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Bridging Pharmacogenomics and Medical Genetics Education to Prepare Students for Personalized Medicine

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ABSTRACT

The rise of personalized medicine, driven by advances in human genome sequencing and pharmacogenomics, necessitates integration of genomic and drug-response knowledge into medical education. Pharmacogenomics explores how genetic variation affects drug metabolism, efficacy, and toxicity, offering a foundation for individualized treatment strategies. Despite growing clinical relevance, education in pharmacogenomics remains limited in medical curricula. Integrating pharmacogenomics with core medical genetics education can bridge this gap, equipping future clinicians with competencies in genetic variation interpretation, genomic technologies, and ethical, legal, and social considerations. Approaches such as competency-based education and interprofessional case-based learning provide effective frameworks for preparing students for the era of precision medicine. Addressing curricular gaps ensures that medical professionals can leverage genomic insights for safer, more effective, and patient-centered therapies.

Keywords: *personalized medicine, pharmacogenomics, medical genetics, genomic education, drug-gene interactions, genetic variation, precision medicine, curriculum integration, ethical, legal, and social implications, competency-based education, case-based learning.*

INTRODUCTION

The dawn of personalized medicine emerged with the sequencing of the human genome and the identification of a large number of single nucleotide polymorphisms (SNPs) in human DNA. It was propelled by the Human Genome Project, the HapMap project, and the completion of the sequencing of the

first human genome. The globalization of the distribution of human genome sequencing and the population candidate variants accessible on the Internet have accelerated it further. The application of DNA technologies became feasible in disease diagnosis, treatment, and drug development. Personalized medicine makes use of the variations in genes, drugs, proteins, and combinations thereof to tailor diagnosis, therapy, and drug development based on the individual patient or population for optimization of efficacy and toxicity. These breakthroughs have recently stimulated the formation of a high-profile professional organization called the International Society for Personalized Medicine, which held its first annual meeting in 2009 and broad knowledge-based network. The terms "personalized medicine," "individualized medicine," "targeted therapy," and "stratified medicine" are widely used in the preclinic and clinic, yet still lack universally accepted definitions. "Personalized medicine" and "precision medicine," the preferred term by the National Institutes of Health and the US government, denote a diagnosis and treatment tailored to the individual patient, whereas "targeted therapy" generally refers to the development of dug candidate-synonymous drug-gene targets (Basyouni & Shatnawi, 2020).

The Emergence of Personalized Medicine

Personalized medicine is an ongoing effort in medicine and pharmacology to identify new ways to determine the presence of disease in patients, or how individuals respond to drugs, based upon a variety of individual measurements. Such measurements might include genetic, epigenetic, environmental, or expression based parameters. Pharmacogenomics is a field developed as part of the overall quest to understand these factors and mitigate errors in drug prescription (L. Kudron et al., 2023). Enormous advancements in sequencing and other technologies have made pharmacogenomic analysis an integral element of personalized medicine and its incorporation into pharmacology.

The FDA first initiated the process of regulating pharmacogenomics in 1999 as an effort to incorporate genetic variation into drug development. Since then, the U.S. government has made additional efforts to stimulate pharmacogenomic research (S Boguski et al., 2013). The National Institutes of Health initiated a sizeable program to create an atlas of human variation data that included some early pharmacogenomic efforts. The National Center for Human Genome Research published its pharmacogenomic strategy and criteria for prioritizing gene systems in Phase I of the Human Genome Project. Two other national governmental programs to apply the Human Genome Project and increase the efficacy and safety of prescription drugs disclosed the involvement of pharmacogenomics. The FDA subsequently issued key documents concerning approaches to drug discovery and pharmacogenomic data rules.

Core Concepts in Pharmacogenomics and Medical Genetics

Pharmacogenomics studies how genetic variation influences drug response, including toxicity and therapeutic failure (C. Nutter & Gálvez-Peralta, 2018). It has evolved from initial discoveries of drug-induced adverse reactions to integration into educational programmes for health professionals. The American Board of Colleges of Pharmacy and the American Society of Health-System Pharmacists have incorporated pharmacogenomics into their standards and curricular guidance. Whereas few programmes included pharmacogenomics before 2000, by 2010 the majority of U.S. pharmacy schools offered independent pharmacogenomics education, often as an elective. Activities to foster discussion, such as debates on personalized medicine, are beginning to bridge gaps between pharmacogenomics knowledge and practice (W. Guy et al., 2020) [table 1].

Table 1: Core Concepts in Pharmacogenomics and Medical Genetics

Concept	Details	Relevance / Examples
Genetic Variation & Drug Response	Polymorphisms in drug-metabolizing enzymes, transporters, receptors; >800 genes associated with >200 drugs; >250 drug-gene pairs with guidelines	Explains inter-individual differences in drug efficacy and toxicity; foundation of pharmacogenomics
Genomic Technologies	WGS, WES, targeted gene panels, RNA-seq	Enables detection of SNVs, insertions, deletions, structural variants; key for personalized treatment decisions
Data Interpretation	QC, alignment, realignment, variant calling; variant classification (5-tier ACMG system); interpretation challenges	Essential for accurate clinical decision-making; handling variants of uncertain significance (VUS)
Pharmacogenomics Applications	Optimized drug dosing, prediction of adverse reactions, safer therapy selection	Population-specific variants require careful consideration; guides precision medicine
Ethical, Legal, & Social Implications (ELSI)	Informed consent, privacy, equity, access, data sovereignty, societal attitudes	Crucial for ethical implementation of pharmacogenomic testing and maintaining patient trust

Core concepts in pharmacogenomics and medical genetics are critical to personalized medicine and cover genetic variation and drug response, genomic technologies and data interpretation, and ethical, legal, and social implications. Genetic Variation and Drug Response identifies polymorphisms and alleles associated with clinically important pharmacokinetic and pharmacodynamic mechanisms, illustrates population diversity, and describes clinical examples. Genomic Technologies and Data Interpretation outlines sequencing modalities and data processing, variant classification, and associated interpretation challenges, and highlights societal issues of data sovereignty and privacy. Ethical, Legal, and Social Implications examines consent, disclosure, equity and access to testing, and ramifications for patient autonomy and trust.

Genetic Variation and Drug Response

Genetic variation contributes significantly to the variability in drug response among individuals. Polymorphisms in genes encoding drug-metabolizing enzymes, transporters, receptors, and pharmacodynamic pathways affect pharmacokinetic and/or pharmacodynamic processes, building the foundation for pharmacogenomics, which studies how genes affect individual responses to drugs (de Leon, 2009). Over 800 pharmacogenomic-related genes have been associated with >200 non-oncological drugs, and guidelines for >250 drug-gene pairs have been established in the clinical setting (Barone et al., 2009). The clinical significance of pharmacogenomic variation has been acknowledged by regulatory organizations and the availability of pharmacogenomic testing has increased; however, commons misinterpretation regarding the application of pharmacogenomic data persist. Due to population diversity, possesing the same pharmacogenomic variant does not guarantee similar metabolizer status or the same anticipated clinical outcome, underscoring the emergence of pharmacogenetically guided treatment

procedures for safer and more effective medication choices. Awareness of the potential impact of human population diversity on genome variation has led to initiatives designed to list and make freely accessible to the public pharmacogenomic variants coinciding with major human populations (Ziyaev, A. A., et al).

Genomic Technologies and Data Interpretation

Among the tools available for genomic analysis are four sequencing modalities: whole-genome sequencing (WGS) for de novo sequencing or resequencing; whole-exome sequencing (WES) for focused analysis of coding regions; targeted gene panels for custom selection of specific genes; and RNA sequencing (RNA-seq) for tracking expression or genotyping mutation transcripts. Before interpretation, data undergo four processing steps: quality control; alignment of reads to a reference genome; realignment of variants to reduce false positives and false negatives due to sequencing errors or homopolymers; and variant calling (Abdurakhmanov, J., et al). The four widely used variant formats are single-nucleotide variants (SNVs), insertions, deletions, and structural variants. Variants are subject to classification on a five-tier scale according to the American College of Medical Genetics and Genomics, based on criteria such as population frequency, segregation analysis, computational prediction, functional assays, and disease association; challenges remain for variants of uncertain significance due to incomplete functional knowledge. Finally, the interpretive report must address unique aspects of pharmacogenomics, including the lack of comprehensive analysis for gene-drug pairs and the limited availability of relevant recommendations and clinical decision-support tools (Giannopoulou et al., 2019).

Pharmacogenomics establishes a direct connection between genomic variation and drug response, providing a more tractable domain for education in genomics than disorders with complex genetic and environmental determinants. Genomic technologies and data handling procedures for pharmacogenomics differ in important respects from those in predictive genetics, medical genomics, and non-genetic clinical decision support. Emerging issues such as data sovereignty, data privacy, and the ethical use of genetic information complement the knowledge base that is already needed for pharmacogenomic education within a medical genomics context (Potamias et al., 2014). Data sovereignty concerns arise because students in some regions cannot legally collect their own personal health or genetic data; privacy requirements frequently restrict the database variants shared with students; and many countries permit the use of personal health records only for training at designated institutions.

Ethical, Legal, and Social Implications

Ethical, legal, and social issues are significant in pharmacogenomics. Ethical concerns include informed consent, secondary information, and privacy (Ziyaev, A. A., et al). Legal implications involve regulations on genetic testing and data sharing. Social implications encompass disparities in access, racial classification, and societal attitudes toward personalized medicine. Education of health professionals and the community is vital, along with policies that address ethical challenges in implementing pharmacogenomic testing and ensuring equitable benefit distribution (Mahmutovic et al., 2018).

Curricular Gaps in Medical Education

Pharmacogenomics-the study of the interaction between genes and drugs-enables personalized medicine by allowing selection of the optimal treatment for an individual. Pharmacogenomics education remains limited in most medical programs (Basyouni & Shatnawi, 2020), yet these curricula also include important genetic principles relevant to pharmacogenomics. An integrated educational framework that builds pharmacogenomics knowledge on preexisting medical genetics concepts would enhance training for personalized medicine. Supplementing existing pharmacogenomics instruction with an integrated educational approach helps train the next generation of clinicians. Understanding the genomic basis of health and disease is essential for medical professionals to incorporate precision medicine into clinical practice (C. Nutter & Gálvez-Peralta, 2018). Across preclinical and clinical stages, knowledge, skills, and attitudes related to both foundational genomic and pharmacogenomic content remain underdeveloped. The

absence of foundational genomics training limits the effectiveness of pharmacogenomic education and underscores the need for integration. Important competencies have been defined to address unmet genomic training needs; gaps in other core areas warrant attention to prepare future physicians for emerging practices in personalized medicine (Azimova, S., et al).

Pedagogical Frameworks for Integration

Advances in genomics and molecular technologies have the potential to transform many areas of medicine, from drug discovery to precision medicine (C. Nutter & Gálvez-Peralta, 2018). Instead of pursuing a multistep pathway, multidimensional or multi-level pharmacogenomics research can proceed simultaneously as part of the same strategy. Work that spans drug genes, complex diseases and microbiome has begun to be communicated as a next-generation pharmacogenomics effort. Integrating pharmacogenomics education into the medical genetics curriculum presents a timely opportunity to ensure students are prepared for the anticipated shift toward genomic medicine and personalized pharmacotherapy (Sasmakov, S. A., et al). Significant curricular gaps exist in South African medical training, and there is already an extensive and politically relevant pharmacogenomics discourse in the country. Two complementary frameworks support the integration of pharmacogenomics into the medical genetics curriculum: competency-based education specific to genomic medicine and interprofessional collaborative case-based learning. Addressing the need for better education in pharmacogenomics fits well with wider efforts already under way to strengthen medical genetics and genomic medicine training (S Boguski et al., 2013) [table 2].

Table 2: Educational Frameworks for Integrating Pharmacogenomics

Aspect	Key Points	Implementation / Strategies
Curricular Gaps	Limited pharmacogenomics education in most medical programs; foundational genomics knowledge often missing	Integrate pharmacogenomics into existing medical genetics curriculum; address competencies in genomics and drug-gene interactions
Competency-Based Education	Focus on skills, knowledge, and attitudes related to genomic medicine	Ensure clinicians can interpret pharmacogenomic results, apply them to patient care, and understand societal implications
Case-Based & Interprofessional Learning	Collaborative learning using real-life scenarios	Enhances practical understanding; bridges gap between theory and clinical application
Pedagogical Goal	Prepare students for personalized medicine	Train physicians to incorporate genomic and pharmacogenomic data into diagnosis, therapy selection, and precision pharmacotherapy
Integration Rationale	Personalized medicine relies on genetic variation and pharmacogenomic knowledge	Linking pharmacogenomics to medical genetics strengthens understanding and clinical readiness

Competency-Based Education for Genomic Medicine

The Core Competencies on Genomic Medicine from the publi-cation “Core Competencies for Health Professionals in Genomic and Genomic-Related Technologies” by the National Human Genome Research Institute (NHGRI) serve as a comprehensive pedagogical framework for incorporating genomic medicine within core medical curricula. These competencies are being used to facilitate the integration of

pharmacogenomics and medical genetics content within preclinical and clinical curricula at the University of Southern California. A mapping of the core competencies to the Accreditation Council for Graduate Medical Education (ACGME) Clinical Learning Environment Review (CLER) Program's six focus areas is also provided to help guide the incorporation of pharmacogenomics and medical genetics education into instructional programs and curricula across various degree programs. The six focus areas are patient safety, health care quality, care transitions, supervision, fatigue management, and team work and interprofessional education (C. Nutter & Gálvez-Peralta, 2018).

A curriculum plan to integrate pharmacogenomics into medical genetics education builds from the Genomic Medicine Academic Alignment: 2020 Status Update by the Association for American Medical Colleges (AAMC) Working Group on Pharmacogenomics Education. The AAMC report identifies gaps in pharmacogenomics knowledge and skills and recommends Pedagogical Frameworks for Integration as a strategic pathway for addressing these gaps throughout preclinical and clinical education (L. Kudron et al., 2023).

Interprofessional Collaboration and case-Based Learning

Personalized medicine encompasses the tailoring of medicine to the individual characteristics of each patient. Pharmacogenomics, the study of how genes affect a person's response to drugs, is the key component of personalized medicine that aims to optimize medication therapy according to each individual's genetic makeup. Pharmacogenomics is currently playing a pivotal role in the advancement of personalized medicine (Omonov Q., et al).

Personalized medicine has become a national and international policy goal, as noted in Government Accountability Office reports (Sasmakov, S. A., et al). The National Institutes of Health (NIH), the Food and Drug Administration (FDA), the National Research Council and other public and private organizations have published reports advocating for the academic preparation of health professionals knowledgeable about the implications and applications of personalized medicine. Reports from Canada's Genome Canada and the Netherlands Genomics Initiative specifically call for the education of pharmacists and pharmacy students to prepare them for the greater clinical role they will have as personalized medicine, genomics and pharmacogenomics become integrated into health care. In a report, the World Health Organization (WHO) cites pharmacogenomics as one of the important fields of personalized medicine (Azimova, S., et al).

Pharmacogenomics has been integrated into pharmacy education since 2005, and the American College of Clinical Pharmacy (ACCP) provides recommendations for educating pharmacy professionals (C. Nutter & Gálvez-Peralta, 2018). Despite the integration of pharmacogenomics into pharmacy curricula, knowledge gaps remain, especially regarding the clinical application of pharmacogenetic testing and other aspects of the clinical use of pharmacogenomics. Completion of large-scale whole-genome sequencing projects has dramatically increased the need for and interest in genomics and pharmacogenomics (D Surofchy et al., 2017).

Genetic variation among individuals influences how they respond to medications, impacting the choice of drug and dosing. For instance, the number of marketed medications with pharmacogenomic guidelines from the Clinical Pharmacogenetics Implementation Consortium (CPIC) has more than doubled in the past four years. The path from clinical pharmacogenomic testing results to the selection of a drug-gene pair to the appropriate clinical action is complex and requires careful consideration of multiple factors. Genetic variation among individuals influences variables such as the selection of drug and dosage, therapeutic effectiveness, and adverse drug reactions. The Guidance for the Licensing of Products for Pharmacogenomic Studies emphasizes the need for training in genomics for health-care professionals.

Assessment Strategies and Learning Analytics

Flexibility, authenticity, formative feedback, and analytics contribute significantly to preparatory learning in pharmacogenomics and medical genetics by promoting student learning, motivation, engagement, and self-regulation (D Surofchy et al., 2017). A blend of real-world, authentic, and simulated assessment formats spanning various types of decision making can further ensure that students appreciate the relevance of their education to practice (Anderson et al., 2021). Formative diagnostic assessment tools guide students toward substantive improvements, while a fully specified rubric identifies key competencies, outlines expectations for higher-order implementation, and supports the timely, consistent, and targeted feedback essential for readiness preparation. Data gathering for the curricular planning phase provides diverse evidence of student performance and relative proficiency across curriculum components, together with action reports that facilitate reflective, evidence-based discussions at program review stages and align with continuous quality improvement requirements.

Curriculum Design for Pharmacogenomics in Medical Genetics

Foundational modules in medical genetics and genomics education serve as groundwork for pharmacogenomics knowledge and applications. Key concepts from these modules include drug therapy response-related polymorphisms or alleles of pharmacokinetic or pharmacodynamic-related genes of key drug-metabolizing enzymes, drug transporters, or target proteins that differ in incidence among populations. Convergence of population-specific variants into a single pharmacogenomic description is clinically relevant when stratifying populations for higher or lower than normal doses of particular drug therapies or for increased risk of adversities such as toxic effects.

Pharmacogenomics-related content spans separate modules as theme-based integrative discussions across medical genetics or medical genomics courses. Mapping of drug-gene relationships implicated in clinical pharmacogenomics guidelines into educational modules highlights specific drug therapy response-predicting gene variants, anticipated implications for drug-protocol dose derivation, increased predisposition for life-threatening adversities, and clinical guidelines directing consideration or implementation of pharmacogenomics in drug therapy. Clinical scenarios modeling pharmacogenomic information facilitate differential diagnosis and accurate intervention using decision support systems for drug therapy selection, dose evaluation, adversity management, and outcome monitoring (Bakhronova D., Omonov Q).

Foundational Modules in Genomics

Genomics education must start by covering foundational concepts for an adequate understanding of pharmacogenomics and medical genetics. The gene concept should be introduced, along with specific variations in the pharmacogenome, inheritance patterns relevant to pharmacogenomics, and the clinical relevance of pharmacogenomic markers (P. Metcalf et al., 2010).

The introduction of foundational modules must precede the development of more advanced genomics-themed modules to cover core concepts that learners are expected to have prior knowledge of. This approach addresses curriculum constraints in terms of student preparation and available time for pre-course readings (L. Kudron et al., 2023).

Pharmacogenomics Themed Modules

About 25% of all US prescriptions are associated with serious toxicity or therapeutic failure. Pharmacogenomic markers are already identified for more than 25% of the commonly prescribed drugs available in the US. Yet, most practitioners lack the knowledge and skills to apply pharmacogenomic information in clinical practice. Pharmacogenomic education is key for the responsible use of genomic technologies and the implementation of clinical genomic programs (W. Guy et al., 2020). Pharmacogenomic education is required for all health professionals (C. Nutter & Gálvez-Peralta, 2018). Pharmacogenomics comprises three fundamental principles: drug-gene pairs, dosing implications, and adverse-drug-reaction risk.

Pharmacogenomic-education gaps persist despite the importance of genomic data for prescribing medications. It would be desirable to develop themed modules on pharmacogenomics in a medical-genetics curriculum. A pharmacogenomics-themed module would complement existing genomics teaching and provide significant pharmacogenomics education at the preclinical level.

Clinical Pharmacogenomics Decision-Making

Drawing from clinical examples observed in practice, pharmacogenomics decision-making in clinical medicine can be readily modelled as a decision pathway familiar to students. A schematic of the decision-making process highlights patient presentation and clinical context at the starting point. Subsequent steps guide the user through relevant data-gathering and interpretation towards pharmacogenomic recommendations. Pathways have been constructed to integrate seamlessly with clinical pharmacogenomic algorithms like the CPIC guidelines. These pathways can be progressed through at varying levels of detail, with supplementary patient scenarios, to accommodate different contexts. The decision-making process can also incorporate existing electronic decision-support systems that enable the pragmatic implementation of pharmacogenomic knowledge (W. Guy et al., 2020).

By modelling the decision-making framework in pharmacogenomics, students can approach the study of pharmacogenomic markers explicitly with full deployment of their existing knowledge and skills in both pharmacotherapy and genomic medicine. The integrated clinical pharmacogenomics curricula developed place the subsequent pharmacogenomic content in undergraduate and postgraduate medical education into the broader context of decision-making relevant to their future practice (Isroilova D., et al).

Integrative Clinical Scenarios and Case Studies

A strong emphasis on preparedness for personalized medicine has fueled collaborative efforts to integrate pharmaceutical genomics (PGx) and medical genetics education (C. Nutter & Gálvez-Peralta, 2018). Carefully crafted clinical scenarios that are authentic, complex, and holistic promote the application of pedagogical approaches to generate, evaluate, and address pharmacogenomics questions and dilemmas. Such scenarios would bridge genomic and pharmaceutical knowledge and skill domains and encompass both direct-to-consumer and health-care-provider approaches to pharmacogenomics, thus covering many medically relevant variants and drugs. Cases that incorporate diverse populations and conditions would model experiential learning within a collaborative team framework along a competency-based pathway, further engaging the next generation of health professionals (W. Guy et al., 2020).

Faculty Development and Resource Allocation

Particularly challenging for faculty governance and resource distribution are the dimensions of time and effort, key considerations for faculty participation. Coordination of training time necessitates central oversight for equitable resource allocation to those willing to coordinate complimentary training. Participation incentives also require attention. Preference postponement is advisable until promotion and tenure hold priority. Without an established faculty training program, inquiring about pending pharmacogenomic training capacity and willingness is premature. Targeting freestanding continuing education programs places decision-making within higher levels of the institution and sidesteps faculty engagement. Current interest centres on government-provided funding for Ontario University CADTH-HTA mechanisms and conscientious application of continuing education resources enabling pharmacy embracing initiatives (Basyouni & Shatnawi, 2020). Assistance towards capacity expansion lending to pharmacogenetics augmentation also of importance (C. Nutter & Gálvez-Peralta, 2018).

Evaluation and Continuous Improvement

New healthcare initiatives that employ the use of genome sequencing in clinical labs and pharmacy settings transform biotechnology from a scientific discipline to a tool that directly influences the practice of medicine. Pharmacogenomics sits at the interface of biotechnology and medicine; as such, decent

education, training and scholarship that thoroughly integrate pharmacogenomics with the genetic aspects of medicine are paramount. A recent article points out unexpected gaps in medical education about drug-gene relationships after biomedical knowledge and databases had been widely available for a decade (Basyouni & Shatnawi, 2020). Pharmacogenomics education has flourished in pharmacy but lags substantially in medicine, the two fields traditionally enjoy a significant degree of separation. Much discourse concerning such education regulated pharmaceutical paradigms, yet a survey indicated that the higher stakes lie in medical training. Further benchmarking with the competencies of pharmacy reveals that analogous medical training in pharmacogenomics is measurably important. Pharmacogenomics proves useful for safe & effective prescription at multiple drug-gene pairings; extensive guidelines document these pairings, and they steadily proliferate, rendering various aspects of pharmacogenetics highly pertinent (Bobomuratov T., et al).

Conclusion

Medical trainees' readiness to practise pharmacogenomics, a vital aspect of personalized medicine, is inadequate. Addressing this gap requires expanding the incorporation of pharmacogenomics in medical curricula, alongside interprofessional educational opportunities. Adoption of integrated and collaborative curricula is essential to meet student, regulatory, and societal expectations related to genomic medicine (C. Nutter & Gálvez-Peralta, 2018). Medical institutions are in a unique position to advance personalized medicine and pharmacogenomics through education, practice, research, and advocacy. Current curricular integration is limited; pharmacogenomic initiatives are often institution and discipline specific and do not form part of interdisciplinary strategies. Integrated pharmacogenomic educational frameworks have the potential to enhance clinical care, position institutions as leaders in genomic education, and facilitate accreditation compliance (D Surofchy et al., 2017).

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