



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**.

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