Bloom Syndrome

Bloom syndrome is a rare genetic condition that affects physical and mental development. It is caused by mutations in the BLM gene, which can lead to an increased risk of cancer and other health problems.

Play +9771264768, we could not determine if you have the variant we tested.

This test is intended to detect a variant in the BLM gene, but your result could not be determined.

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 may need to have this condition, or to have any other information about your results.

Review the Carrier Status report
See Scientific Details

We could not determine your carrier status.

We could not rule 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. The average person of Ashkenazi Jewish descent may have up to a 1 in 4 chance of carrying a variant for Bloom syndrome.

See Scientific Details

About Bloom Syndrome

Also known as: Bloom Syndrome, Neill Syndrome, Congenital Telomeric Synthetase Deficiency

When symptoms develop:
Symptoms typically develop during infancy.

Typical signs and symptoms:
• Small, broad nose
• Recurrent infections
• Cataracts at any age
• Sun sensitivity
• Hair loss in men
• Early signs in carrier

Read more at:
Genetics Home Reference® GeneTests® National Organization for Rare Disorders®

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

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

Connect with a GC

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

Print support

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

Print support

Learn more about this condition and connect with support groups.

Learn more
Bloom syndrome is an autosomal recessive disorder caused by mutations in the BLM gene, which is located on chromosome 15. Patients with Bloom syndrome have a higher risk of developing cancer due to their inability to repair DNA damage. The diagnosis is typically made based on the presence of typical physical features, such as sunburned appearance, and a blood test that shows an abnormal number of white blood cells.

**Indications for Use**

The Bloom syndrome test is designed for individuals who are at risk of developing cancer due to genetic mutations in the BLM gene. The test can help identify individuals who may benefit from cancer surveillance and targeted therapies.

**Test Details**

**Performance Summary**

- **Specificity:** 100%
- **Sensitivity:** 100%

**Analysis Report**

The test was performed on a blood sample from the patient. The results show no mutations in the BLM gene, indicating a normal risk for developing cancer.

**References**


**Change Log**

- **Date**: [Date]
- **Change**: Bloom Syndrome report created.