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 BLM gene in order to have this condition.

† play+5651cb:3024, you have the variant we tested.

You could pass this variant on to your children.

1 variant detected
is at 40,024

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 the condition, or you have any concerns about your results.

Review the Carrier Status tutorial
See Scientific Details

Intended Uses

• To test for the BLM pathogenic variant in the BLM gene

Limitations

• You can test both of you for possible carriers for the condition.

Important Ethnecities

• This test is most relevant for people of Ashkenazi Jewish descent.

You are a carrier.

You could pass this variant on to your children.

We detected one variant for Bloom syndrome.

People with only one variant are not expected to have Bloom syndrome.

Your results may be relevant for you if you’re thinking about starting a family.

If you and your partner are both carriers, each child has approximately 25% chance of having this condition. Your physician may also wish to consider testing your fetus prior to having children.

About Bloom Syndrome

Also known as: Bloom Syndrome, Bloom氏综合征, Albus-Bloom Syndrome

When symptoms develop

Symptom typically develop during infancy

Typical signs and symptoms

• Small body size

• Repeating infections

• Cancer at young age

• Sun sensitivity skin

• Infertility in men

• Early enucleate in variant

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Read more at: Genes in Health Resources™, SanfordGenetics™, National Organization for Rare Disorders™

Consider talking to a healthcare professional if you are thinking about having children.

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

Contact with a GC

Share your results with your family.

Give your results.

If you have other concerns about your results, consult with a healthcare professional.

Find support.

Learn more about this condition and connect with support groups.

Speak more.
Bloom syndrome is caused by variants in the Bloom gene. The Bloom 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 Bloom change this protective function, which can lead to harmful changes and rearrangements in DNA. Read more at Genetics Home Reference.

You have one variant detected by this test.

**Variants Detected**

<table>
<thead>
<tr>
<th>Variant Type</th>
<th>Genotype*</th>
</tr>
</thead>
<tbody>
<tr>
<td>BLRM&lt;sup&gt;mo&lt;/sup&gt;</td>
<td>R.R</td>
</tr>
<tr>
<td>ATCCTA</td>
<td>Typical copy from one of your parents</td>
</tr>
<tr>
<td>TAGACC</td>
<td>Variant copy from your other parent</td>
</tr>
</tbody>
</table>

**Additional Information**

- **Biological explanation:**
- **Typical vs. variant DNA sequence:**
- **Percent of 23andMe customers with variant:**
- **Reference:** [1, 2, 3] [4, 5]

**Test Details**

**Indications for Use**

The 23andMe PEDIGREE Status Test for Bloom Syndrome is indicated for the detection of the BLRM<sup>mo</sup> variant in the Bloom gene. This test is intended to be used to determine prior status for Bloom syndrome in adults, but cannot determine if a person has two copies of a variant 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 rates do not reflect the method by which sufficient data is available.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Test Performance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>&gt;90%</td>
</tr>
</tbody>
</table>

**Analytical Performance**

Accuracy was determined by comparing results from the 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 know all variants that could cause this condition.
- This test does not diagnose any health conditions.
- Possible results in individuals whose ethnicities are not commonly associated with this condition may be missed.
- Individuals in this situation should consider genetic counseling and follow-up testing.
- Results should be used in consultation with a healthcare professional.

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.

<table>
<thead>
<tr>
<th>Date</th>
<th>Change</th>
</tr>
</thead>
<tbody>
<tr>
<td>[ ]</td>
<td>Bloom Syndrome report created.</td>
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</table>