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Short Communication

Pharmacogenomics: Current state, implementation challenges, and practical recommendations

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Abstract

Pharmacogenomics (PGx) — the analysis of the ways in which genetic variation affects drug response — is increasingly transitioning from research to standard clinical practice. Increasing evidence, carefully curated implementation guidelines, and regulatory endorsement now substantiate the use of genotype data to tailor therapy for many of the most widely used drugs. Despite obvious advantages (enhanced efficacy, reduced adverse drug reactions), there are still barriers: disparate test quality, uneven reimbursement, low clinician familiarity, and dispersed clinical decision support. This brief communication provides an overview of the state of clinical pharmacogenomics, identifies successes, and challenges of implementation, and provides useful suggestions for implementing PGx into daily practice.

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1. Introduction

Adverse drug reactions and therapeutic failures are principal causes of morbidity, mortality, and healthcare expenses. Pharmacogenomics provides an avenue to individualize therapy by aligning drug choice and dosing with a patient's genetic background (e.g., CYP, TPMT, HLA alleles). The past decade has seen the discipline come of age: evidence syntheses, clinical practice guidelines, and regulatory gene–drug association tables give advice that clinicians can apply today. Large programmes — in particular the CPIC — issue peer-reviewed, gene–drug guidelines that translate genotype to prescribing recommendation.[1] Regulators like the FDA have pharmacogenomic biomarker tables and increasingly cite PGx data in labelling, upholding clinical utility.[2]

1.1. Current evidence and guideline landscape

There are now dozens of CPIC guidelines covering high-impact genes (CYP2C19, CYP2D6, CYP3A5, TPMT, HLA-B*15:02, etc.) with actionable prescribing recommendations for many commonly used drugs.[1] Systematic evaluations have shown substantial concordance between CPIC guidance and actionable regulatory labeling across many gene–drug pairs, although gaps and inconsistencies persist.[3] Recent reviews conducted by Rim et al.[4] also report growth in both the evidence base and implementation projects evidencing decreases in adverse events and earlier achievement of therapeutic targets upon the use of PGx.

1.2. Implementation models & real-world outcomes

Three primary models of PGx delivery have arisen:

1. Reactive testing—genotype ordered at time of prescribing;
2. Pre-emptive panel testing—multi-gene panels done once and results kept in electronic health record (EHR) for future reference;

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3. Embedded pharmacy programs—pharmacists preside over testing and interpretation within clinics or hospitals.

Preemptive strategies prevent point-of-care delays and have been successfully used in several health systems.[5] Integrated pharmacist-led services enhance uptake and patient counseling, and active clinical decision support is important for the translation of genotype into practice.[6]

2. Discussion

The path of pharmacogenomics demonstrates both scientific advance and translational lag. Although the clinical advantages of PGx are underpinned by numerous studies, such as enhanced outcomes in cardiovascular medicine, psychiatry, and oncology,[4,6] adoption into everyday practice is unclear. This mirrors a larger problem with precision medicine: evidence development has ahead of infrastructure, reimbursement, and education.[7] Globally, well-supported informatics and policy-sensitive health systems (such as the U.S. and some areas of Europe) have proceeded more rapidly in integrating PGx into pathways of care.[5] But in resource-poor settings, the problem is compounded by varying allele frequencies and restricted laboratory capabilities.[3] This highlights the necessity of developing panels that capture local genetic diversity, not importing Eurocentric models for testing. Clinician acceptance and workflow are also an important issue. Surveys routinely indicate that numerous doctors are not sure how to interpret PGx results; integrating pharmacists and genetic counselors has been successful.[6]

Furthermore, clinical decision support needs to weigh beneficial reminders against avoiding "alert fatigue" within electronic health records.[4] In the future, wider implementation will depend on policy incentives, payer acceptance of cost-effectiveness, and international collaboration to harmonize testing. Improvements in sequencing technology and declining costs will make pre-emptive multi-gene testing possible, and PGx a standard inclusion in preventive health screening.[1,2] Finally, it will take a synergy of evidence, technology, policy, and education for sustained progress.

3. Key Barriers and Practical Challenges

Laboratory standardization & interpretation: Commercial PGx tests differ in their coverage, nomenclature, and translation to phenotype; clinicians need to ensure tests report clinically actionable alleles and standardized calls of the phenotype.[3]

Clinical decision support & EHR integration: Without CDS embedded at the prescribing point, PGx findings are not utilized adequately.[4]

Reimbursement & health-economic evidence**:** Inconsistent coverage policies exist; more robust health economic information are required.[7]

Clinician education & workflow: Prescribers often receive little training in genomic medicine; pharmacists can fill the gap.[6]

Equity & population diversity: Most PGx information over-represents European populations; allele frequencies vary worldwide, influencing panel design and guideline applicability.[3]

4. Practical Recommendations

Begin with high-priority gene–drug pairs: Target actionable CPIC/FDA-recommended pairs with good evidence, including CYP2C19–clopidogrel, HLA-B*15:02–carbamazepine, TPMT–thiopurines, and CYP2D6/CYP2C19–antidepressants.[1,2]

Employ standardized reports and CDS**:** Implement tests with standardized allele and phenotype calls, and link to CDS to provide clear prescribing guidance.[4]

Involve pharmacists and genetic counsellors: Pharmacists and geneticists facilitate uptake and offer patient counselling.[6]

Design panels in accordance with population genetics: Make sure PGx panels contain alleles of significance to the local population.[3]

Gather outcome and cost information: Local clinical effect and economic evidence is imperative for sustainability.[5]

5. Conclusion

Pharmacogenomics has evolved from an experimentally driven discipline into a clinically actionable one with robust evidence, consensus recommendations, and regulatory guidance. Conversion of gene–drug pairings to prescribing advice using tools like CPIC and the FDA biomarker table illustrates that PGx is no longer something of the future but an established reality. Clinical application has already yielded tangible gains, such as fewer adverse drug events, better dosing, quicker therapeutic response, and enhanced patient satisfaction.[4,6] Despite these achievements, various systemic issues persist. Standardization of tests, integration with EHRs, clinician training, and reimbursement are key hurdles that should be addressed to facilitate the widespread uptake. Equity issues are just as significant; PGx testing panels and recommendations need to accommodate global genetic variation instead of Eurocentric allele frequencies.[3] The mismatches underscore the necessity for international coordination, consistent nomenclature, and diverse population studies.

In the future, the use of pharmacogenomics in standard care is likely to increase with improvements in sequencing technologies, lowering costs, and growth of pre-emptive, panel-based testing models. Incorporating PGx into preventive care, primary care, and hospital formularies has the potential to make drug prescribing a more accurate, data-

driven process. In addition, interdisciplinary collaboration between pharmacists, physicians, genetic counsellors, bio-informaticians, and policymakers will be needed to develop sustainable and scalable programs. Finally, pharmacogenomics is a paradigm that shifts modern therapeutics away from trial and error prescribing and toward evidence-based individualization. Healthcare systems that make an investment today in establishing PGx infrastructure will not only mitigate the burden of adverse drug reactions but also be at the vanguard of precision medicine. The coming decade offers a unique opportunity to translate decades of genetic research into everyday clinical practice, making personalized prescribing a universal standard of care.

6. Conflict of Interest

None.

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