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Using pharmacogenomics, clinicians can make informed decisions about which drug to administer to which patient, and at what dose.

Scientists Use Pharmacogenomics to Provide Personalized Medicine

By studying genome variants, researchers can identify which drugs are safe and effective for each individual patient.

Written By Rebecca Roberts, PhD

Published Dec 20, 2025 (UTC)

Medications do not work the same way for everyone. Many people carry genetic mutations that affect their response to drugs, from painkillers to cancer treatments. Researchers investigate pharmacogenomics to learn how such variations in the

genome affect a patient's therapeutic response. In this article, we delve into the science of pharmacogenomics, how clinicians use it to inform personalized medicine, and some key examples of how pharmacogenomics is applied in clinical settings.

What Is Pharmacogenomics?

Pharmacogenomics, a portmanteau of pharmacology and genomics sometimes abbreviated as PGx, is the study of how an individual's genetic variation affects their response to medication. The aim of pharmacogenomic testing is to increase drug efficacy and safety for each unique patient.[1]

Scientists use pharmacogenomics as a tool to inform clinical decisions and personalize drug treatments; by studying a person's genome, scientists can predict if a specific drug will be effective and at what dose, or if the patient is likely to experience adverse drug reactions (ADRs).

For example, many individuals have mutations in the cytochrome P450 (CYP) gene family, which encodes liver enzymes responsible for metabolizing a variety of medications. These mutations can reduce CYP enzyme activity, preventing the body from effectively metabolizing and eliminating drugs. Using genome sequencing, clinicians can identify if a patient carries these mutations, reducing the risk of drug-related toxicity or overdose.[2]

Genes that are known to affect drug responses, like the CYP family, are called pharmacogenes. Because they typically encode drug-metabolizing enzymes, transporters, or receptors, these genes can affect either pharmacokinetics (drug metabolism and transport) or pharmacodynamics (therapeutic actions).[3,4] There is also a growing body of evidence that epigenetic mechanisms can influence drug responses.[4]

What is the difference between pharmacogenomics and pharmacogenetics?

The terms pharmacogenomics and pharmacogenetics are often used

interchangeably by scientists.[5] However, pharmacogenetics may refer to the study of specific genes that affect a person's ability to metabolize drugs, while pharmacogenomics is a broader term that includes all the possible genomic variations that can affect drug responses.[5]

Pharmacogenomic Testing

There are several types of pharmacogenomic testing. In concurrent pharmacogenomic testing, clinicians sequence individual target genes after a drug is prescribed but before it is administered to a patient.[6] In contrast, reactive pharmacogenomic testing occurs when a drug has shown limited efficacy in a patient or after an ADR occurs.[7]

Pharmacogenomics panels

Scientists have developed pharmacogenomics panels that include a wide range of relevant genes that affect common medications. Using these panels, clinicians can perform pharmacogenomic testing pre-emptively, before prescribing certain drugs.[6] Several pharmacogenomics working groups and international consortia have recommended pharmacogenomic panel screening integration into standard healthcare settings. The Clinical Pharmacogenetics Implementation Consortium, for example, has published clinical practice guidelines for more than 100 pharmacogenes.[6]

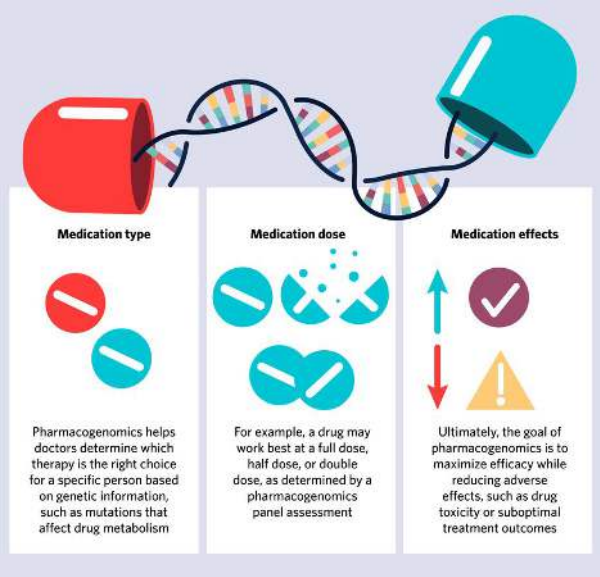
Pharmacogenomics workflow

In a standard pharmacogenomics workflow, clinicians use a blood, saliva, or buccal swab sample to isolate DNA from a patient, then perform either targeted genotyping of a specific gene or pharmacogenomics panel, or comprehensive next-generation sequencing.[8,9] Using these data, clinicians can make informed decisions about which drug to recommend to the patient and determine the appropriate dose.

Clinical Pharmacogenomics Applications

Pharmacogenomic testing for antidepressants

Scientists have demonstrated that pharmacogenomic testing can help tailor personalized treatments for depression and anxiety. Genetic variations, including those in the CYP gene family, account for around 60 percent of the variability in response to antidepressant drugs, such as serotonin reuptake inhibitors, serotonin-norepinephrine reuptake inhibitors, noradrenergic and serotonergic modulators, tricyclic and tetracyclic compounds, and monoamine oxidase inhibitors.[4]



Pharmacogenomics helps scientists optimize personalized therapeutic recommendations, such as which medication and dose will provide the best efficacy with the fewest adverse events.

Informed by their CYP mutations, patients can be categorized based on whether they will metabolize these drugs slowly, normally, or rapidly; patients who are genetically predisposed to slow therapeutic metabolism are at increased risk of ADRs and therefore require lower dosages, while those with ultrarapid metabolizing enzymes need higher or more frequent dosages for these drugs to be effective.[10]

Researchers have also shown that mutations in the monoamine and γ-aminobutyric acid metabolic enzymes, transporters, and receptors; uridine 5'-diphospho-glucuronosyltransferase enzymes; and P-glycoprotein ATP-binding cassette transporters affect antidepressant drug pharmacokinetics and pharmacodynamics.[4]

Cardiovascular pharmacogenomics

Cardiovascular disease treatment is another example of how clinicians can apply pharmacogenomics. Warfarin is an anticoagulant that doctors frequently prescribe to patients with cardiovascular disease, including myocardial infarction (heart attack), atrial fibrillation, and thrombosis (blood clots). However, warfarin has a very narrow therapeutic window; excessive anticoagulation can cause dangerous bleeds,

while inadequate responses can result in other adverse events.[1]

Research has shown that mutations in the CYP 2C9 enzyme and the vitamin K epoxide reductase complex subunit 1 gene can put individual patients at risk of either excessive bleeding while on warfarin, or adverse events, such as heart attack.[1] Based on robust clinical evidence, an international consortium published a pharmacogenomic algorithm that clinicians can implement to determine the correct dosages of warfarin for individual patients.[1]

Pharmacogenomics in cancer

Pharmacogenomics is also highly applicable in the context of cancer treatment. Many cancer treatments have narrow therapeutic windows and are associated with severe toxicity; using pharmacogenomics, clinicians can tailor cancer therapies to individual patients and avoid potential ADRs.[11]

Researchers have identified a range of genetic variants they can use to predict cancer treatment efficacy. This includes mutations in the genes that code for dihydropyrimidine dehydrogenase (DPYD), thymidylate synthetase, methylenetetrahydrofolate reductase, thiopurine S-methyltransferase, nucleoside diphosphate-linked moiety X-type motif-15, uridine 5'-diphospho-glucuronosyltransferase, ATP-binding cassettes, and others.[11]

For example, mutations in *DPYD* are associated with severe fluoropyrimidine toxicity that can be fatal.[12] Clinicians can use pharmacogenomics to identify patients that carry these *DPYD* mutations and significantly reduce the risk of these patients experiencing severe toxicity by reducing the fluoropyrimidine dose by 50 percent.[12]

Ethical Issues in Pharmacogenomics

Because pharmacogenomics is so effective in determining which drugs and dosages should be administered to patients, researchers and clinicians have proposed that pharmacogenomic testing be widely implemented in standard healthcare setting across the globe.[13] However, other scientists and bioethicists have voiced concerns over potential ethical issues in pharmacogenomics, such as the large-scale collection

of genomic data and ownership of such data, and the accessibility of treatments that are inferred from pharmacogenomic data.[13]

Future Outlooks

As researchers continue to learn more about how the genome affects drug responses and pharmacogenomics becomes integrated into standard healthcare, patients will hopefully benefit from more personalized treatment plans. By increasing the likelihood of drug efficacy and minimizing the risk of ADRs, pharmacogenomics could reduce the burden on global healthcare systems.

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