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.

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 BLM A68 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.

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 this report.

If you think you might have symptoms or if this condition runs in your family, consult with a healthcare professional.

If you’re starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Learn more about this condition and connect with support groups.

Bloom syndrome is caused by variants in the BLM gene.

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.

Your result cannot be determined.

<table>
<thead>
<tr>
<th>Variants Detected</th>
<th>View All Tested Markers</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td>1</td>
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</table>

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>BLM Ash</td>
<td>Not determined</td>
<td></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 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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
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<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>&gt;99%</td>
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</table>

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

4. Sene MM et al. (1993). "Bloom’s Syndrome"