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.

Overview | Scientific Details

Jamie, 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.
- Does **not cover** other types of thalassemia.

Important Ethnicities

- This test is most relevant for people of **Cypriot, Greek, Italian, and Sardinian** descent.

You are likely not a carrier.

This result may be less relevant for you because the variants that cause beta thalassemia are less common in people of your ethnicity.

We ruled out the tested variants for beta thalassemia.

These variants are most common in people of **Sardinian, Cypriot, Italian, or Greek** descent.

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

We cannot estimate your chances because sufficient data is not available for your ethnicity.

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).

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, Southeast Asian, and Southern Chinese descent.

How it's treated

Treatment focuses on managing symptoms and preventing complications, with some individuals requiring frequent blood transfusions. Others may benefit from curative hematopoietic stem cell transplants. In addition, scientists are currently working on more treatment options for this condition.

Read more at: MedlinePlus 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

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Overview | Scientific Details

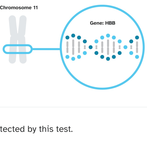
Beta thalassemia and related hemoglobinopathies are caused by variants in the HBB gene.

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 MedlinePlus®

Chromosome 11



You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*			Additional Information
-29A>G Gene: HBB Marker: rs34598529	T Typical copy from one of your parents	T Typical copy from one of your parents	T Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [4, 17, 26, 26] ClinVar®
IVS1-(-13)G>C Gene: HBB Marker: rs33960103	C Typical copy from one of your parents	C Typical copy from one of your parents	C Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [29, 41] ClinVar®
IVS1-5G>C Gene: HBB Marker: rs33915217	C Typical copy from one of your parents	C Typical copy from one of your parents	C Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [9, 15, 24, 29, 32, 39] ClinVar®
IVS1-6T>C Gene: HBB Marker: rs35724775	A Typical copy from one of your parents	A Typical copy from one of your parents	A Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [3, 5, 9, 32, 36, 39, 43] ClinVar®
IVS1-110G>A Gene: HBB Marker: rs35664220	C Typical copy from one of your parents	C Typical copy from one of your parents	C Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [3, 5, 7, 9, 32, 34] ClinVar®
IVS2-654C>T Gene: HBB Marker: rs34451549	G Typical copy from one of your parents	G Typical copy from one of your parents	G Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [6, 16, 24, 25] ClinVar®
IVS2-745C>G Gene: HBB Marker: rs34698099	G Typical copy from one of your parents	G Typical copy from one of your parents	G Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [5, 9, 32, 35, 39] ClinVar®
W15X Gene: HBB Marker: rs33958763	C Typical copy from one of your parents	C Typical copy from one of your parents	C Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [5, 16, 24, 31, 40] ClinVar®
Q39X Gene: HBB Marker: rs11554487	G Typical copy from one of your parents	G Typical copy from one of your parents	G Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [3, 5, 14, 31, 40] ClinVar®
HbC Gene: HBB Marker: rs33930165	C Typical copy from one of your parents	C Typical copy from one of your parents	C Typical copy from one of your parents	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andme customers with variant References [12, 13, 21] 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 259	[27]
Cypriot	1 in 71	[22]
Italian, particularly from Sicily	1 in 61	[2, 16]
Greek	1 in 37	[38]
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 19 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 via complete blood count and hemoglobin electrophoresis for people of African, Southeast Asian, Mediterranean, Middle Eastern, and West Indian 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%	[19]
Cypriot	99% (averaged between Greek and Turkish Cypriot)	[19]
Italian, particularly from Sicily	82%	[19]
Greek	75%	[19]
Turkish	66%	[19]
Balkan	41-89%, depending on region of ancestry	[19]
South Asian	28-79%, depending on region of ancestry	[19]
Southeast Asian	11-73%, depending on region of ancestry	[19]
North African	59-61%, depending on region of ancestry	[19]
Middle Eastern	29-64%, depending on region of ancestry	[19]
Chinese, particularly from Southern China	5-36%, depending on region of ancestry	[37]

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. **Ahmed M et al. (2001). "The beta-globin genotype E121G/W15X (cd121GAA->CAA/cd15TGG->TGA) underlines Hb d/beta-(0) thalassaemia marked by domination of haemoglobin D." Ann Hematol. 80(11):629-33.** ^
2. **Amato A et al. (2014). "Carrier screening for inherited haemoglobin disorders among secondary school students and young adults in Latium, Italy." J Community Genet. 5(3):265-8.** ^
3. **Amselem S et al. (1988). "Determination of the spectrum of beta-thalassemia genes in Spain by use of dot-blot analysis of amplified beta-globin DNA." Am J Hum Genet. 43(1):95-100.** ^
4. **Antonarakis SE et al. (1984). "beta-Thalassemia in American Blacks: novel mutations in the "TATA" box and an acceptor splice site." Proc Natl Acad Sci U S A. 81(4):1154-8.** ^
5. **Aulehla-Scholz C et al. (1998). "Molecular basis of beta-globin gene mutations in Turkey: detection of rare mutations by direct sequencing." Hum Genet. 64(2):195-7.** ^
6. **Boonyawat B et al. (2014). "Molecular analysis of beta-globin gene mutations among Thai beta-thalassemia children: results from a single center study." Appl Clin Genet. 7:253-6.** ^
7. **Busslinger M et al. (1981). "Beta + thalassemia: aberrant splicing results from a single point mutation in an intron." Cell. 27(2 Pt 1):289-98.** ^
8. **Canatan D et al. (2006). "Hemoglobinopathy control program in Turkey." Community Genet. 9(2):124-6.** ^
9. **Chehab FF et al. (1987). "The molecular basis of beta-thalassemia in Lebanon: application to prenatal diagnosis." Blood. 69(4):1045.** ^
10. **Cheng TC et al. (1984). "beta-Thalassemia in Chinese: use of in vivo RNA analysis and oligonucleotide hybridization in systematic characterization of molecular defects." Proc Natl Acad Sci U S A. 81(7):2825-9.** ^

See all references

Change Log

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

Date	Change
Nov. 3, 2021	The carrier detection rate ranges were updated for Balkan, Middle Eastern, North African, South Asian, and Southeast Asian.
Sept. 13, 2018	Information specific to people of Chinese descent was added. Customers who self-report having Chinese ancestry may now see carrier frequency and carrier detection rate information specific to that ancestry.
March 2, 2016	The carrier frequency was updated for customers with no variants detected with the following self-reported ethnicities: Balkan, Middle Eastern, North African, South Asian, and Southeast Asian.
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: rs33915217, rs34598529, rs33758763. This may also update the overall report result for these customers.
Oct. 21, 2015	Beta Thalassemia and Related Hemoglobinopathies report created.

23andMe

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