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Beta Thalassemia and Related Hemoglobinopathies

Beta thalassemia is a genetic disorder characterized by anemia and fatigue as well as bone deformities and organ problems. A person must have two variants in the HBB gene in order to have this condition.

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

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

0 variants detected
in the HBB gene

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 HBB gene.
- To identify carrier status for beta thalassemia and related hemoglobinopathies.

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 **Cypriot, Greek, Italian, and Sardinian** descent.

You are likely not a carrier.

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

We ruled out the most common variants for beta thalassemia in people of Cypriot descent.

You still have a chance of being a carrier for beta thalassemia.

You may still have up to a **1 in 59 chance** of carrying a variant not covered by this test.

See Scientific Details

About Beta Thalassemia and Related Hemoglobinopathies

Also known as: Cooley's Anemia, Mediterranean Anemia

When symptoms develop

Symptoms typically develop any time from late infancy (severe form) into adulthood (intermediate form).

How it's treated

Treatment focuses on managing symptoms and preventing complications. Some individuals may require frequent blood transfusions.

Typical signs and symptoms

- Anemia
- Fatigue
- Enlarged liver and spleen
- Poor growth and weight gain
- Bone deformities
- Iron buildup in multiple organs

Ethnicities most affected

This condition is most common in people of Mediterranean, Middle Eastern, North African, Transcaucasian, Central Asian, South Asian, and Southeast Asian descent.

Read more at

Genetics Home Reference

GeneReviews

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.

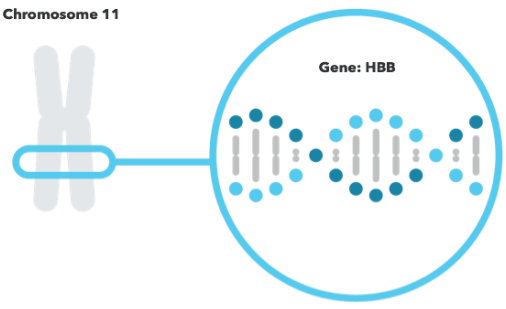
Learn more

Beta thalassemia and related hemoglobinopathies are caused by variants in the HBB gene.

HBB

The HBB gene contains instructions for making a protein called beta-globin. This protein is part of a larger protein called hemoglobin that is found in red blood cells. Hemoglobin transports oxygen from the lungs to all other cells of the body. Certain variants in HBB alter the structure of hemoglobin, making it defective in transporting oxygen.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	10

Marker Tested	Your Genotype*		Additional Information
-29A>G Gene: HBB Marker: rs34598529	**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** [4 , 15 , 18 , 23] | ClinVar
IVS1-(-1)G>C Gene: HBB Marker: rs33960103	**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** [25 , 35] | ClinVar
IVS1-5G>C Gene: HBB Marker: rs33915217	**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** [9 , 14 , 21 , 25 , 27 , 33] | ClinVar
IVS1-6T>C Gene: HBB Marker: rs35724775	**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** [3 , 5 , 9 , 27 , 31 , 33 , 36] | ClinVar
IVS1-110G>A Gene: HBB Marker: rs35004220	**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** [3 , 5 , 7 , 9 , 27 , 29] | ClinVar
IVS2-654C>T Gene: HBB Marker: rs34451549	**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 , 10 , 21 , 22] | ClinVar
IVS2-745C>G Gene: HBB Marker: rs34690599	**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 , 9 , 27 , 30 , 33] | ClinVar
W15X Gene: HBB Marker: rs63750783	**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** [1 , 5 , 16 , 21 , 26 , 34] | ClinVar
Q39X Gene: HBB Marker: rs11549407	**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** [3 , 5 , 13 , 26 , 34] | ClinVar
HbC Gene: HBB Marker: rs33930165	**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** [11 , 12 , 19] | 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 Sardinian, Cypriot, Italian, Greek, and Turkish 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

Sardinian	1 in 250	[24]
Cypriot	1 in 59	[20]
Italian, particularly from Sicily	1 in 61	[2]
Greek	1 in 37	[32]
Turkish	1 in 65	[8]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Beta Thalassemia and Related Hemoglobinopathies is indicated for the detection of 10 variants in the HBB gene. This test is intended to be used to determine carrier status for beta thalassemia in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Sardinian, Cypriot, Italian, and Greek descent.

Special Considerations

- Symptoms of beta thalassemia may vary between people with the condition depending on the variants involved.
- Carrier screening for beta thalassemia and related hemoglobinopathies is recommended by ACOG for people of African, Southeast Asian, and Mediterranean descent considering having children.

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.

Sardinian	97%	[17]
Cypriot	90% (averaged between Greek and Turkish Cypriot)	[17]
Italian, particularly from Sicily	82%	[17]
Greek	75%	[17]
Turkish	66%	[17]
Balkan	Albanian: 80%Macedonian: 72%Croatian: 41%	[17]
South Asian	Bangladesh: 70%Maharashtran: 60%Azerbaijan: 42%Pakistani: 42%Punjabi: 32%Pathan: 20%	[17]
Southeast Asian	Indonesian: 73%Taiwanese: 47%Malaysian: 38%Singapore: 35%Thai: 11%	[17]

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.

North African	Algerian: 61%Egyptian: 54%Tunisian: 50%	[17]
Middle Eastern	Oman: 64%United Arab Emirates: 63%Saudi Arabia: 60%Lebanon: 51%Jordan: 48%Bahrain: 44%Syria: 41%Yemen: 40%Kuwait: 33%Iran: 29%	[17]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for samples with known variant status. 461 out of 461 genotype results were correct. About 1 in 11,200 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.

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