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.

How To Use This Test

This test does not diagnose any health conditions. Please talk to a healthcare professional if this condition exists in your family, you think you might have this condition, or you have any concerns about your health.

Intended Uses

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

Limitations

- Does not test for all possible variants for the condition.
- DNA report format has 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 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 estimate your chances because this condition is rare and not well studied in your ethnicity.

About Bloom Syndrome

Also known as: Bloom Syndrome, Bloom Syndrome, Malignant Neoplasms, Cancer, Chromosome Abnormalities, Anemia, Infertility, Cancer, Infections, Neoplasms

When symptoms develop

Symptoms typically develop during infancy.

How it’s treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications such as infections and cancer.

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.

Read more on:

- Genetics Home Reference
- Genetic Alliance
- National Organization for Rare Disorders

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

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

Share your results 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 BRCA2. This protein helps repair DNA when it is damaged or copied. Genetic errors in the BLM gene prevent repair of DNA damage, leading to a higher risk of birth defects and premature aging in humans.

Read more at [Bloom Syndrome’s Home Reference](#)