

Bloom Syndrome

Bloom syndrome is a rare genetic disorder characterized by impaired growth and increased risk of infections and cancer. A person must have two variants in the BLM gene in order to have this condition.

Erin, we could **not determine** if you have the variant we tested.

This test is intended to detect a variant in the BLM gene but your result could not be determined.



This can be caused by random test error, other factors that interfere with the test, or if you have two copies of a tested variant.

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 BLM^{Ash} variant in the BLM gene.
- To identify carrier status for Bloom syndrome.

- 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 **Ashkenazi Jewish** descent.

We could not determine your carrier status.

Carrier testing for Bloom syndrome may still be relevant for you because you have **Ashkenazi Jewish** ancestry.



We could not rule out the tested variant for Bloom syndrome.

This variant is most common in people of **Ashkenazi Jewish** descent.

You still have a chance of being a carrier.

The average person of Ashkenazi Jewish descent may have up to a **1 in 107 chance** of carrying a variant for Bloom syndrome.

[See Scientific Details](#)



About Bloom Syndrome

Also known as: Bloom-Torre-Machacek Syndrome, Congenital Telangiectatic Erythema



When symptoms develop

Symptoms typically develop during infancy.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications such as infection and cancer.



Typical signs and symptoms

- Small body size
- Recurring infections
- Cancer at a young age
- Sun-sensitive skin
- Infertility in men
- Early menopause in women



Ethnicities most affected

This syndrome is most common in people of Ashkenazi Jewish descent.

Read more at

[Genetics Home Reference](#)

[GeneReviews](#)

[National Organization for Rare Disorders](#)

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](#)



If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.

[Print report](#)



Share your results with a healthcare professional.

[Print report](#)



Learn more about this condition and connect with support groups.

[Learn more](#)

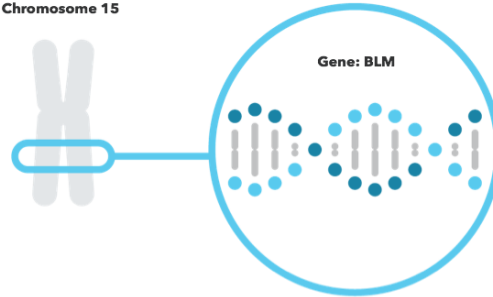
Bloom syndrome is caused by variants in the BLM gene.

BLM

The BLM gene contains instructions for making a protein called Bloom Syndrome Protein, also known as RecQ2. This protein helps protect DNA when it is copied and repaired. Certain variants in BLM disrupt this protective function, which can lead to harmful breaks and rearrangements in DNA.

Read more at [Genetics Home Reference](#) [↗](#)

Chromosome 15



Your result for this test could not be determined.

Variants Detected		View All Tested Markers
0		1
Marker Tested	Your Genotype*	Additional Information
BLM Ash Gene: BLM Marker: i4000396	Not determined	<ul style="list-style-type: none">> Biological explanation> Typical vs. variant DNA sequence(s)> Percent of 23andMe customers with variant> References [1, 2, 3] 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

You have received a **Not Determined** test result. This result can be caused by random test error or other factors that interfere with the test. It can also be caused by having two copies of the BLM^{Ash} variant.

Because you received a **Not Determined** test result, your chance of being a carrier for Bloom syndrome may be up to 1 in 107. This is the average chance of being a carrier for a person of Ashkenazi Jewish descent.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Bloom Syndrome is indicated for the detection of the BLM^{Ash} variant in the BLM gene. This test is intended to be used to determine carrier status for Bloom syndrome 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 descent.

Special Considerations

- Symptoms of Bloom syndrome may vary between people with the condition even if they have the same genetic variants.
- Carrier testing for Bloom syndrome is recommended by ACMG for people of Ashkenazi Jewish descent considering having children. This test includes the variant 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	>99%	[3]
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Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 70 samples with known variant status. 70 out of 70 genotype results were correct. Fewer than 1 in 100,000 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 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. Ellis NA et al. (1998). "The Ashkenazic Jewish Bloom syndrome mutation blmAsh is present in non-Jewish Americans of Spanish ancestry." *Am J Hum Genet.* 63(6):1685-93. [↗](#)
2. German J et al. (2007). "Syndrome-causing mutations of the BLM gene in persons in the Bloom's Syndrome Registry." *Hum Mutat.* 28(8):743-53. [↗](#)
3. Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." *Genet Med.* 10(1):54-6. [↗](#)
4. Sanz MM et al. (1993). "Bloom's Syndrome" [↗](#)