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.

Overview | Scientific Details

Jamie, you **do not have the variants** we tested.

You could still have a variant not covered by this test.

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.

You are likely not a carrier.

This result is relevant for you because you have **European** ancestry.

We ruled out the most common variants for cystic fibrosis in people of European descent.

You still have a chance of being a carrier for cystic fibrosis.

You may still have up to a **1 in 230 chance** of carrying a variant not covered by this test.

See Scientific Details

About Cystic Fibrosis

When symptoms develop

Symptoms typically develop during infancy.

Typical signs and symptoms

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

Ethnicities most affected

This condition is common in all ethnicities, but most common in people of European descent.

How it's treated

There is currently no known cure. For people with certain CFTR variants, medications can improve the function of the CFTR protein or increase the amount of CFTR protein, which helps treat the underlying cause of the condition. Other treatments focus on managing symptoms and preventing complications such as lung infections and malnutrition.

Read more at: MedlinePlus GeneReviews Mayo Clinic

Consider talking to a healthcare professional if you are concerned about your results.

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

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

Learn more

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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.

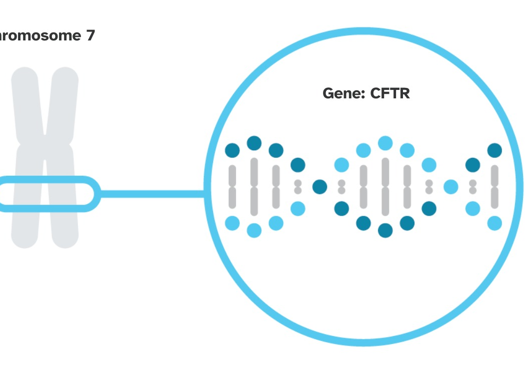
Overview | Scientific Details

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 MedlinePlus



You have no variants detected by this test.

Variants Detected | View All Tested Markers

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 [6] | ClinVar
DeltaI507 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 [6] | 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 [6] | 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 [6] | 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 [1, 6] | 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 [6] | 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	**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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [1] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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 [6] | 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
2789+5G>A Gene: CFTR Marker: i4000320	**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 [6] | 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 Interpretation

This report provides an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk**.

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

View technical article on estimating post-test carrier risk.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of Ashkenazi Jewish, European, Hispanic/Latino, African American, and Asian descent only.

- For people with partial ethnicity from one or more groups mentioned above, post-test carrier risk depends on the exact mixture in the person's background.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

Ashkenazi Jewish	1 in 390	[3]
European	1 in 230	[3]
Hispanic/Latino	1 in 210	[3]
African American	1 in 170	[3]
Asian	1 in 210	[3]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Cystic Fibrosis is indicated for the detection of 29 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 22 of 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	94%	[2, 6]
European	89%	[2, 6]
Hispanic/Latino	73%	[2, 6]
African American	65%	[2, 6]
Asian	55%	[2, 6]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

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. 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.
3. Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.
4. Gramegna A et al. (2020). "From Ivacaftor to Triple Combination: A Systematic Review of Efficacy and Safety of CFTR Modulators in People with Cystic Fibrosis." Int J Mol Sci. 21(16):5882.
5. Ong T et al. (2001). "Cystic Fibrosis and Congenital Absence of the Vas Deferens." [Accessed Dec 8, 2020].
6. 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.

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
March 2, 2018	The variant 2789+5G>A (i4000320) was added to the report. Customers who have this variant will see this variant detected in their result, and see updated content in their report. The carrier detection rate was updated for customers who self-report having Ashkenazi Jewish ancestry. The chances of still being a carrier were also updated for customers with no variants detected who self-report having Ashkenazi Jewish ancestry. The chances of still being a carrier were updated for customers with no variants detected who self-report having European or African American ancestry.
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for one or more of the following genetic markers may see a genotype at these markers: i4000291, i4000294, i4000296, i4000300, i4000301, i4000305, i4000307, i4000308, i4000311, i4000314, i4000315, i4000317, i4000318, i4000321, i4000323, i4000324, i4000325. This may also update the overall report result for these customers.
Oct. 21, 2015	Cystic Fibrosis report created.

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