

CHEAT SHEET

Disruptive therapeutics and diagnostics series | Volume 4 of 6

Pharmacogenomics

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Key takeaways

- Pharmacogenomics (PGx) is the science of understanding the influence of a person's genome (i.e., their complete set of DNA and genes) on drug treatment outcomes.
- The push towards pharmacogenomics is particularly important right now because it holds the promise of contributing to smart drug development, improved patient outcomes, and lower total cost of care.
- Pharmacogenomics is disrupting three elements of healthcare: care delivery, cost/payment models, and clinical evidence.

DISRUPTIVE THERAPEUTICS AND DIAGNOSTICS SERIES

VOLUME 4 [This volume](#)

Pharmacogenomics

Pharmacogenomics (PGx) is the science of understanding the influence of a person's genome (i.e., their complete set of DNA and genes) on drug treatment outcomes.

VOLUME 1 [Read now](#)

At-Home Diagnostics

Home tests – also called self-tests or home-use tests – are typically sold over the counter and allow users to test self-collected specimens and interpret the results on their own without the help of trained health professionals.

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Biomarker Testing

Biomarker testing (also known as mutation, genomic, or molecular testing) uses laboratory tests to help the health care team gather as much information as possible about a patient's disease state.

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Deep Brain Stimulation

Deep Brain Stimulation (DBS) is the implantation of a device that sends a signal to brain areas responsible for body movement to improve the lives of patients with Parkinson's, other neurodegenerative diseases, and behavioral and mental health disorders.

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Psychedelic-Assisted Therapy

Psychedelic-assisted therapy entails using a high dose of a psychedelic substance to treat a variety of behavioral and mental health issues such as addiction, mental health conditions.

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Ultra High-Cost Drugs

Ultra high-cost drugs (UHCDs) are expensive, potentially durable, or curative therapies currently approved to treat conditions that are orphan and rare disease.

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What is it?

Pharmacogenomics (PGx) is the science of understanding the influence of a person's genome (i.e., their complete set of DNA and genes) on drug treatment outcomes. PGx testing can help providers treat patients precisely by predicting drug response and informing appropriate prescribing. These tests fall under the umbrella of precision medicine, which uses multiple data sources to treat patients effectively and proactively. Scientists and researchers have discovered that patients metabolize certain drugs differently, in large part due to genetic variations. PGx helps providers prescribe targeted medications that improve outcomes by understanding a patient's gene-drug interactions.

Relationship between precision medicine and PGx

PGx makes up one component of precision medicine



1. Inpatient care management relies more heavily on RNs and social workers to staff their programs.

Why does it matter?

Pharmacogenomics can help pharmaceutical companies develop drugs more efficiently, providers better target patients who would benefit from a given therapy, and patients better understand their risk for and avoid adverse drug reactions.

The push towards pharmacogenomics is particularly important right now because it holds the promise of contributing to smart drug development. By studying a drug only in people likely to benefit from it, drug companies might be able to speed up the drug's development and maximize its therapeutic benefit. This would allow some individuals to receive potentially lifesaving medicines that might otherwise have difficulty gaining approval because of the risk they pose for other patients. Pharmacogenomics can play a major role in developing new therapeutics for rare diseases that would fall under the ultra high-cost drug umbrella.

Over 97% of the US population carries a high-risk PGx variant, indicating that they have genetic mutations that affect how they metabolize drugs. This may put them at high risk of adverse drug reactions.

Talking to them about their options with PGx and providing them with more precise treatment can help improve their experience across the care continuum and change the face of healthcare delivery.

700K

Annual emergency department visits caused by adverse drug reactions



~\$4K

Annual savings per patient who received PGx testing for major depressive disorder



>97%

Of the US population carries a high-risk PGx variant



Source: "Preventable adverse drug reactions: a focus on drug interventions," U.S. Food and Drug Administration; Dunnenberger H, et al., "Preemptive clinical pharmacogenetics implementation: current programs in five US medical centers," *Annual Review of Pharmacology and Toxicology*, January 2015; "Medication errors and adverse drug events," Patient Safety Network; Maciel A, et al., "Estimating cost savings of pharmacogenetic testing for depression in real-world clinical settings," *Neuropsychiatric Disease and Treatment*, 2018.

What makes it disruptive?

Areas of disruption

Care delivery

PGx can disrupt the way physicians prescribe drugs by introducing another round of lab testing between diagnosis and prescribing. However, this enables clinicians to tailor care to what is most likely to work for individuals, reducing adverse drug reactions, trial-and-error prescribing patterns, and avoidable costs in disease management.

There are also workforce implications when implementing pharmacogenomics. Because genetic testing requires an addition of another round of testing to be managed by the care team, this may pose both a staffing and administrative burden.

Cost/payment models

Pharmacogenomics isn't applicable to all marketed drugs, and thus will need to undergo rigorous evaluation to justify costs. This will need to be on a case-by-case basis so that payers are able to justify investing resources in research and development of pharmacogenomics and make reimbursement decisions based on the evidence.

Although reimbursement is an evolving landscape for PGx, it is most commonly accepted for specific therapeutic areas- care in oncology, CV, and mental health where cost-effective evidence is most abundant. There will be difficulty in developing payment models for pharmacogenomic based therapies in therapeutic areas outside of these.

Additionally, genetic testing is expensive, and plans are still figuring out when paying for these tests is worthwhile.

(CONT.)

What makes it disruptive?

Clinical evidence

Currently, coverage can vary greatly and depends mostly on drug-gene evidence. The large amounts of genomic data that is collected will need more rigorous review, standardization, and protocols in place for every step in the process, from data collection to processing, analysis, and interpretation. This can place challenges on workforce shortages, burnout, and more.

Another challenge is that although pharmacogenomics holds the promise to enhance patient care by enabling treatments tailored to genetic make-up and lowering risk of serious adverse events, the evidence is still new and clinicians may not fully understand the ripple effects of looking into a patient's genome. Rates of genomic literacy among should be improved upon, so that there isn't a widening gap in knowledge when translating this to the patient.

A note on health equity

PGx has the potential to support health equity efforts if there is adequate representation in clinical trial data and evidence.

However, some new therapies or tests may be taken up less quickly and in lower numbers in more underserved and minority populations due to barriers in access, availability, ability to pay privately, and lack of understanding of healthcare information. This may result in studying drugs in groups on specific populations, as a result, frustrate health equity efforts.

Conversations you should be having

Sector	Conversations
Payers	<ul style="list-style-type: none"> • How can my organization gain buy in from physicians, influential stakeholders in the medical group, pharmacy, IT, informatics, and administration? • How can we integrate biomarker testing into our efforts to improve clinical outcomes and manage costs?
Health systems	<ul style="list-style-type: none"> • What are the clinical use cases for PGx in our patient population? • What internal and external resources are required to introduce and scale a PGx program?
Pharma	<ul style="list-style-type: none"> • How can my organization better equip providers with the tools to incorporate PGx into their clinical decision making? • Do we have appropriate and consistent frameworks for thinking about the evidence and data needs related to biomarkers?
Medtech	<ul style="list-style-type: none"> • How can we create the infrastructure to support and ensure genetic data integration with physician workflow?

(CONT.)

Conversations you should be having

Sector	Conversations
Cross-sector	<ul style="list-style-type: none"> • How can patient genomic data be managed and safeguarded? • How do we ensure diverse patients are represented in PGx research and have access to PGx-informed prescribing? • How can we support providers in incorporating PGx into clinical decision making? • What level of pharmacogenomic impact on patient outcomes warrants paying for expensive genetic testing?

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