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+c88abc834, you do not have the variant we tested.

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

0 variants detected in the BLM 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 results. See Scientific Details

Intended Uses

- To test for the BLM7Y™ variant in the BLM gene.
- To identify carrier status for Bloom syndrome.

Limitations

- Does not test for all possible variants for the syndrome.
- Does not detect if someone has two copies of a tested variant.

Important Equalities

- 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 determine your chances because this condition is rare and not well studied in your ethnicity.

About Bloom Syndrome

Also known as: Bloom-Tansley Syndrome, Congenital Telangiectatic Erythema

When symptoms develop

- Symptoms typically develop during infancy.

Typical signs and symptoms

- Small body size
- Repeated infections
- Cancer at a young age
- Scurvy-like gums
- Inability to tan
- Early, premature in women

Ethnicity 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 infections and cancer.

Consider talking to a healthcare professional if you are concerned about your results.

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

Contact with a c7c7

Share your results with a healthcare professional.

Find support

Learn more about this condition and connect with support groups.

More info
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.

You have no variants detected by this test.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of Ashkenazi Jewish descent only.

- Few people of European (non-Ashkenazi) Jewish descent are carriers.
- For people of European (non-Ashkenazi) Jewish descent, the risk test may not be the best option.
- Results for both carrier parents are needed because sufficient data is not available.

Test Indications

The Clinava PGS Carrier Status Test for Bloom Syndrome is indicated for the detection of the BLM* variant or the BLM** variant.

- This test is intended to assess whether a person is a carrier of Bloom syndrome in adults. But, determination if a person has a two-copy variant is not available.
- The test is also relevant for people of Jewish descent.

Special Considerations

- Symptoms of Bloom syndrome may vary among people with the condition even if they have the same genetic variant.
- Carrier testing for Bloom syndrome is recommended by ACMG for people of Ashkenazi Jewish descent considered by children. This test includes the variant recommended for testing by ACMG.

Test Performance Summary

Carrier Detection Rate & Relevant Variations

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be detected by this test. Carrier detection rate differs by ethnicity and is provided only when sufficient data is available.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Detection Rate</th>
</tr>
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<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>98% [1]</td>
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</table>

Analytical Performance

A determinant was determined by computing results from the test with results from sequencing. Greater than 99% of test results were concordant. With this in mind, the test may provide 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 is not intended to screen for any factors that could cause this condition.
- This test does not diagnose any health conditions.
- Positive results in individuals who are not related to the carrier may occur. In this situation, further testing should be performed.
- This test is not intended for use by healthcare professionals.

Test Details

- This test is not covered by your current insurance.
- This test is not available in all states.

References