Bloom Syndrome

Prelims – *Current events of national and international importance | Health*

Why in News?

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

- It is a ***rare genetic disorder***, characterised by short stature and blood-related complications, and carries a risk of blood cancers such as acute myeloid leukemia.
- People with Bloom syndrome are usually smaller than 97% of the population in both height and weight from birth, and they rarely exceed 5 feet tall in adulthood.
- **Causes -** Bloom Syndrome is an autosomal recessive genetic disorder, meaning a person must inherit *2 copies of the mutated BLM gene, one from each parent*.
- The BLM gene provides instructions for making a protein called a *RecQ helicase*, which is crucial for DNA stability.
- Mutations in the BLM gene lead to a non-functional BLM protein, resulting in genomic instability and a high number of chromosomal breaks.
- **Symptoms**
  - **Growth Deficiency -** Individuals typically experience severe growth delays both before and after birth.
  - **Photosensitive Skin -** A characteristic butterfly-shaped rash develops on the face after sun exposure, along with telangiectases (small, enlarged blood vessels) and other skin changes.
  - **Increased Cancer Risk -** There is a greatly increased risk of developing many types of cancer, often at an early age.
  - **Immune Deficiencies -** People with Bloom Syndrome may have immune system issues, making them more vulnerable to infections.
  - **Other Complications -** These can include insulin resistance, an

increased risk for diabetes, and chronic obstructive pulmonary disease.

- **No Cure -** There is ***currently no treatment*** that targets the underlying genetic cause of the disorder.
- **Recent case -** The girl had
  - Monosomy 7, a chromosomal abnormality involving the loss of one copy of chromosome 7, and
  - Myelodysplastic Syndrome (MDS), a disorder in which the bone marrow fails to produce healthy blood cells.
- There is a risk of MDS progressing to leukemia.
- She underwent bone marrow transplant (BMT) using stem cells from her younger brother.

Reference

The Hindu | Bloom Syndrome

