

Understanding Medication Genetic Testing

How pharmacogenetic testing may help guide medication choices — and what it cannot tell us

A Guide for Patients, Families & Caregivers

What is pharmacogenetic (PGx) testing?

Pharmacogenetic testing looks for inherited differences in genes that affect how the body handles certain medications. Many medicines are broken down by enzymes in the liver. Gene variants can change how quickly some of these enzymes work. Testing usually uses a cheek swab or saliva sample; sometimes blood is collected.

This is not the same as liver-function testing. Blood tests such as AST, ALT, bilirubin, and albumin assess aspects of liver health; PGx testing examines inherited enzyme activity. A person can have normal liver tests and still metabolize certain drugs differently.

How liver metabolism can affect a medication

Possible result	What it may mean
Poor metabolizer	Some drugs may build up and cause adverse effects; drugs that need enzyme activation may work less well.
Intermediate metabolizer	Reduced enzyme activity may change exposure to certain medications.
Normal metabolizer	Typical activity for that gene; does not guarantee a medication will work or be safe.
Rapid / ultrarapid metabolizer	Some drugs may be cleared faster; some prodrugs may be activated more quickly.

Important: The meaning depends on the specific drug. “Fast” is not always better and “slow” is not always worse. A result for one enzyme does not apply to every medicine.

Genes you may see on a report

Gene / pathway	Examples of relevance
CYP2D6	Some antidepressants, antipsychotics, atomoxetine, and codeine/tramadol activation.
CYP2C19	Some antidepressants, including citalopram/escitalopram and certain tricyclics; clopidogrel activation.
CYP2C9	Warfarin and selected other drugs; interpretation depends on medication.
CYP3A4 / CYP3A5	Important metabolic pathway for many drugs; clinically actionable genetic findings are limited for many psychiatric medicines.
UGT1A4 / UGT2B7	Glucuronidation pathways; commercial psychiatric panel claims may exceed validated dosing evidence.
HLA-B*15:02 / HLA-A*31:01	Immune-related risk of severe skin reactions with certain antiseizure/mood-stabilizing drugs; not liver-metabolism genes.

What are the potential benefits?

- May identify a higher likelihood of adverse effects or unusual drug exposure for certain medications.
- Can support a dose adjustment or an alternative medication when there is an established gene–drug guideline.

- May be particularly useful after unexpected side effects, poor response, or when considering a medication with strong gene–drug evidence.
- Results usually remain relevant over a lifetime because inherited DNA generally does not change.

What are the limitations?

- A test cannot predict whether a medication will definitely work, whether symptoms will improve, or every side effect.
- Many medications have no established, clinically useful genetic dosing recommendation. Commercial “green/yellow/red” categories can oversimplify the evidence.
- Age, kidney or liver disease, other medications, smoking, diet, and medical conditions may matter more than genetics.
- Some drugs inhibit or induce enzymes, temporarily making a genetically normal metabolizer behave differently (“phenoconversion”).
- Panels differ in genes and variants tested; some variants are missed, and performance may vary across ancestral backgrounds.
- Results do not diagnose depression, bipolar disorder, dementia, or other psychiatric conditions.

Special considerations for older adults and dementia

Medication sensitivity in later life often reflects multiple factors: reduced physiologic reserve, frailty, dehydration, kidney function, drug interactions, falls, and cognitive impairment. PGx may contribute useful information but cannot replace a careful medication review, “start low, go slow” when appropriate, and monitoring for benefit and harm.

For dementia-related agitation, a PGx result does not establish that an antipsychotic, antidepressant, or other medication is safe or effective. Risks such as sedation, falls, stroke, and increased mortality with antipsychotics in dementia-related psychosis still require individualized discussion.

What to expect: before, during, and after testing

- Before: Your clinician reviews current and past medications, side effects, treatment goals, family history, and whether testing would change a decision. Ask about insurance coverage and privacy.
- Sample: A cheek swab, saliva sample, or blood sample is collected. There is usually no special preparation.
- Results: Laboratory turnaround is often days to a few weeks, depending on the laboratory. The report may assign metabolizer categories for specific genes.
- Review: Your clinician interprets the report for each medication alongside your age, health, doses, other medicines, and symptoms. Do not change medication doses based on a color-coded report alone.
- Follow-up: Any treatment change still requires symptom and side-effect monitoring; blood levels or laboratory testing may be needed for some drugs.

Examples: where evidence is stronger — and where it is not

Medication situation	What testing can / cannot help with
Certain SSRIs and tricyclic antidepressants	CYP2C19 and/or CYP2D6 may inform dosing or selection for particular drugs.
Some antipsychotics	CYP2D6 may affect exposure to selected agents; relevance differs by drug and guideline.
Carbamazepine / oxcarbazepine	HLA testing may help identify risk for severe skin reactions in relevant populations; this is not a metabolism test.
Lamotrigine	Routine commercial enzyme panels do not reliably predict response or prevent serious rash; clinical titration remains essential.
Lithium / valproate	PGx is not a substitute for lithium levels, renal/thyroid tests, valproate-related monitoring, or clinical assessment.

Questions to ask the prescribing clinician

- Which specific medication decision might this test change?
- Is there a recognized CPIC, DPWG, or FDA-supported gene–drug recommendation for this medication?
- Could another medication, medical condition, or organ-function change explain my response?
- Will insurance cover the test, and who can access the genetic results?
- How will we monitor safety and benefit after interpreting the report?

Trusted educational resources

- Clinical Pharmacogenetics Implementation Consortium (CPIC): cpicpgx.org — evidence-based gene–drug guidelines.
- PharmGKB: pharmgkb.org — curated information about medication–gene relationships.
- MedlinePlus Genetics: medlineplus.gov/genetics — accessible explanations of genes and testing.
- U.S. Food and Drug Administration: fda.gov — pharmacogenetic association tables, labeling, and cautions about unvalidated claims.
- National Society of Genetic Counselors: nsgc.org — finding a genetic counselor when appropriate.

Take-home message: PGx testing can be one useful piece of the medication puzzle, especially for certain gene–drug pairs. It is not a “best medication” test and cannot replace clinical judgment, follow-up, or ordinary safety monitoring.

Educational information only. Decisions about genetic testing and medication changes should be made with a qualified healthcare professional.