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.

play+cd8abc8b34, you **do not have the variant** we tested.

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

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.

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.

You are likely not a carrier.

This result may be less relevant for you because the variants that cause Bloom syndrome are rarely found in people of your ethnicity.

We ruled 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 for Bloom syndrome.

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.

About Bloom Syndrome

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

When symptoms develop

Symptoms typically develop during infancy.

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.

How it's treated

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

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.

Share your results with a healthcare professional.

Learn more about this condition and connect with support groups.

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Scientific Details

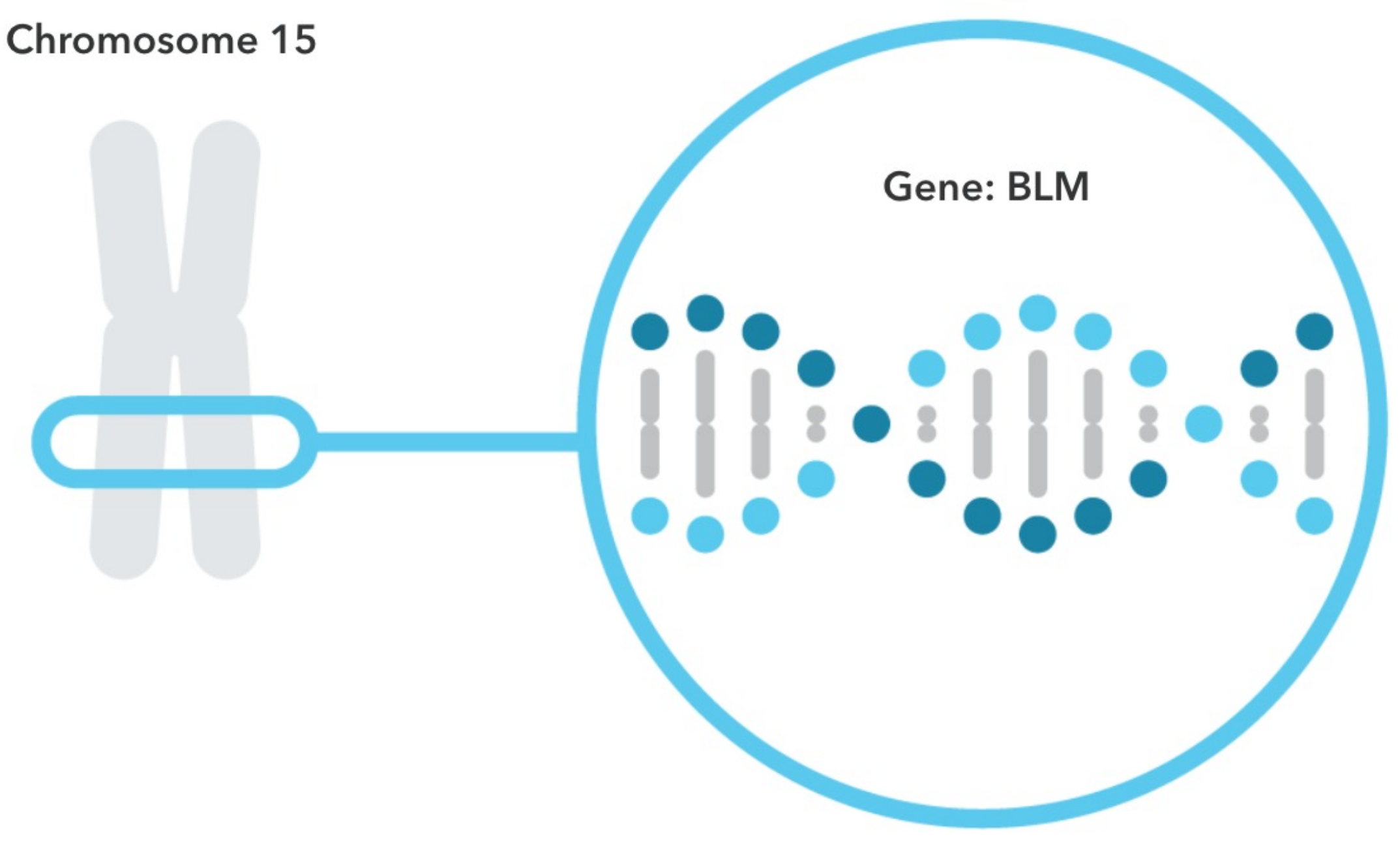
Bloom syndrome is a rare genetic disorder characterized by impaired growth and increased risk of infections and cancer. A person must have two variants in the BLM gene in order to have this condition.

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



You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
BLM^{Ash} **Gene:** BLM **Marker:** i4000396	**ATCTGA** Typical copy from one of your parents	**ATCTGA** Typical copy from your other parent	**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

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 Ashkenazi Jewish descent only.

- For people of partial Ashkenazi Jewish descent, post-test carrier risk is less than that for those who are fully Ashkenazi Jewish. The exact post-test risk depends on how much Ashkenazi Jewish ancestry a person has.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

Ashkenazi Jewish	1 in 11,000	[3]

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]

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. 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. (2006). "Bloom's Syndrome." [Updated 2016 Apr 7].

Change Log

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

Date	Change
[redacted]	Bloom Syndrome report created.

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