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.

We could not determine your carrier status.

Your test result could not be determined. This variant is most common in people of Ashkenazi Jewish descent.

About Bloom Syndrome

This variant is most common in people of Ashkenazi Jewish descent.

We cannot estimate your risk because the condition is rare and not well studied in your ethnicity.

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

If you or your family have a history of this condition or you think you have symptoms, consult with a healthcare professional.

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 Re claims. This protein helps protect DNA when it is copied and repaired. Certain changes in BLM slow down this protective function, which can lead to harmful breaks and rearrangements in DNA.

Your result for this test could not be determined.

Test Interpretation

You have received a Not Determined result. This result can be caused by several test errors or other factors that interfere with the test. It can also be caused by having two copies of a BLM variant.

Because you received a Not Determined result, we have no new information about your chance of having a parent with Bloom syndrome. We cannot estimate your chance because the average chance of having a parent with Bloom is not known for your family.

Test Details

Indications for Use

The 23andMe PGD Carrier Status Test for Bloom Syndrome is indicated for the detection of the BLM variant in the BLM gene. This test is intended to be used to determine carrier status for Bloom syndrome if this condition is suspected. However, it cannot confirm the presence of a family history of Bloom syndrome.

Special Considerations

- Individuals with Bloom syndrome may vary in their symptoms, even if they share the same genetic variants.
- This test is not intended to diagnose or confirm a Bloom syndrome diagnosis. If a Bloom syndrome diagnosis is being considered, please consult with your healthcare provider.

Performance Test Summary

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
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<tbody>
<tr>
<td>Detection Rate</td>
<td>99%</td>
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Analysis and Reporting

The result is determined by comparing the test results with results from a large database of known variants. A variant is considered significant if it is found in 0.1% or more of the population and has been associated with Bloom syndrome.

Warnings and Limitations

- The test does not cover all variants that could cause this condition.
- It does not diagnose or confirm any health conditions.
- Results may vary, and the presence of a Bloom syndrome variant does not confirm a diagnosis. A healthcare professional should interpret the test results.

References