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.

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.

See Scientific Details

About Bloom Syndrome

Also known as: Bloom-Torre-MacKerrow 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 this report.

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

Connect with a GC

Learn more about this condition and connect with support groups.

Receive up to $25 when you refer family and friends to 23andMe. Get started today.
Bloom Syndrome

Bloom syndrome is a rare genetic disorder characterized by increased risk of infections and cancer. A person must have two variants in the BLN1 gene in order to have this condition.

**Genetic Home Reference**

The BLN1 gene contains instructions for making a protein called Bloom Syndrome Protein, also known as RecG2. This protein helps protect DNA when it is copied and repaired. Certain variants in BLN1 disrupt this protective function, which can lead to harmful changes and rearrangements in DNA.

Your result cannot be determined.

**Variants Detected**

<table>
<thead>
<tr>
<th>Variant Detected</th>
<th>Your Genotype*</th>
</tr>
</thead>
<tbody>
<tr>
<td>BLN1**</td>
<td>Not determined</td>
</tr>
</tbody>
</table>

**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 or having children. This test includes the variants recommended for testing by ACMG.

**Test Performance Summary**

The carrier detection rate is an estimate of the percentage of carriers for the condition that would be identified by the test. Carrier detection rates differ by ethnicity and are provided only where sufficient data is available.

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

**Analysis**

Accuracy was determined by comparing results from this test with results from sequenced tests. Greater than 99% of test results were correct. While unlikely, it is possible to receive false positive or false negative results. For more details about the analytical performance of this test, refer to the package insert.

**Warnings and Limitations**

- This test does not assess 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.
- Results may not be the last word. They may not be available on our genotyping platform, or may not pertain to your testing standards.

See the Package Insert for more details on use and performance of this test.

**References**


**Change Log**

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