TEST

Pharmacogenetic testing

Pharmacogenetic testing looks for inherited DNA differences that may affect your response to certain medicines.

WHAT IT MEASURES
Inherited DNA variants that can change medicine response

SAMPLE
Cheek swab, saliva, or blood

FASTING NEEDED?
No

HOW LONG IT TAKES
Usually 5 to 15 minutes

RESULTS IN
Several days to weeks, varying by laboratory

USED FOR
Choosing or dosing certain medicines

MAIN LIMITATION
Many results do not reliably predict individual response

Overview

Pharmacogenetic testing examines parts of your DNA that can affect how your body responds to medicines. DNA is the genetic material carried in most cells.

Some genetic differences change how quickly your body breaks down a medicine. Others affect how a medicine reaches its target or whether it is more likely to cause a particular side effect.

The test may help with choosing or dosing certain medicines. However, it does not predict your response to every medicine, and it cannot guarantee that a treatment will work or be free of side effects.

Pharmacogenetics usually focuses on one or several genes related to a particular medicine. "Pharmacogenomics" is a broader term that may describe testing many genes at once. In practice, the terms are often used in similar ways.

Most results reflect inherited DNA and usually remain the same throughout life. Their interpretation can change as new evidence becomes available.

Why the test is ordered

The test may be ordered before you start a medicine when a genetic result could affect treatment. It may also be considered after an unexpected side effect, an unusually strong response, or a lack of benefit.

Examples of medicine-gene relationships include:

These examples do not mean that everyone taking these medicines needs testing. Whether a result is useful depends on the medicine, available evidence, treatment alternatives, and your clinical circumstances.

Testing is not intended to explain every side effect. Kidney or liver function, other medicines, dose, illness, age, pregnancy, and adherence can all influence treatment response.

- CYP2C19 and clopidogrel: some variants reduce activation of this antiplatelet medicine
- CYP2D6 and codeine: some variants affect how codeine is converted into morphine
- DPYD and fluoropyrimidines: some variants increase the risk of serious toxicity from certain cancer medicines
- TPMT or NUDT15 and thiopurines: some variants increase the risk of severe effects on blood cells
- Specific HLA variants and certain medicines: some variants are linked to a higher risk of severe immune reactions

Who should have the test

Pharmacogenetic testing may be useful if you are about to take a medicine with a well-supported gene-medicine relationship. It may also be considered if you have had an unusual response to a relevant medicine.

Testing can sometimes help when several reasonable treatment options are available. A result may support one option, but it should be considered alongside your medical history and current health.

A broad panel is not automatically better than a targeted test. Panels may include genes with limited evidence or genes unrelated to medicines you are likely to use. Results from direct-to-consumer tests may also cover only selected variants.

Your ancestry can affect which genetic variants are more common, but race or ethnicity cannot replace testing. People from the same population do not all carry the same variants.

How to prepare

You do not need to fast. You can usually take your usual medicines unless you have been given different instructions.

For a cheek swab or saliva sample, the laboratory may ask you not to eat, drink, smoke, chew gum, or brush your teeth for a short period beforehand. Follow the collection kit instructions because requirements differ.

Tell the testing service if you have had a bone marrow or stem cell transplant. Blood cells after a donor transplant may carry the donor's DNA, so a blood sample may not represent your inherited genetic makeup.

Also mention a recent blood transfusion. Modern transfusions contain relatively few white blood cells, but the laboratory may still need this information when choosing or interpreting a sample.

What happens during the test

A cheek swab is rubbed firmly along the inside of your mouth to collect cells. This usually takes less than a minute, although several swabs may be needed.

For a saliva test, you spit into a collection tube. It may take a few minutes to provide enough saliva.

For a blood test, a healthcare professional places a needle into a vein, usually in your arm, and collects a small tube of blood.

The sample is sent to a laboratory. DNA is extracted and examined for selected variants. Some tests check one gene, while panels check many genes.

Understanding your results

Pharmacogenetic reports may list specific variants, combinations called alleles , or a predicted response category. Some laboratories use "star allele" names, such as ` 1` or ` 2`.

A report may describe how your body is expected to process a medicine. Terms can include poor, intermediate, normal, rapid, or ultrarapid metabolizer. These categories are gene-specific and do not apply equally to all medicines.

A result might suggest:

A "normal" result does not mean that you cannot have side effects or treatment failure. It may

only mean that the tested variants were not found.

There is no universal normal range for this test. Reporting and interpretation vary by laboratory. Clinical meaning may also vary with age, sex, pregnancy status, medicines, and the wider clinical context.

Do not start, stop, or change a prescribed medicine based only on a test report. Some medicines can cause harm if stopped suddenly.

- using the medicine in the usual way
- considering a different starting dose
- monitoring more closely
- considering another medicine
- confirming the result with additional testing

What affects the results

Your inherited DNA usually does not change with food, exercise, time of day, illness, or most medicines. However, several factors can make a result incomplete or misleading without anything being wrong with the laboratory.

The test can only find variants it was designed to detect. A limited panel may miss rare variants, gene deletions, extra gene copies, or complex rearrangements. This is especially important for genes such as CYP2D6 , which can have complicated structural changes.

Poor-quality samples can cause problems. Food residue, too little saliva, contamination, or mixing up samples may prevent testing or produce an unreliable result.

A donor stem cell or bone marrow transplant can make blood-based results reflect the donor's DNA. Recent transfusion should also be reported, although its effect depends on the blood product and testing method.

Medicines do not usually change your genotype, but they can change what the result means in practice. A medicine that blocks or increases the activity of a drug-processing enzyme can make your actual response differ from your genetically predicted response.

Kidney and liver function, age, pregnancy, smoking, diet, other illnesses, and interactions between medicines may also affect drug levels. These factors can outweigh or modify a genetic effect.

Accuracy and limitations

A well-validated laboratory test can identify the particular variants it examines accurately. However, not every laboratory examines the same variants or uses methods that can detect complex gene changes.

The test can miss an important genetic difference if that variant is outside the panel. In that situation, the report may suggest usual activity even though an untested variant affects medicine response.

A result may also predict increased risk when you would have tolerated the medicine. Genetic risk does not mean that a reaction will definitely happen. Conversely, you may have a side effect despite having no identified high-risk variant.

Sample contamination, technical errors, and incorrect allele assignment can occasionally produce a wrong result. Confirmation may be appropriate when a result is unexpected or would lead to a major treatment decision.

Pharmacogenetic testing is not a replacement for monitoring how you feel or for tests that measure the medicine's effects. Drug-level testing, blood counts, liver or kidney tests, clotting tests, or other monitoring may be more useful for detecting a current problem.

Evidence is much stronger for some gene-medicine pairs than for others. A commercial report may include associations that have not been shown to improve treatment decisions.

Risks and complications

A cheek swab or saliva collection has no significant physical risk.

A blood draw may cause brief pain, a small bruise, or light-headedness. These effects are usually minor and short-lived.

Genetic information can have privacy and family implications. Your result may indirectly suggest information about biological relatives, and legal protections for genetic information differ by location and type of insurance or employment.

What happens next

The result should be considered with your current medicines, health conditions, treatment goals, and previous responses. Routine follow-up may include discussing whether the result changes a medicine choice, dose, or monitoring plan.

Keep a copy of the report. Because inherited results usually remain relevant, they may be

useful if you need a related medicine later. Ask whether the full laboratory report can be added to your health record rather than recording only a broad category.

Interpretation may change as research and prescribing guidance develop. An older result may need to be reviewed again rather than repeating the DNA test.

Pharmacogenetic testing does not create an emergency by itself. Seek urgent medical help for possible severe medicine reactions, such as trouble breathing, swelling of the face or throat, fainting, or a widespread blistering or peeling rash. Do not wait for a genetic result if you are acutely unwell.

Common questions

Can I drink coffee before the test?

Yes. Coffee does not change your DNA. If you are providing saliva, follow the kit's instructions about how long to avoid drinks before collection.

Does the time of day matter?

No. Your inherited genetic variants are not affected by the time of day.

Can I exercise beforehand?

Yes. Exercise does not change the pharmacogenetic result. It may affect other blood tests collected at the same appointment, so follow any separate instructions.

Will this test tell me which medicine will work best?

Usually not by itself. It may indicate that certain medicines or doses are more or less suitable, but your health, other medicines, and treatment goals remain important.

Do I need to be tested again?

Usually not for the same inherited variants. You may need updated interpretation or additional testing if the first panel did not examine a relevant gene or variant.

Can I use a home testing kit?

Some samples can be collected at home. Check that testing is performed by an appropriately accredited laboratory and that the report clearly states which variants were examined. Consumer reports may need confirmation before treatment is changed.

Sources



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