

# Harlequin Ichthyosis

April 23, 2021

**In News:** Odisha reported its first-ever case of a baby born with harlequin ichthyosis, a rare genetic condition, at a hospital in Berhampur in the state's Ganjam district April 22, 2021.

## About Harlequin Ichthyosis

- Mutations in the ABCA 12 gene are stated to cause harlequin ichthyosis.
- The ABCA12 protein plays a major role in transporting fats in cells which make up the outermost layer of skin.
- Severe mutations in the gene lead to the absence or partial production of the ABCA12 protein.
- The disease affected one in three million births and is caused due to a mutated gene inherited from the parents.
- The disease sees the skin form large diamond-shaped plates across the body that are separated by deep cracks..
- The skin is dry and scaly, almost like fish skin and hence the term 'ichthyosis', derived from 'ikthus', Greek for fish.
- The facial features of the baby, including the mouth, eyes and ears were deformed, restricting breathing and eating.
- The baby was kept in the intensive care unit.
- The condition of the mother was good, Indira Palo, assistant professor at the college and hospital's gynaecology department.
- India's first recorded case of a baby born with harlequin ichthyosis was in 2016, at a private hospital in Nagpur, Maharashtra.
- Such cases were also reported in Delhi, Patna and West Bengal.

- The infants could not survive for long and succumbed to the disease days after birth.
- There have been maybe 200 to 250 such cases across the world.