

# Bloom syndrome

**Bloom syndrome** (also known as **Bloom-Torre-Machacek Syndrome**) is an inherited disorder characterized by a high frequency of breaks and rearrangements in an affected person's chromosomes.

## Signs and Symptoms

People with Bloom syndrome are much smaller than average, and often have a high-pitched voice and characteristic facial features including a long, narrow face; small lower jaw; and prominent nose and ears. They tend to develop pigmentation changes and dilated blood vessels in the skin, particularly in response to sun exposure. These changes often appear as a butterfly-shaped patch of reddened skin on the face. The skin changes may also affect the hands and arms.

Other features of the disorder may include learning disabilities, mental retardation, chronic lung problems, diabetes, and immune deficiency that leads to recurrent pneumonia and ear infections. Men with Bloom syndrome usually do not produce sperm, and as a result are unable to father children (infertile). Women with the disorder generally experience menopause earlier than usual.

## Related Problems

Chromosome instability in Bloom syndrome results in a high risk of cancer in affected individuals. Affected individuals develop the full range of cancers found in the general population, but the cancers arise unusually early in life. People with Bloom syndrome may be first diagnosed with cancer at about 25 years old.

## Epidemiology

Bloom syndrome is a very rare disorder in most populations, and its overall frequency is unknown. It is more common in people of Central and Eastern European (Ashkenazi) Jewish background, among whom 1 in 48,000 are affected. Approximately one third of people with Bloom syndrome are of Ashkenazi Jewish descent.

## Chances of Developing Bloom Syndrome

### Genetics

Mutations in the BLM gene cause Bloom syndrome. The BLM gene provides instructions for producing a protein called the Bloom (BLM) syndrome protein, which is a member of the DNA helicase family. DNA helicases are enzymes that unwind the two spiral strands of a DNA molecule so that they can be copied. When a cell prepares to divide to form two cells, the chromosomes are duplicated (replicated) so that each new cell will get a complete set of chromosomes. The replication process involves unwinding the DNA so that it can be copied. The BLM protein is important in maintaining the stability of the DNA during this process. Mutations in the BLM gene alter or reduce the BLM protein's DNA helicase activity, which causes errors in the copying process during replication. As a result, people with Bloom syndrome have a higher frequency of chromosome breakage and rearrangement than unaffected people. This increase in chromosome breakage and rearrangement leads to the signs and symptoms of Bloom syndrome.

### Heredity

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## References

- Gene Review: Bloom's SyndromeThis link leads to a site outside Genetics Home Reference.
- MedlinePlus - Health informationHealth Topic: CancerThis link leads to a site outside Genetics Home Reference.
- Educational resources - Information pages (6 links)
- Patient support - For patients and families (2 links)
- Gene ReviewsThis link leads to a site outside Genetics Home Reference. - Clinical summary
- Gene TestsThis link leads to a site outside Genetics Home Reference. - DNA tests ordered by healthcare professionals
- Genetic ToolsThis link leads to a site outside Genetics Home Reference. - Teaching cases
- ClinicalTrials.govThis link leads to a site outside Genetics Home Reference. - Linking patients to medical research
- PubMedThis link leads to a site outside Genetics Home Reference. - Recent literature
- Online Books - Medical and science textsScriver's Online Metabolic and Molecular Bases of Inherited Disease (OMMBID):Bloom SyndromeThis link leads to a site outside Genetics Home Reference.

- OMIMThis link leads to a site outside Genetics Home Reference. - Genetic disorder catalog

## External links

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