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 50 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.

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.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>-29A&gt;G</td>
<td>T</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>T</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs4598529</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>IVS1-41G&gt;C</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>C</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs39660103</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>IVS1-5G&gt;C</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>C</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs3915217</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>IVS1-6T&gt;C</td>
<td>A</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>A</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs38724775</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>IVS1-110G&gt;A</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>C</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs39004220</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>IVS2-654C&gt;T</td>
<td>G</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>G</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs34451549</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>IVS2-745C&gt;G</td>
<td>G</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>G</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs34690599</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>W15X</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>C</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs63750783</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>Q39X</td>
<td>G</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>G</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs11549407</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
<tr>
<td>HbC</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HBB</td>
<td>C</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs39301665</td>
<td>Typical copy from one of your parents</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [6, 16, 21, 22]</td>
</tr>
</tbody>
</table>

*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

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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Risk</th>
<th>Ref ID</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sardinian</td>
<td>1 in 250</td>
<td>[ 24 ]</td>
</tr>
<tr>
<td>Cypriot</td>
<td>1 in 59</td>
<td>[ 20 ]</td>
</tr>
<tr>
<td>Italian, particularly from Sicily</td>
<td>1 in 61</td>
<td>[ 2 ]</td>
</tr>
<tr>
<td>Greek</td>
<td>1 in 37</td>
<td>[ 32 ]</td>
</tr>
<tr>
<td>Turkish</td>
<td>1 in 65</td>
<td>[ 8 ]</td>
</tr>
</tbody>
</table>

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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Detection Rate</th>
<th>Ref ID</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sardinian</td>
<td>97%</td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>Cypriot</td>
<td>90% (averaged between Greek and Turkish Cypriot)</td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>Italian, particularly from Sicily</td>
<td>82%</td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>Greek</td>
<td>75%</td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>Turkish</td>
<td>66%</td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>Balkan</td>
<td></td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>South Asian</td>
<td></td>
<td>[ 17 ]</td>
</tr>
<tr>
<td>Southeast Asian</td>
<td></td>
<td>[ 17 ]</td>
</tr>
</tbody>
</table>

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%

Middle Eastern

Oman: 64%
United Arab Emirates:
43% Saudi Arabia:
60% Lebanon:
51% Jordan:
48% Bahrain:
44% Syria:
41% Yemen:
40% Kuwait:
33% Iran: 29%

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.

References

17. HbVar Database


