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## Bloom syndrome

### What is Bloom syndrome?

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Bloom syndrome is a rare inherited condition characterised by:

- Telangiectases (visible broken blood vessels under the skin) on the face
- [Photosensitivity](#)
- Unusually small size at birth
- Increased susceptibility to infections and respiratory illness
- Increased susceptibility to cancers of many sites and types.

The condition was first described by dermatologist Dr David Bloom in 1954 and is also known as congenital telangiectatic erythema.

### What causes Bloom syndrome and who gets it?

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Bloom syndrome is an autosomal recessive inherited disorder, which means that two abnormal Bloom's syndrome genes are needed for the disease to be apparent (one from each parent). If a person has one affected gene, he or she is called a carrier of Bloom Syndrome and does not manifest symptoms of the disease. If both parents are carriers, there is a 1 in 4 chance of having an affected child with each pregnancy.

The gene for Bloom syndrome is located on chromosome 15 (gene locus is band 15q26.1). Mutations in this gene cause errors in the copying process during DNA replication and result in a higher number of chromosome breakages and rearrangements. This leads to the signs and symptoms of Bloom syndrome.

Bloom syndrome is more common in eastern European Ashkenazi Jews. At least 1 in 100 Ashkenazi Jews is a carrier of the disease. It appears to be slightly more common in males than females.

### What are the signs and symptoms?

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Bloom syndrome is apparent from birth with affected newborns being unusually small. Most parents seek medical help when the infant does not grow normally. Over 50% of children are significantly underdeveloped in physical stature until age 8 years, and most fail to reach 1.5 metres (5 feet) in adulthood.

Other features of Bloom syndrome become apparent as the child grows older. These include:

- Skin features
  - Telangiectatic erythema - broken spidery veins and redness of the skin of the face, mainly across the bridge of the nose and adjoining upper cheek area (butterfly distribution)
  - Pigmentation of sun exposed areas
  - Cheilitis (swelling of the lips) with crusting or bleeding
  - Skin problems are aggravated by sun exposure (photosensitivity) and vary in severity from mild to severely disfiguring
  - There is some improvement with age
- Skull, face and body features
  - Lack of subcutaneous fat creates a birdlike appearance, with a long narrow face and prominent nose

- Relatively large protruding ears
- Long limbs, particularly long arms in proportion to body length
- Disproportionately large hands and feet
- A high-pitched voice may be present
- Increased susceptibility to ear, respiratory and gastrointestinal infections, some of which can be life-threatening.
  - Patients with Bloom syndrome have decreased immunoglobulin A and M.
- Increased susceptibility to cancer
  - Patients with Bloom syndrome have an overall 150- to 300-times increased risk of developing some form of cancer compared with the general population
  - 20% of patients with Bloom syndrome develop cancer
  - Cancers usually arise unusually early in life, with the diagnosis made at about 25 years of age
- Infertility
  - Men with Bloom syndrome are sterile
  - Women have reduced fertility and shortened reproductive span

## Treatment

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Because it is a genetic disorder, no cure is available. Genetic counselling may be appropriate for affected individuals and carriers (their parents and siblings). A genetic test to identify the mutation is available in some regions.

Treatment is primarily preventative. Patients with Bloom syndrome should be monitored closely for signs of precancerous or cancerous conditions. Bacterial infections should be treated promptly with antibiotics.

### Related information

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#### References:

- [Bloom Syndrome](#) – eMedicine Dermatology, the online textbook

#### On DermNet NZ:

- [Skin signs of systemic disease](#)
- [Photosensitivity](#)

#### Other websites:

#### Books about skin diseases:

See the [DermNet NZ bookstore](#)

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If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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