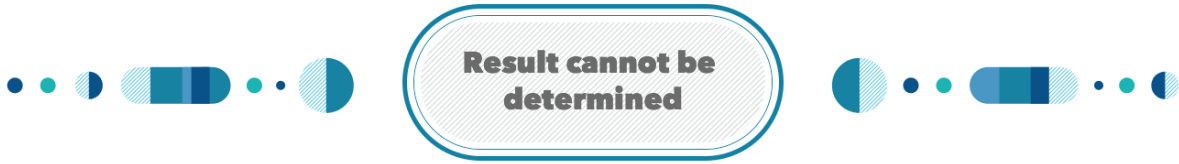


# Sickle Cell Anemia

Sickle cell anemia is a genetic disorder characterized by anemia, episodes of pain, and frequent infections. A person must have two HbS variants in the HBB 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

- To test for the HbS variant in the HBB gene.
- To identify carrier status for sickle cell anemia.

## - Limitations

- Does **not report** if someone has two copies of a tested variant.
- This report only discusses sickle cell anemia, not other forms of sickle cell disease. [See the beta thalassemia report for more information.](#)

## 🌐 Important Ethnicities

- This test is most relevant for people of **African** descent.

## About Sickle Cell Anemia



### When symptoms develop

Symptoms typically develop by early childhood.

### How it's treated

Treatment focuses on managing pain and preventing complications. Certain medications or blood transfusions may improve symptoms.



### Typical signs and symptoms

- Anemia
- Fatigue
- Episodes of pain
- Frequent infections
- Stroke
- Injury to multiple organs



### Ethnicities most affected

This condition is most common in people of African descent.

### Read more at

[Genetics Home Reference](#) [↗](#)

[GeneReviews](#) [↗](#)

[Centers for Disease Control and Prevention](#) [↗](#)

[National Heart, Lung, and Blood Institute](#) [↗](#)

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](#)

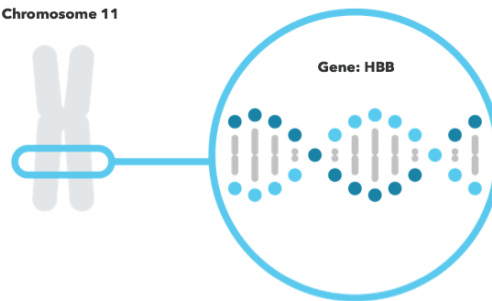
Sickle cell anemia is caused by the HbS variant in the HBB gene.

### 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](#)

Chromosome 11



Your result cannot be determined.

Variants Detected		View All Tested Markers	
0		1	
Marker Tested	Your Genotype*	Additional Information	
<b>HbS</b> Gene: HBB Marker: i3003137	<b>Not determined</b>	<ul style="list-style-type: none"><li>&gt; <b>Biological explanation</b></li><li>&gt; <b>Typical vs. variant DNA sequence(s)</b></li><li>&gt; <b>Percent of 23andMe customers with variant</b></li><li>&gt; <b>References [ 1, 3, 4, 5, 6, 7 ]   ClinVar</b></li></ul>	

\*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 Sickle Cell Anemia is indicated for the detection of the HbS variant in the HBB gene. This test is intended to be used to determine carrier status for sickle cell anemia in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of African and African American descent.

### Special Considerations

- Carrier screening for hemoglobinopathies such as sickle cell anemia is recommended by ACOG for people of African 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 is provided only where sufficient data is available.

African American	100% (This report covers the only variant that causes sickle cell anemia)	[ 2 ]
African	100% (This report covers the only variant that causes sickle cell anemia)	[ 2 ]

### Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 54 samples with known variant status. 54 out of 54 genotype results were correct. About 1 in 10,400 samples may receive a **Not Determined** result. 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 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

- ACOG Committee on Obstetrics. (2007). "ACOG Practice Bulletin No. 78: hemoglobinopathies in pregnancy." *Obstet Gynecol.* 109(1):229-37. [↗](#)
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- Turhan A et al. (2002). "Primary role for adherent leukocytes in sickle cell vascular occlusion: a new paradigm." *Proc Natl Acad Sci U S A.* 99(5):3047-51. [↗](#)