Harlequin Ichthyosis: Rare Genetic Disorder

Why in News

Recently, **Odisha reported its first-ever case of a baby born with Harlequin Ichthyosis,** a rare genetic skin condition.

- **India's first recorded case** of a baby born with harlequin ichthyosis **was in 2016,** at a private hospital in Nagpur, Maharashtra.

Key Points

- **About:**
    - **Harlequin Ichthyosis** is a rare **genetic skin disorder** to a newborn infant.
    - It's a type of ichthyosis, which refers to a group of disorders that cause persistently dry, scaly skin all over the body.
    - It is inherited in an **autosomal recessive pattern.**

Autosomal Recessive Pattern

- Autosomal (that means inherited) Recessive Pattern is a way, where **a genetic trait or condition can be passed down from parent to child.**
- A genetic condition **can occur when the child inherits one copy of a mutated (changed) gene from each parent.**
- If an individual receives one normal gene and one abnormal gene for the disease, the person will be **a carrier for the disease.**
- The risk to have a child who is a carrier, like the parents, is 50% with each pregnancy. The chance for a child to receive normal genes from both parents is 25%. The risk is the same for males and females.

- **Causes:**
    - It can be **caused by changes (mutations) in the ABCA12 gene.**
    - ABCA12 **Gene gives instructions for making a protein that is necessary for skin cells to develop normally.**
    - It plays a key role in the transport of fats (lipids) to the most superficial layer of the skin (epidermis), creating an effective skin barrier.
    - When this gene is mutated, the skin barrier is disrupted.
- **Impact:**
    - Newborn infants **are covered with plates of thick skin** that crack and split apart and can **restrict breathing and eating.**
    - Premature birth is typical, leaving the infants at risk for complications from early delivery.
- **Affected Population:**
    - It affects males and females in equal numbers.

    - It affects **approximately one in 5,00,000 persons.**
    - There are around 250 such cases across the world.
- **Treatment:**
    - A newborn with Harlequin ichthyosis **requires neonatal intensive care,** which may include spending time in a heated incubator with high humidity.
- **Related Disorder:**
    - **Lamellar ichthyosis** is an inherited skin disorder characterized by broad, dark, plate-like scales separated by deep cracks.
        - Its **symptoms are similar to Harlequin Ichthyosis.**

Source DTE