enhanced efficacy, reduced adverse drug reactions, and improved cost-effectiveness. Though pharmacogenomics comes with numerous benefits for the general population, a number of challenges still hinder its widespread use and implementation in routine clinical practice, including inadequate clinical evidence, high cost of implementation, as well as ethical considerations. To enable the successful integration of pharmacogenomics, these challenges must be strategically addressed.

Judging from the ongoing progress and development in areas such as research, technology, and policy development in the field of pharmacogenomics, it is evident that the future holds great promise of more personalized drug therapy which is safe and effective, providing better health outcomes. As healthcare moves towards personalized medicine, collaboration among clinicians, researchers, policymakers, and patients will be essential to realize its full potential.

Pharmacogenomics truly exemplifies the shift toward precision medicine, where treatments are designed for individuals based on their genetic profile, leading to better health outcomes for patients and a more efficient healthcare system.

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