

Bloom Syndrome

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Bloom Syndrome Latest News

Recently, a 12-year-old girl diagnosed with the Bloom Syndrome, underwent a bone marrow transplant using stem cells from her younger brother at a private hospital in Chennai.

About Bloom Syndrome

- Bloom Syndrome or **BSyn** is a **genetic disorder**, in which the **BLM gene**, a protein-coding gene, does **not function as it should**.
- It leads to growth delays, increased vulnerabilities to infections, sensitivity to the sun and an increased risk for certain cancers.
- The BLM gene is a **gene that makes a protein involved** in maintaining the structure of DNA, when DNA is copied in a cell, and in repairing damaged DNA.
- Mutations in the BLM gene may **cause cells to lose their ability** to repair damaged DNA, which may **lead to abnormal cell growth**.

- **Other names** for Bloom Syndrome are **Bloom-Torre-Machacek syndrome and congenital telangiectatic erythema.**

How does it Occur?

- It is **inherited in an autosomal recessive** pattern. This means that if both parents have a mutation in one of their two BLM genes and if the child inherits two of the disease-causing variants of the gene, then the disorder occurs.
- It is seen **most commonly in the Eastern European** (Ashkenazi) Jewish population.

Signs and Symptoms

- **Poor growth** is a common clinical feature — this happens during the development of the foetus and after birth. **Below normal average height** and head circumference may be seen. There may be abnormalities in certain features such as **narrowing of the head and face**, prominent ears and nose, long arms and legs and high-pitched voices.
- **Skin lesions:** The skin is very sensitive to sunlight and red rashes may develop after exposure.
- **Insulin resistance** leads to an increased risk of diabetes, while other issues include immune deficiencies.
- People with Bloom Syndrome are at a **higher risk of contracting infections** such as ear and lung infections and also chronic obstructive pulmonary disease (COPD).
- In adults, male sterility and female **infertility may occur.**
- Bloom Syndrome increases the risk of **developing cancer, and at an early age.**
- **Treatment:** It involves a multi-disciplinary approach. Since there is no specific treatment, the goal is to manage symptoms.

Source: TH

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