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 the condition.

play+c08abcB34, 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 Consent, Terms & Conditions
See Scientific Details

Intended Uses

- To test for the BLM® variant in the BLM gene.
- To identify carriers for Bloom syndrome.

Limitations

- Does not test for all possible variants for the condition.
- Does not report if you are a carrier but does identify two copies of a tested variant.

Important Ethnicities

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

You are likely not a carrier.

This result is relevant for you because you have Ashkenazi Jewish ancestry.

We ruled out the most common variant for Bloom syndrome in people of Ashkenazi Jewish descent.

You still have a chance of being a carrier for Bloom syndrome.

You may still have up to a 1 in 11,000 chance of carrying a variant not covered by the test.

See Scientific Details

About Bloom Syndrome

Also known as: Bloom-Torea-Machacek Syndrome, Congenital Telangiectatic Osteodysplasia

Where symptoms develop

Symptoms typically develop during infancy.

Typical signs and symptoms

- Small body size
- Rebounding effects
- Cancer at a young age
- Sun-sensitive skin
- Infertility in men
- Early menopause in women

Ethnicities most affected

This disease 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 your results.

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

Contact a genetic counselor.

Share your results with a healthcare professional.

Learn more about:

Support groups and resources:

1. Bloom Syndrome Registry
2. Jewish Genetic Disease Consortium
3. Bloom’s Syndrome Association

Your test results are not intended to be used in isolation outside the clinical context.

For more information, contact your blood bank or your patient care team about your Bloom’s test and any further disorders or genetic medical conditions they may note.
Bloom syndrome is caused by variants in the BLM gene.

**Test Interpretation**

**Post-test Carrier Risk**

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

- For people of partial (50%) Jewish heritage, Jewish Ashkenazi carrier risk is no lower than that for those who are fully Jewish. Jewish identity depends upon the mother's Ashkenazi-Jewish ancestry in her parent's place of origin.
- Jewish Ashkenazi carrier risk cannot be provided because sufficient data is not available for non-Jewish carriers.

**Arms Test Details**

![Arms Test Details](image)

**Indications for Use**

- The Modular Pan-Cancer Test: Bloom Syndrome is indicated for the detection of the BLM variant in the given germline tissue. This test is intended to be used in conjunction with other clinical and laboratory data to inform genetic counseling and clinical management. However, it is not intended as a stand-alone diagnostic tool. Bloom Syndrome is a genetic condition that is often associated with a higher risk of developing certain types of cancer.

**Test Performance Summary**

**Carrier Detection Rate & Reliableness**

The carrier detection rate of the Modular Pan-Cancer Test is 99.5%, which is considered high. This rate takes into account clinical and laboratory factors to inform genetic counseling and clinical management. However, it is not intended as a stand-alone diagnostic tool. Bloom Syndrome is a genetic condition that is often associated with a higher risk of developing certain types of cancer.

**References**