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Role of Genetic Polymorphisms in Individual Variations in Drug Response

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Abstract

Interindividual variability in drug response remains a major challenge to safe and effective pharmacotherapy. Genetic polymorphisms in genes encoding drug-metabolizing enzymes, transporters, therapeutic targets, and immune recognition proteins contribute to differences in drug exposure, efficacy, and adverse reactions. This narrative review synthesizes published international evidence concerning the molecular basis and clinical relevance of pharmacogenetic variation. Six domains are considered: the biological foundations of polymorphism; cytochrome P450 variation; cardiovascular pharmacogenetics; oncology and analgesic prescribing; HLA-associated hypersensitivity; and clinical implementation. Well-established gene–drug associations include CYP2C19 with clopidogrel, CYP2C9 and VKORC1 with warfarin, SLCO1B1 with statin-associated toxicity, TPMT and NUDT15 with thiopurines, DPYD with fluoropyrimidines, and selected HLA alleles with severe drug reactions. Although preemptive testing has demonstrated clinical promise, not every genetic association is actionable, and the predictive value of genetic results depends on clinical context, ancestry-inclusive test design, concurrent medicines, and physiological factors. Integrating validated pharmacogenetic guidance with clinical judgment offers an important route toward more individualized prescribing.

Keywords: Pharmacogenetics, Genetic Polymorphisms, Drug Metabolism, CYP450, Pharmacogenomics, Personalized Medicine, Adverse Drug Reactions, Precision Therapeutics

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1. Introduction

Patients receiving the same medication at similar doses may experience different therapeutic outcomes: some improve as intended, others show little benefit, and some develop serious toxicity. Variation in body size, age, disease severity, kidney and liver function, adherence, and interactions between drugs contributes to this heterogeneity. Inherited genetic differences can also alter the way medication is absorbed, activated, transported, metabolized, eliminated, or recognized by the immune system. Pharmacogenetics studies these relationships to make drug selection and dosing more informed. The clinical relevance of polymorphisms differs markedly across gene–drug pairs. A well-validated variant may support selection of an alternative medicine, whereas another variant may have

only a small, inconsistent association with outcomes. This narrative review considers six distinct themes and distinguishes mechanistic associations from evidence that changing prescribing practice improves patient outcomes. It is an interpretive review of published findings, not a new cohort study or systematic meta-analysis.

Molecular Foundations of Pharmacogenetic Variation

Genetic polymorphisms include single-nucleotide substitutions, insertions or deletions, gene-copy-number changes, and structurally complex alleles. Their biological effect depends on whether a variant changes protein structure, alters gene expression, disrupts splicing, or occurs in linkage with another functional variant. Because medicines interact with multiple biological pathways, the same inherited difference may be clinically important for one drug and irrelevant for another. The central purpose of pharmacogenomics is to translate this molecular variation into reliable prescribing decisions (Relling & Evans, 2015; Evans & Relling, 2004). Enzymatic polymorphisms can alter metabolic clearance or the activation of prodrugs. Variants in transporters can modify drug entry into hepatocytes, while variants in receptors or other targets may influence sensitivity at a given concentration. Immune-associated polymorphisms are mechanistically different because they can predispose a patient to hypersensitivity rather than simply increasing circulating drug levels. These categories should not be conflated when interpreting the likelihood of benefit or harm (Whirl-Carrillo et al., 2021). Genotype is the inherited combination of alleles, whereas phenotype describes the predicted or observed functional state. Clinical reports often classify certain enzymes as poor, intermediate, normal, rapid, or ultrarapid metabolizers. The classification is gene-specific and rests on experimentally and clinically supported allele-function assignments. Standardized terminology helps clinicians interpret laboratory reports consistently across testing platforms and electronic records (Caudle et al., 2017). Genetic effects also interact with nongenetic conditions. An enzyme inhibitor can make a genetically normal metabolizer behave functionally like a poor metabolizer, a process sometimes described as phenoconversion. Kidney or liver impairment can independently change drug exposure. Thus, inherited polymorphisms explain an important fraction, but never all, of response variability. Applying pharmacogenetic results requires attention to indication, disease stage, concomitant drugs, and the expected consequences of changing therapy (Swen et al., 2011). Molecular literacy is therefore essential for clinical interpretation: a variant's existence does not establish its usefulness as a screening test. Successful translation requires replicated associations, clear allele nomenclature, analytical validity, clinical relevance, and evidence that a prescriber can take an appropriate action. The expanding evidence base supports a targeted, clinically contextual approach rather than indiscriminate reliance on genetic profiles.

Cytochrome P450 Polymorphisms and Metabolic Phenotypes

The cytochrome P450 family comprises major enzymes involved in the oxidative metabolism of many prescribed drugs. Clinically relevant genes include CYP2D6, CYP2C19, CYP2C9, and CYP3A5. Their variants can change the formation of active metabolites and the clearance of pharmacologically active parent compounds. Consequently, a reduced-function genotype may increase exposure to an active drug while decreasing the activation of a prodrug. Understanding the direction of this effect is fundamental to safe interpretation (Crews et al., 2021). CYP2D6 is particularly complex because its locus contains whole-gene deletions, duplications, rearrangements, and numerous function-altering alleles. Clinical laboratories commonly translate detected combinations into activity scores and predicted metabolizer categories. A patient classified as ultrarapid may have increased activation of certain prodrugs, whereas a poor metabolizer may have limited activation. Yet such generalizations cannot be transferred automatically to drugs that have different metabolic pathways or active metabolites (Crews et al., 2014). CYP2C19 affects several antidepressants, proton pump inhibitors, and clopidogrel. Some alleles abolish function, whereas others increase activity. For antidepressants, a higher or lower concentration can influence tolerability or treatment response, but clinical interpretation must be based on the particular medication rather than the enzyme category alone. Contemporary guidance supports action for selected CYP2D6, CYP2C19, and CYP2B6–antidepressant relationships while finding insufficient support for routine prescribing decisions using some proposed

serotonin-transporter and receptor associations (Bousman et al., 2023). CYP2C19 also illustrates population variation in allele frequency. Alleles associated with decreased function are distributed differently across ancestry groups, so laboratory panels designed around only a narrow set of populations can miss relevant variation. A negative result for the variants included in a test is not proof of normal metabolism if the test does not interrogate important alleles or structural changes. Accurate interpretation requires transparent reporting of assay coverage, uncertainty, and phenotype assignment (Lee et al., 2022). Routine care should integrate enzyme predictions with clinical assessment, therapeutic drug monitoring when appropriate, and recognition of drug–drug interactions. The most effective use of CYP testing is targeted interpretation for a specific prescription and patient, not the assumption that a single metabolizer label predicts response to all medicines. This disciplined approach is more likely to improve medication safety and avoid unnecessary changes (Swen et al., 2011).

Selected Pharmacogenetic Mechanisms and Clinical Relevance

Gene / variant	Drug example	Functional implication	Clinical relevance
CYP2C19 loss of function	Clopidogrel	Reduced prodrug activation	Reduced antiplatelet response in relevant indications
CYP2D6 activity variation	Codeine	Variable morphine formation	Risk of low efficacy or toxicity
CYP2C9 / VKORC1	Warfarin	Altered clearance and target sensitivity	Variable dose requirements
SLCO1B1 reduced function	Some statins	Reduced hepatic uptake	Elevated muscle-toxicity susceptibility
TPMT / NUDT15 decreased function	Thiopurines	Impaired handling of thiopurine metabolites	Increased myelosuppression risk
DPYD decreased function	5-FU / capecitabine	Reduced drug breakdown	Elevated severe-toxicity risk
HLA-B*57:01	Abacavir	Immune predisposition	Hypersensitivity risk

Cardiovascular Pharmacogenetics and Treatment Selection

Cardiovascular treatment provides some of the clearest examples of clinically significant inherited differences. Clopidogrel is a prodrug requiring CYP2C19-dependent activation. Loss-of-function alleles can reduce active metabolite formation, leading to weaker platelet inhibition in certain patients. Studies in acute coronary syndromes and percutaneous coronary intervention linked these variants with diminished antiplatelet effects and higher risk of ischemic events, establishing a rationale for genotype-informed treatment selection (Mega et al., 2009; Simon et al., 2009). The magnitude of benefit from alternative antiplatelet agents is dependent on clinical indication and the evidence supporting that setting. Updated guidelines advise considering agents that do not rely on CYP2C19 activation for certain intermediate and poor metabolizers, subject to contraindications and individualized bleeding assessment. Genetic testing does not replace clinical decision-making, and results cannot be interpreted independently of cardiovascular history or procedural context (Lee et al., 2022). Warfarin presents a different mechanism because dose requirements reflect both metabolism and pharmacodynamic sensitivity. CYP2C9 affects elimination of the more active S-warfarin component, while VKORC1 variants influence sensitivity at the drug target. Algorithms combining genetic and clinical factors can estimate initial dose more accurately than approaches based on simple demographic characteristics alone, although accuracy varies across populations and very unusual dose requirements (International Warfarin Pharmacogenetics Consortium, 2009). Clinical utility should be judged by patient outcomes, not only predictive accuracy. A randomized European study reported

improved anticoagulation control when genotype-guided warfarin initiation was compared with a standard regimen, but findings from other trials and healthcare settings have not always matched. Such differences emphasize comparator choice, allele coverage, ancestry, and adherence to dose algorithms (Pirmohamed et al., 2013). Statin-related adverse effects highlight transporter polymorphisms. SLCO1B1 variants can reduce hepatic uptake of certain statins, increasing systemic exposure and susceptibility to muscle toxicity. Genome-wide evidence first demonstrated a strong association with simvastatin-induced myopathy, while updated implementation guidance also considers ABCG2 and CYP2C9 for relevant statins. This example shows how transporter and metabolic variation can jointly influence safe cardiovascular prescribing (SEARCH Collaborative Group, 2008).

Cancer Therapy, Analgesia, and Toxicity Prevention

Cancer treatment frequently involves narrow therapeutic margins, making inherited differences in drug processing clinically important. Thiopurines such as mercaptopurine, azathioprine, and thioguanine are influenced by TPMT and NUDT15 activity. Patients carrying decreased-function alleles can accumulate toxic metabolites and experience profound myelosuppression with conventional starting doses. Genotype-guided recommendations help clinicians consider safer initial regimens while maintaining treatment monitoring (Relling et al., 2019). The consequences of NUDT15 variants are especially relevant to global implementation because allele frequencies differ among populations. A recent guideline update emphasizes recognition of compound deficiencies affecting both TPMT and NUDT15 and illustrates why testing only one gene may overlook substantial toxicity risk. Treatment recommendations nevertheless depend on indication, conventional starting dose, and clinical response; genotyping complements rather than replaces blood-count surveillance (Maillard et al., 2026). Fluoropyrimidines provide another important safety example. Reduced activity of dihydropyrimidine dehydrogenase, encoded by DPYD, can markedly increase exposure to fluorouracil or capecitabine and predispose patients to serious toxicity. Clinical recommendations interpret selected DPYD variants to inform initial dose or alternative treatment decisions, while acknowledging that standard variant panels do not detect every person at risk. Close clinical monitoring remains essential even after a reassuring result (Amstutz et al., 2018). Prospective investigation of upfront DPYD*2A testing found that individualized fluoropyrimidine management could improve safety and offered a model for evaluating clinical and economic consequences of pharmacogenetic screening. The central lesson is that safety interventions should be judged by prevention of severe adverse reactions, feasibility, and effects on treatment delivery, rather than genetic association alone (Deenen et al., 2016). Pain management illustrates the opposite direction of metabolism-dependent effects for certain prodrugs. CYP2D6 activity influences codeine conversion into morphine: low activity may yield poor analgesia, while exceptionally high activity can create toxicity risk. Updated opioid guidance emphasizes clinically supported CYP2D6 associations and distinguishes them from genetic markers that currently lack adequate evidence for treatment selection. Prescribers must also consider respiratory risk, concomitant sedatives, patient age, and available nonopioid options (Crews et al., 2021).

Principles for Clinically Responsible Genotype Interpretation

Clinical step	Questions and safeguards	Purpose
Confirm indication	Is the gene–drug relationship relevant to this patient?	Avoid inappropriate extrapolation
Review genotype	Which alleles and copy-number changes were tested?	Recognize technical limitations
Assign phenotype	Use current validated gene-specific translation tables	Reduce interpretation errors
Consider interactions	Review other drugs, organ function, age, and disease	Account for phenoconversion

Select action	Choose standard care, an adjusted regimen, or an alternative according to guidance	Improve efficacy and safety
Monitor outcomes	Follow clinical response and toxicity despite testing	Identify residual risk
Document results	Record findings and update interpretation as evidence changes	Enable safe future prescribing

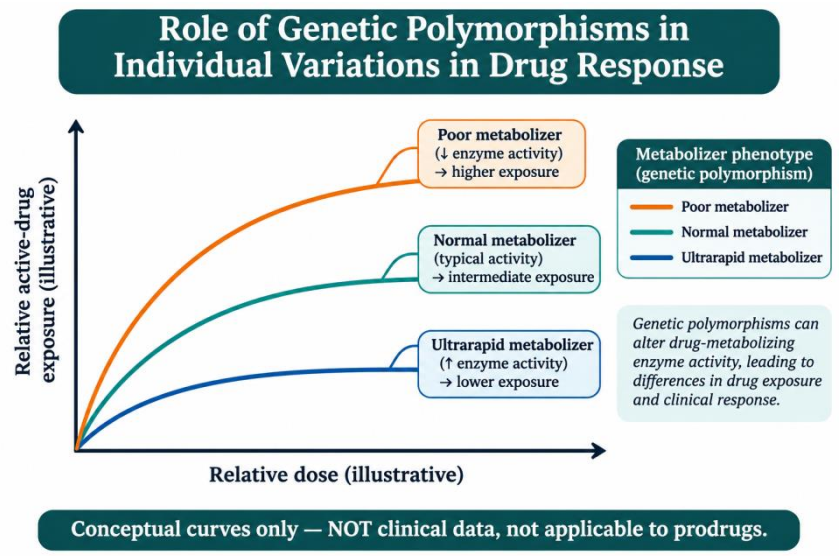
Immune-Mediated Hypersensitivity and HLA Risk Alleles

Not all inherited drug-response differences involve concentration or metabolism. Human leukocyte antigen (HLA) polymorphisms can influence drug-related immune recognition, sometimes creating a marked risk of severe hypersensitivity reactions. The clinical significance of these associations depends on a particular drug, allele, and immunological phenotype. Strong associations can support avoiding the implicated drug before exposure, offering a form of precision medicine focused on prevention of catastrophic adverse reactions (Mallal et al., 2008). HLA-B*57:01 is a well-established predictor of abacavir hypersensitivity. A prospective randomized screening study demonstrated the value of identifying at-risk individuals before starting therapy and withholding the drug where indicated. The result remains a landmark example because the intervention is clear and the adverse outcome is clinically meaningful. Screening does not imply that every carrier would otherwise experience toxicity; it provides a strategy for managing risk (Mallal et al., 2008). Other HLA associations involve severe cutaneous adverse reactions. The HLA-B15:02 allele was strongly associated with carbamazepine-related Stevens–Johnson syndrome in a Han Chinese population, demonstrating that immunogenetic susceptibility can show substantial population differences. These findings led to practical guidance on considering HLA status before carbamazepine and related prescriptions in relevant clinical contexts (Chung et al., 2004). Guidelines now address both HLA-B15:02 and HLA-A31:01 in carbamazepine prescribing and recognize different reaction spectra. Importantly, the absence of a high-risk genotype does not eliminate all risks of severe skin reactions, and a positive test does not predict the time or certainty of an event. Correct clinical application requires matching the right test to the right medicine and interpreting results alongside prior exposure and alternative treatment availability (Phillips et al., 2018). HLA-B58:01 and allopurinol-induced severe cutaneous adverse reactions provide another relevant gene–drug association. Recommendations aim to avoid exposure in genetically susceptible patients when clinically appropriate. Together, these examples highlight the need for accurate HLA typing, clear documentation, and caution in assuming that ancestry alone can replace individual genetic assessment (Saito et al., 2016).

Implementation, Equity, and the Future of Precision Prescribing

Genetic discoveries improve care only when the results reach prescribers in a usable form and lead to appropriate therapeutic decisions. Implementation involves sample collection, accurate laboratory analysis, clear interpretation, decision-support alerts, clinician education, data stewardship, and follow-up. Pharmacogenetic guidance generally explains what to do when a genotype is known; it does not always mandate universal testing before every prescription (Swen et al., 2011). Prospective international implementation research has tested the effect of broader pharmacogenetic panels rather than isolated gene–drug pairs. The PREPARE trial examined a 12-gene approach across healthcare settings and found reductions in clinically relevant adverse drug reactions among patients with actionable results who received genotype-guided prescribing. Interpretation still depends on study population, outcome definitions, uptake, and the medicines actually prescribed (Swen et al., 2023). Healthcare systems face practical obstacles, including testing cost, laboratory capacity, clinician familiarity, and the risk of alert fatigue. Preemptive programs have demonstrated how electronic health records can preserve results for future prescriptions, though programs require sustained technical and clinical oversight. Results may remain useful throughout life, but interpretation must be updated when allele-function knowledge or prescribing guidance changes (Dunnenberger et al., 2015).

Equity is central to responsible implementation. Many discovery datasets and clinical panels have historically overrepresented selected ancestry groups, while structural and rare variants may be missed by restricted assays. A more inclusive approach requires validation across populations, transparent test limitations, accessible counseling, protection of genetic privacy, and assessment of whether clinically important improvements extend to under-resourced services (Whirl-Carrillo et al., 2021). Standardized terminology reduces ambiguity when results are transmitted between laboratories and healthcare organizations. Clinicians should be able to identify the genotype, predicted phenotype, evidence level, and medication-specific implication. Future advances in sequencing, implementation science, and decision support may broaden access, but pharmacogenetic information must remain integrated with clinical assessment and shared decision-making rather than treated as a substitute for them (Caudle et al., 2017).



Summary

Inherited polymorphisms can substantially influence drug response through metabolism, transport, molecular targets, and immune pathways. Evidence supporting clinical action is particularly strong for selected CYP, TPMT, NUDT15, DPYD, SLCO1B1, and HLA associations. Nevertheless, a genetic result is not a complete prediction of therapeutic success or toxicity; drug interactions, physiological conditions, clinical indication, and the design of genetic assays remain decisive. Responsible precision prescribing depends on validated gene–drug guidance, clear interpretation, appropriate alternatives, outcome monitoring, and equitable testing access. Future work should prioritize ancestry-inclusive evidence, robust prospective outcome studies, and durable electronic clinical decision support.

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