HEALTH BASICS

Pharmacogenetics

Pharmacogenetics studies how inherited genetic differences can affect your response to medicines.

WHAT IT IS
How inherited genes influence responses to medicines

WHERE IT COMES UP
Medication choices, dosing decisions, and genetic test reports

WHY IT MATTERS
May support safer or more effective medicine choices

WHAT IT IS NOT
Not a disease or diagnosis

WHO TO ASK
Prescriber, pharmacist, or clinical genetics service

HOW IT IS CHECKED
Blood or cheek-swab genetic testing

Overview

Pharmacogenetics is the study of how inherited differences in your genes may affect your response to medicines. These differences can influence how quickly your body processes a drug, whether the drug is likely to work, or whether you have a higher chance of certain side effects.

Pharmacogenetics is not a disease or diagnosis. It is one source of information that may help with choosing or dosing some medicines.

The term pharmacogenomics is often used in a similar way. Pharmacogenetics usually refers to one or a few genes affecting a medicine. Pharmacogenomics can refer more broadly to many genes across the genome. In everyday healthcare, the terms may be used interchangeably.

Only some medicines have well-established gene-related guidance. For many medicines, genetic testing does not add useful information. Your age, kidney and liver function, other medicines, health conditions, and treatment goals can remain just as important.

How it works

Genes are sections of DNA that carry biological instructions. Most people have small differences called genetic variants . A variant is not automatically harmful. Some variants change the activity of proteins involved in processing or responding to medicines.

For example, the liver uses proteins called enzymes to break down many drugs. A genetic variant may cause an enzyme to work more slowly or more quickly than usual. This can change the amount of medicine or active drug product in your body.

A laboratory may use your genetic result to predict a metabolizer status , such as poor, intermediate, normal, rapid, or ultrarapid metabolizer. These labels describe expected enzyme activity. They do not describe your general metabolism, weight, digestion, or overall health.

Genes can affect medicines in several ways:

Testing usually uses blood or cells collected with a cheek swab. A targeted test may look for variants in one gene, while a panel looks at several genes. Different laboratories may test different variants.

A result does not guarantee that a medicine will work or cause a side effect. It changes an estimate of likelihood. Other medicines can also block or increase enzyme activity, sometimes making your actual response different from the genetic prediction.

- Changing how quickly a drug is activated or removed
- Changing how a drug interacts with its target
- Increasing the chance of a particular immune reaction
- Changing the likelihood that a usual dose will work as intended

Why it matters for your health

A pharmacogenetic result may help a clinician select a medicine, choose a starting dose, or decide whether closer monitoring is appropriate. The result is only useful when there is reliable evidence connecting that gene to the medicine being considered.

Examples of established drug-gene relationships include:

These examples do not mean everyone needs testing. Whether a test is useful depends on the medicine, your clinical situation, whether treatment has started, and whether the result would change a decision.

- CYP2C19 and clopidogrel: Some variants reduce activation of clopidogrel, an antiplatelet medicine used to reduce blood clots. This can affect how well it works in certain clinical situations.
- HLA-B and abacavir: The HLA-B*57:01 variant is linked to a serious immune reaction to abacavir, a medicine used for HIV.
- TPMT or NUDT15 and thiopurines: Reduced activity can increase the risk of severe bone marrow suppression from medicines such as azathioprine, mercaptopurine, and thioguanine.
- CYP2D6 and codeine: Codeine must be converted into morphine to relieve pain. Very low conversion may limit pain relief, while unusually fast conversion can increase opioid toxicity.
- CYP2C9, VKORC1, and warfarin: Variants can contribute to differences in the dose needed for this blood-thinning medicine.
- Certain HLA variants and carbamazepine: Some variants are associated with severe skin reactions. Which variants are relevant can be connected to genetic ancestry.

What can go wrong

Problems can occur when a clinically important drug-gene relationship is missed or when a test result is treated as more certain than it is.

Related situations include:

A "normal" result does not rule out side effects. A higher-risk result does not mean a reaction will definitely happen. Genetic testing also does not replace routine blood tests, symptom review, or other monitoring required for a medicine.

Contact your prescriber or pharmacist promptly about a new or worsening side effect, poor treatment response, or uncertainty about a test result. This is usually a routine or same-day medication review rather than an emergency.

Seek urgent medical care for a spreading rash with blisters, skin peeling, mouth sores, or fever. Call emergency services for trouble breathing, swelling of the face or tongue, collapse, unresponsiveness, or severe uncontrolled bleeding.

- Treatment failure: A drug may not be activated properly or may be removed too quickly.
- Drug toxicity: Slow removal or unusually fast activation may lead to excessive drug effects.

- Severe hypersensitivity: Certain HLA variants can raise the chance of specific immune reactions, such as abacavir hypersensitivity.
- Bone marrow suppression: Thiopurines can sharply reduce blood cell production in people with low TPMT or NUDT15 activity.
- Serious skin reactions: Stevens-Johnson syndrome and toxic epidermal necrolysis are rare but severe reactions linked to certain medicine and HLA combinations.
- Incorrect interpretation: A consumer test may not include all relevant variants or may make claims that are not supported for a particular medicine.

What you can do

Do not stop, start, or change the dose of a medicine based only on a genetic report. The result must be matched to the exact medicine, reason for treatment, and other factors affecting your care.

If testing is being discussed, useful questions include:

Keep a copy of the full laboratory report, including the gene, exact variant or genotype, laboratory name, and test date. A phone app summary or colour-coded result may leave out important details.

Tell future prescribers and pharmacists that you have a pharmacogenetic result. The DNA result itself usually does not change, but scientific interpretation and prescribing guidance may change over time.

If you are not taking or considering a medicine connected to the result, there may be nothing to do now. Keep the report for future medication decisions.

- Which gene and medicine is this test meant to assess?
- Would the result change the medicine, dose, or monitoring?
- What variants does the laboratory test include?
- Will insurance or another payment system cover the test?
- How will the result be stored and shared?

Common questions

Can pharmacogenetic testing find the best medicine for me?

Usually not by itself. It may help rule out an option, adjust a dose, or identify a medicine that

needs closer monitoring. It cannot compare every possible treatment or predict your full response.

Is this the same as a test for an inherited disease?

No. Pharmacogenetic testing focuses on medicine response. Some results may have implications for biological relatives, but they generally do not diagnose a disease.

Can a home DNA test provide reliable medication advice?

Its usefulness depends on the variants tested, laboratory quality, and evidence behind each claim. Consumer reports may be incomplete. A clinical laboratory may need to confirm a result before it is used for prescribing.

Do I need to repeat the test?

Your inherited DNA usually remains the same. Repeat testing may be considered if an earlier test covered too few variants, used a different method, or did not assess a newly relevant gene.

Does ancestry affect testing?

Some variants are more common in people with particular genetic ancestries, but appearance or self-identified ethnicity cannot show which variants you carry. Tests should not be interpreted from ancestry alone.

Will my relatives have the same result?

They may share some variants, but not necessarily the same combination. Your report should not be used to make medication decisions for another person.

Sources

- MedlinePlus Genetics: What is pharmacogenomics? - US National Library of Medicine
- US Food and Drug Administration: Table of Pharmacogenetic Associations
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