Hunter Syndrome: MPS II

Why in News

Two brothers suffering from **Mucopolysaccharidosis II or MPS II (Hunter Syndrome, Attenuated Type)** have approached the Delhi High Court seeking direction to the Centre and AIIMS to provide them free treatment.

- **MPS II is a rare disease** that is passed on in families.

Key Points

- **About:** MPS II mostly **affects boys** and their bodies **cannot break down a kind of sugar** that **builds bones, skin, tendons and other tissues.**
- **Cause:** It is **caused by changes (mutations) of the *IDS* gene** that regulates the production of the iduronate 2-sulfatase (I2S) enzyme.
    - This enzyme is **needed to break-down complex sugars,** known as **glycosaminoglycans (GAGs),** produced in the body.
- **Impact:** Lack of I2S enzyme activity **leads to the accumulation of GAGs within cells,** specifically inside the **lysosomes.**
    - Lysosomes are compartments in the cell that **digest and recycle different types of molecules.**
    - Conditions that cause molecules to build up inside the lysosomes, including MPS II, are called **lysosomal storage disorders.**
    - The accumulation of GAGs increases the size of the lysosomes, which is why **many tissues and organs are enlarged in this disorder.**
- **Symptoms:** It is characterized by **distinctive facial features, a large head, enlargement of the liver and spleen (hepatosplenomegaly),** hearing loss, etc.
- **Inheritance:**
    - MPS II is inherited in an X-linked recessive pattern, which means that this condition occurs almost exclusively in males. **Females are generally unaffected carriers of this condition.**
    - In a family with more than one affected individual, the mother of the affected males must be a carrier. When a carrier female has a child, there is a 25% (1 in 4) chance that she will have an affected son.

Rare Diseases

- A rare disease is a **health condition of low prevalence** that affects a small number of people compared with other prevalent diseases in the general population.
- Though rare diseases are of low prevalence and individually rare, collectively they **affect a considerable proportion of the population.**
- **80% of rare diseases are genetic in origin** and hence disproportionately impact children.
- Recently, the Delhi High Court has directed the Centre to **finalise the National Health Policy for Rare Diseases of 2020** by March 2021 and **make operational provision of crowdfunding**

envisaged under the law for treatment of high-cost rare diseases.

Source:TH