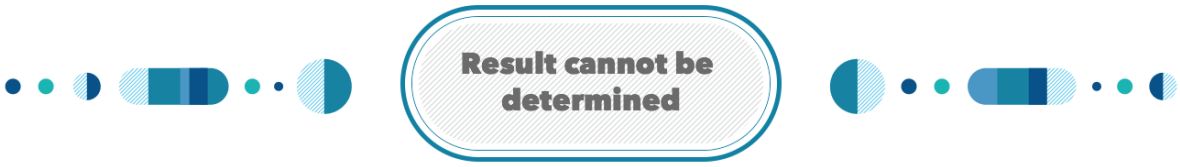
Cystic Fibrosis

Cystic fibrosis is a rare genetic disorder characterized by impaired lung and digestive function. A person must have two variants in the CFTR gene in order to have this condition.

Your result for this test cannot be determined.

We may not always be able to report a result for this test. This can happen if there is a test error or if a person has two copies of a variant tested.



If you are concerned about this report, please consult with a healthcare professional about additional testing.

How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Carrier Status tutorial

See Scientific Details

Intended Uses

- Tests for **multiple variants** in the CFTR gene.
- To identify carrier status for cystic fibrosis.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.

Important Ethnicities

- This test is most relevant for people of **European** , **Hispanic/Latino** , and **Ashkenazi Jewish** descent.

About Cystic Fibrosis

When symptoms develop

Symptoms typically develop during infancy.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications such as lung infections and malnutrition.

Typical signs and symptoms

- Chronic cough
- Lung infections
- Pancreatic insufficiency
- Malnutrition
- Infertility in males

Ethnicities most affected

This condition is most common in people of European, Ashkenazi Jewish, and Hispanic/Latino descent.

Read more at

Genetics Home Reference

GeneReviews

Mayo Clinic

Consider talking to a healthcare professional if you are concerned about this report.

If you think you might have symptoms or if this condition runs in your family, consult with a healthcare professional.

Print report

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC

Learn more about this condition and connect with support groups.

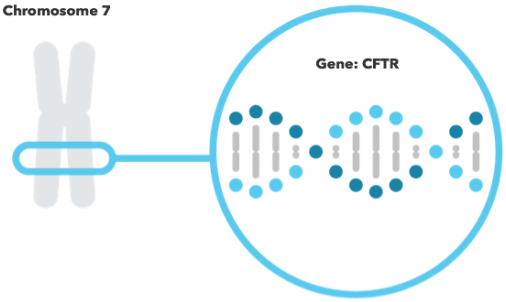
Learn more

Cystic fibrosis is caused by variants in the CFTR gene.

CFTR

The CFTR gene contains instructions for making a protein called cystic fibrosis transmembrane conductance regulator. This protein helps control the salt and water balance of certain organs by allowing chloride ions to pass in and out of cells. Certain variants in the CFTR gene disrupt this function, causing the lungs, pancreas, and other organs to produce abnormally thick mucus. This mucus can clog the respiratory tract, leading to signs and symptoms of cystic fibrosis.

Read more at Genetics Home Reference



Your result cannot be determined.

Variants Detected	View All Tested Markers
0	27

Marker Tested	Your Genotype*		Additional Information
DeltaF508 Gene: CFTR Marker: i3000001	**CTT** Typical copy from one of your parents	**CTT** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
Deltal507 Gene: CFTR Marker: i4000292	**ATC** Typical copy from one of your parents	**ATC** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
G85E Gene: CFTR Marker: i4000294	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
R334W Gene: CFTR Marker: i4000296	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
R347P/H Gene: CFTR Marker: i4000297	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
A455E Gene: CFTR Marker: i4000291	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
V520F Gene: CFTR Marker: i4000299	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [1] | ClinVar
G542X Gene: CFTR Marker: i4000300	**Not determined**		> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
S549N Gene: CFTR Marker: i4000301	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [1] | ClinVar
G551D Gene: CFTR Marker: i4000305	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
R553X Gene: CFTR Marker: i4000306	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar
R560T Gene: CFTR Marker: i4000307	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> Biological explanation > Typical vs. variant DNA sequence(s) > Percent of 23andMe customers with variant > References [5] | ClinVar

R1162X Gene: CFTR Marker: i4000308	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
W1282X Gene: CFTR Marker: i4000309	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
N1303K Gene: CFTR Marker: i4000311	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
394delTT Gene: CFTR Marker: i4000313	**TT** Typical copy from one of your parents	**TT** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1] | ClinVar
621+1G>T Gene: CFTR Marker: i4000314	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
711+1G>T Gene: CFTR Marker: i4000315	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
1078delT Gene: CFTR Marker: i4000316	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
1717-1G>A Gene: CFTR Marker: i4000317	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
1898+1G>A Gene: CFTR Marker: i4000318	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
3120+1G>A Gene: CFTR Marker: i4000321	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
3659delC Gene: CFTR Marker: i4000322	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
3905insT Gene: CFTR Marker: i4000324	**(–)** Typical copy from one of your parents	**(–)** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1] | ClinVar
3849+10kbC>T Gene: CFTR Marker: i4000325	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar
2184delA Gene: CFTR Marker: i4000319	**A** Typical copy from one of your parents	**A** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [5] | ClinVar

3876delA
Gene: CFTR
Marker: i4000323

A
Typical copy from one of your parents

A
Typical copy from your other parent

> **Biological explanation**
> **Typical vs. variant DNA sequence(s)**
> **Percent of 23andMe customers with variant**
> **References [1]** | ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Cystic Fibrosis is indicated for the detection of 28 variants in the CFTR gene. This test is intended to be used to determine carrier status for cystic fibrosis in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Ashkenazi Jewish, European, and Hispanic/Latino descent.

Special Considerations

- Symptoms of cystic fibrosis may vary depending on the variants involved.
- ACMG recommends carrier testing for cystic fibrosis for people of all ethnicities considering having children. This test includes 21 of the 23 variants recommended for testing by ACMG.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

Ashkenazi Jewish	95%	[3 , 5]
European	89%	[3 , 5]
Hispanic/Latino	73%	[3 , 5]
African American	65%	[3 , 5]
Asian	55%	[3 , 5]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 1,514 samples with known variant status. 1,514 out of 1,514 genotype results were correct. About 1 in 610 samples may receive a **Not Determined** result for one or more variants included in this test. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. 510(k): Illumina MiSeqDx Cystic Fibrosis Clinical Sequencing Assay
2. American College of Obstetricians and Gynecologists Committee on Genetics. (2011). "ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis." Obstet Gynecol. 117(4):1028-31.
3. Bobadilla JL et al. (2002). "Cystic fibrosis: a worldwide analysis of CFTR mutations--correlation with incidence data and application to screening." Hum Mutat. 19(6):575-606.
4. Moskowitz SM et al. (1993). " *CFTR* -Related Disorders"
5. Watson MS et al. (2004). "Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel." Genet Med. 6(5):387-91.