

*To what extent can PGx tests aid in the effectiveness of patients' medical treatments and reactions to treatments?*

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As a doctor, you are taught to treat every patient fairly. You hammered in sleepless nights to memorize specific drugs for every condition. You completed med-school and now have your white coat. You know *everything*. Next thing you know, your patient is having adverse effects on your blood thinners and has to be sent to emergency procedures. Why is this? We are all humans, right? Still, every person has a unique genetic makeup and responds differently to medication. Thus, PGX testing is beneficial in adapting treatment to individual patients.

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### **What is PGX testing**

PGx testing analyzes a person's genetic makeup and determines if the genes of a patient can reduce the effectiveness or result in adverse effects of the medication. A sample of one's blood or saliva gets swabbed and sent off for diagnostic testing. PGx falls under the precision medicine umbrella, offering tailor-made treatments based on one's lifestyle, environment, and genes. Instead of following a "one shoe fits all" approach, PGx testing optimizes each patient's health.

#### 1. Cardiovascular Diseases:

- Warfarin (Coumadin) Therapy: PGX testing for CYP2C9 and VKORC1 genes helps determine the appropriate dose to minimize the risk of bleeding complications.
- Clopidogrel (Plavix) Therapy: Testing for the CYP2C19 gene variant can indicate how well a patient will metabolize the drug, which is crucial for patients undergoing stent placement.

#### 2. Psychiatric Disorders:

- Antidepressants: Genetic variations in CYP2D6 and CYP2C19 can affect the metabolism of medications like SSRIs and TCAs, impacting efficacy and risk of side effects.
- Antipsychotics: Variations in genes like CYP2D6 and CYP3A4 can influence the response to drugs such as risperidone and aripiprazole.

#### 3. Pain Management:

- Opioid Analgesics: CYP2D6 testing is important for medications like codeine and tramadol, as variations can lead to poor efficacy or increased risk of toxicity.

- Non-opioid Analgesics: Variations in CYP2C9 can affect the metabolism of NSAIDs like ibuprofen and celecoxib, influencing both efficacy and risk of adverse effects.
4. Oncology:
    - Chemotherapy: Variations in genes like TPMT and DPYD can affect the metabolism of thiopurines (e.g., mercaptopurine) and fluoropyrimidines (e.g., 5-fluorouracil), respectively, guiding dosing to avoid severe toxicity.
    - Targeted Cancer Therapies: Testing for specific mutations (e.g., EGFR, KRAS) can help determine the most effective targeted therapy for cancers such as lung and colorectal cancer.
  5. Neurological Disorders:
    - Antiepileptic Drugs: Genetic testing for HLA-B\*5702 and HLA-A\*3101 can help predict the risk of severe skin reactions to carbamazepine and oxcarbazepine.
  6. Infectious Diseases:
    - HIV Treatment: HLA-B\*5701 testing is recommended before starting abacavir to reduce the risk of a severe hypersensitivity reaction.
  7. Gastroenterology:
    - Thiopurine Drugs: TPMT testing is critical for patients with inflammatory bowel disease who are treated with thiopurine drugs (e.g., azathioprine) to avoid myelosuppression.
  8. Pulmonology:
    - Asthma: PGX testing for ADRB2 can guide the use of beta-agonists like albuterol in asthma management, ensuring optimal therapeutic response.
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### **Problems with PGx testing**

Though PGx testing is an effective method to guide treatment, the guidelines of testing are ambiguous. Certain companies have various metrics to analyze a patient's genetic makeup. Due to the infancy of precision medicine, the pharmacogenetics industry lacks standardization. Going forward, the FDA must further regulate this process and follow CPIC (Clinical Pharmacogenetics Implementation Consortium). Refer back to peer-reviewed guidelines that allow for proper treatments.

## Works Cited

*Center for Individualized Medicine - pharmacogenomics for patients.* Mayo Clinic. (2023, April 28).  
<https://www.mayo.edu/research/centers-programs/center-individualized-medicine/patient-care/pharmacogenomics/patients#:~:text=PGx%20is%20the%20study%20of%20eye%20color%20and%20blood%20type>.

*Fulgent pharmacogenetic testing (PGX) | fulgent genetics.* (n.d.).  
<https://www.fulgentgenetics.com/pgx>

*U.S. National Library of Medicine. (n.d.). Pharmacogenetic tests: MedlinePlus Medical Test.* MedlinePlus. <https://medlineplus.gov/lab-tests/pharmacogenetic-tests/>

*Pharmacogenomics (PGX) individualized medication therapy.* TriHealth. (n.d.).  
<https://www.trihealth.com/services/precision-medicine-and-genetic-services/our-services/pharmacogenomics>