TEST

Cystic fibrosis carrier screening

A genetic test that checks whether you carry a cystic fibrosis gene variant

WHAT IT MEASURES
Disease-causing changes in the CFTR gene

SAMPLE
Blood, saliva, or cheek swab

FASTING NEEDED?
No

HOW LONG IT TAKES
Usually less than 10 minutes

RESULTS IN
Usually 1-3 weeks, varying by laboratory

USED FOR
Carrier screening before or during pregnancy

MAIN LIMITATION
A negative result cannot exclude every CFTR variant

Overview

Cystic fibrosis carrier screening is a genetic test that looks for disease-causing changes, called variants, in the CFTR gene.

Cystic fibrosis is an inherited condition that mainly affects the lungs and digestive system. A carrier has one disease-causing CFTR variant and usually does not have cystic fibrosis.

The condition follows an autosomal recessive inheritance pattern. A child usually develops cystic fibrosis only if they inherit a disease-causing variant from each biological parent.

If both biological parents are carriers, each pregnancy has:

These chances apply separately to every pregnancy. Carrier screening estimates reproductive risk; it does not diagnose cystic fibrosis in a pregnancy or child.

- A 1 in 4 chance that the child will have cystic fibrosis
- A 1 in 2 chance that the child will be a carrier
- A 1 in 4 chance that the child will inherit neither variant

Why the test is ordered

The test is usually ordered before pregnancy or early in pregnancy. Knowing your carrier status can help you understand the chance of having a child with cystic fibrosis and decide whether to consider further testing.

It may also be ordered when:

Carrier screening differs from newborn screening, which checks a baby after birth for signs that cystic fibrosis may be present.

- A biological relative has cystic fibrosis or is a known carrier
- Your reproductive partner has a disease-causing CFTR variant
- You are using donated eggs, sperm, or embryos
- A fertility clinic offers broader carrier screening
- Previous testing found a CFTR variant that needs clarification

Who should have the test

Professional guidance in the United States supports offering cystic fibrosis carrier screening to people who are pregnant or considering pregnancy. Testing is optional.

It may be especially useful when you or your partner has a family history of cystic fibrosis. If possible, obtain the affected relative's genetic report. Testing for the known family variant can be more informative than using only a standard panel.

If one partner is a carrier, the other biological parent can be tested. During pregnancy, both may be tested at the same time to reduce delays.

Because inherited DNA does not normally change, repeat screening is often unnecessary. However, newer tests may examine more variants than older panels. A genetics professional can compare your previous report with current options.

How to prepare

You do not need to fast. Food, coffee, prescribed medicines, exercise, and the time of day do not usually affect inherited DNA testing.

Tell the testing service if you have had a bone marrow or stem cell transplant. Blood and saliva may contain donor DNA, causing the result to reflect the donor rather than you. A different

sample, such as cultured skin cells, may be needed.

Ask whether insurance requires prior authorization and whether genetic counseling is included. Costs vary by laboratory and test type.

For a saliva sample, follow the laboratory's instructions. You may need to avoid eating, drinking, smoking, chewing gum, or brushing your teeth for a short time before collection.

What happens during the test

The sample is usually collected by:

Collection usually takes less than 10 minutes. A genetics laboratory then examines selected CFTR variants or, with broader testing, much of the gene.

Some tests check only a panel of common variants. Others use sequencing, and a separate method may be needed to detect large missing or duplicated sections of DNA.

- Drawing a small tube of blood from a vein
- Spitting into a collection tube
- Rubbing a swab inside your cheek

Understanding your results

Results are provided as a written genetic interpretation rather than a numerical reference range.

A negative result means the test did not find the disease-causing variants it was designed to detect. It lowers, but does not eliminate, the chance that you are a carrier.

A positive or carrier result means one disease-causing or likely disease-causing CFTR variant was found. This usually does not mean you have cystic fibrosis. Testing your reproductive partner can help estimate the risk for a pregnancy.

A variant of uncertain significance means there is not enough evidence to know whether a change causes disease. Laboratories do not always report these during routine screening. It should not be treated as a confirmed carrier result without specialist interpretation.

Rarely, testing finds two CFTR variants. Their meaning depends on the variants and whether they are on the same or opposite copies of the gene. A clinician or genetics professional should review this result; it is not by itself a diagnosis.

What affects the results

Everyday activities such as fasting, exercise, drinking coffee, or taking usual medicines do not change inherited CFTR variants.

Important factors include:

These factors affect how reassuring a negative result is.

- The test panel: Limited panels miss variants they do not include
- Ancestry: Variant frequency differs among populations
- Family information: A negative result may be less reassuring if a relative's exact variant is unknown
- Sample quality: Too little DNA, saliva contamination, or labeling problems may require another sample
- Bone marrow or stem cell transplant: Blood and saliva may contain donor DNA
- Recent blood transfusion: Standard red-cell transfusions usually have little effect, but tell the laboratory about any recent transfusion
- Testing method: Some methods may miss large deletions, duplications, or variants in unexamined regions

Accuracy and limitations

A positive finding from a validated laboratory is generally reliable, although sample errors and later changes in variant classification are possible. Some variants have variable effects, so a result may not predict how severely a child would be affected.

Targeted panels can miss variants outside the panel. Sequencing detects more variants but may still miss some large DNA changes or variants in regions it does not examine.

A negative result cannot rule out carrier status. Your report may give a residual risk, which estimates the chance that you remain a carrier based on the test, ancestry data, and family history.

Carrier screening is not the preferred test for diagnosing cystic fibrosis in someone with symptoms. Diagnosis may involve sweat chloride testing and diagnostic genetic evaluation. During pregnancy, chorionic villus sampling or amniocentesis can test fetal cells when diagnostic information is wanted.

Risks and complications

A blood draw may cause brief discomfort, bruising, or light-headedness. Saliva and cheek-swab collection rarely cause physical problems, although an inadequate sample may need to be repeated.

Results can have implications for relatives and reproductive decisions. Genetic counseling can help you understand these issues.

What happens next

A negative result usually needs no urgent follow-up. Keep a copy of the report because it identifies the variants and methods used.

If you are a carrier, next steps may include testing your reproductive partner and speaking with a genetic counselor. If both partners carry disease-causing CFTR variants, options may include testing during pregnancy, testing after birth, in vitro fertilization with embryo testing, using donor eggs or sperm, or proceeding without further testing.

These choices are personal. Screening does not require a particular decision.

A carrier result is not an emergency. During pregnancy, however, follow-up can be time-sensitive because some tests are available only at certain stages. Contact your pregnancy care team promptly rather than waiting for a later routine visit.

Common questions

Can I drink coffee before the test?

Yes. Coffee does not affect inherited DNA. For a saliva sample, follow instructions about when to stop drinking.

Does the time of day matter?

No. The result is the same at any time.

Can I exercise beforehand?

Yes. Exercise does not change the result.

Can I have the test while pregnant?

Yes. It tests your DNA, not the fetus's DNA.

If my result is negative, can my child still have cystic fibrosis?

The chance is lower but not zero because screening cannot detect every disease-causing variant. The other biological parent's result also matters.

Is this the same as a cystic fibrosis sweat test?

No. A sweat test measures salt in sweat to help diagnose cystic fibrosis. Carrier screening examines DNA to estimate reproductive risk.

Sources

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