



What is Bloom syndrome?

Bloom syndrome is an inherited disease characterized by short stature, sun-sensitivity, and increased risk of cancer caused by the instability of DNA during cell division and the inability of the body to repair damaged DNA. Symptoms associated with Bloom syndrome are the result of unrepaired gaps and breaks in the genetic material that impair normal cell functions.¹ Bloom syndrome is also known as Bloom-Torre-Mackacek syndrome and congenital telangiectatic erythema.²

What are the symptoms of Bloom syndrome and what treatment is available?

The symptoms of Bloom syndrome may include³:

- Prenatal and postnatal growth deficiency, which may be the only symptom apparent at birth, and short stature in all stages of life
- Feeding difficulties in infancy
- Gastrointestinal reflux, which may contribute to repeated lung and ear infections
- Red (butterfly-shaped) rash that appears after sun exposure, typically on the face and less often, on the hands, and forearms
- Immunodeficiency
- Learning problems
- Infertility in males and early menopause in females
- Serious medical complications that appear at unusually early ages, including chronic obstructive pulmonary disease and diabetes mellitus
- Increased risk of multiple cancers at early age (average age of diagnosis 26.6 years, range (<1-49)

There is no cure for Bloom syndrome at this time. Lifespan may be shortened, most commonly by cancer, but survival to the teens or 20's is expected. Preventative management is important, such as avoiding sun exposure and having regular cancer screening from a young age. Treatment focuses on managing symptoms, preventing secondary complications, and surveillance.³

How is Bloom syndrome inherited?

Bloom syndrome is an autosomal-recessive disease caused by mutations in the *BLM* gene.¹ *BLM* is also known as *RECQL3*. An individual who inherits one *BLM* mutation is a "carrier" and is not expected to have related health problems. An individual who inherits two *BLM* mutations, one from each parent, is expected to be affected with Bloom syndrome.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Bloom syndrome?

Bloom syndrome is rare in most ethnic groups with an unknown incidence. In the Ashkenazi (Eastern European) Jewish population, the carrier frequency is 1 in 134⁴, which predicts an incidence of approximately 1 in 72,000.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or a genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.





CARRIER SCREEN

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

- NIH Office of Rare Diseases Research – Genetic and Rare Diseases (GARD) Information Center: http://www.rarediseases.info.nih.gov/GARD/Condition/915/Bloom_syndrome.aspx
- Bloom's Syndrome Foundation: <http://www.bloomssyndrome.org/>

References

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4. Scott, SA, et al. Experience with Carrier Screening and Prenatal Diagnosis for 15 Ashkenazi Jewish Genetic Diseases. *Hum Mut* 2010; 31(11): 1240-1250.

