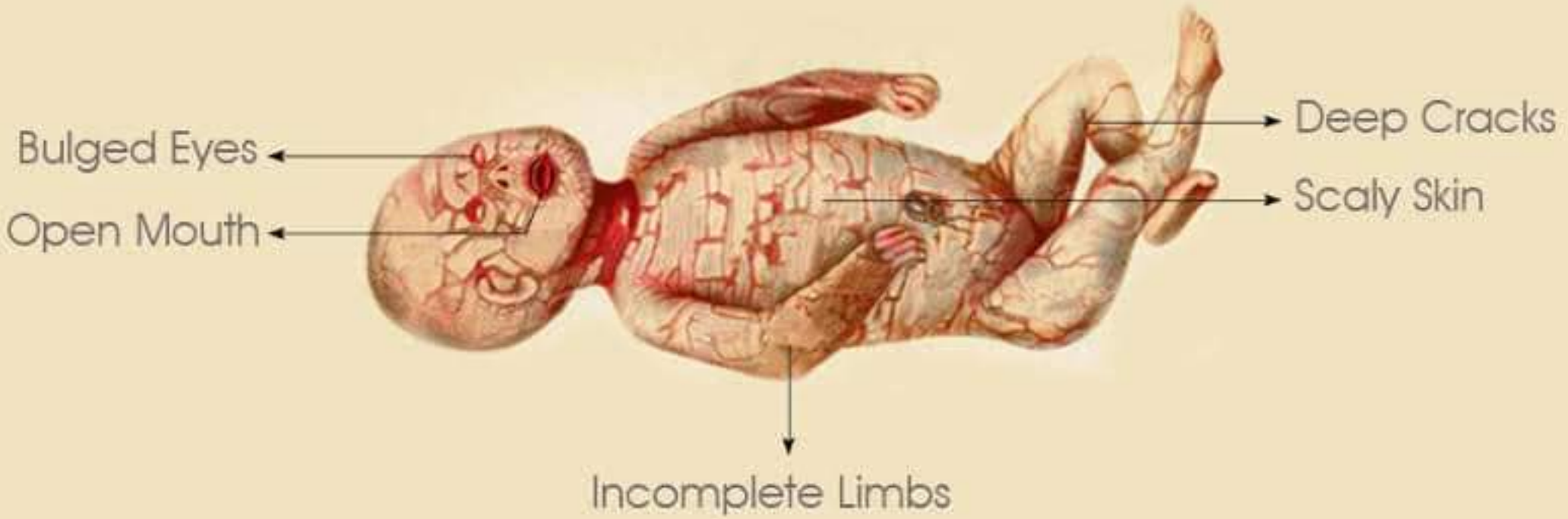
Harlequin Ichthyosis



What is Harlequin Ichthyosis ?

Harlequin ichthyosis is a rare, congenital skin condition. It affects the skin over almost the whole body of an infant, and carries a high risk of neonatal death due to severe and life-threatening infection.

Harlequin ichthyosis occurs in approximately one in every 500,000 people. Males and females both are equally vulnerable to this disorder.

Causes of harlequin ichthyosis

Harlequin ichthyosis is caused by a mutation of the ABCA12 gene. This gene helps to direct the production of the ABCA12 protein involved in transporting fats within the epidermis, which is essential for normal skin synthesis. Various mutations are found. Some can stop the production of this protein, or induce the production of an abnormal form of the protein.

Harlequin ichthyosis is inherited in an autosomal recessive fashion, which means patients with this disorder have two copies of the mutated genes, one inherited from each parent. If two people with the mutation have a child, there is a 25% chance that the child will be affected. The parents are carriers of the mutated gene and show no sign of the disease because they have one normal copy which compensates for the defective one.

Collodion baby

- Most autosomal recessive congenital ichthyosis (ARCI) present at birth as collodion babies
- Baby encased in shiny parchement-like membrane which may impair respiration and sucking .
- Usually peels off within the first 4 weeks of life.
- In 80% of cases, collodion baby is followed by the onset of an ARCI subtype
- Around 10–20% develop into self-improving congenital ichthyosis

Harlequin ichthyosis

- Most devastating type of ARCI. Often lethal in around 44% case
- Mutations in the ABCA12 gene
- Neonates are born with armour-like skin (truncal plates with fissuring)
- Impaired movement and the ability to drink and breath
- More prone to infection of the skin, as well as other organs such as the lungs (major cause of death)
- Biopsy - enormous thickening of stratum corneum. Parakeratosis and hypergranulosis and reduced non-polar lipids
- Electron microscopy – numerous abnormal lamellar bodies in stratum granulosum

PATHOGENESIS

- All patients with harlequin ichthyosis have absent or defective lamellar granules and no intercellular lipid lamellae.
- These granules are responsible for secreting lipids that maintain the skin barrier at the interface between the granular cell layer and the cornified layer.
- The lipid abnormality is believed to allow excessive transepidermal water loss; lack of released hydrolases prevents desquamation, resulting in a severe retention hyperkeratosis

Symptoms of harlequin ichthyosis

- Thick skin plates that crack and split
- Distorted facial features
- Tight skin around eyes and mouth (may force eyelids and lips to turn inside out and affect ability to feed)
- Restricted breathing (when chest or abdomen is affected)
- Hands and feet that are small, swollen and partially flexed
- Deformed ears or ears fused to the head (may appear to be missing)
- High blood sodium levels

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Treatments

This will increase the softness and suppleness of the skin. Oral retinoids are useful in severe cases but their use should be monitored.

The thick plates of skin gradually peel off over several weeks. The skin appears very red and shiny from the peeling.

Physiotherapy benefits some patients who suffer from restricted mobility. Other treatments may be needed for instance, counselling can help patients cope with the psychological challenges of the disease.

Other important aspects include the prevention of infection, dehydration and drying out of the cornea. When required, the skin may have to be incised to allow proper circulation and thoracic movement.

Complications of harlequin ichthyosis

- **External deformities.** The child's appearance is distorted by the rigid skin. Thus some infants may have no apparent external ears, or deformed ears, because the abnormal skin draws them tightly inwards. The eyes and the eyelids are often affected in various ways. In some, the eyelids cannot close properly. In others the swollen eyelids obscure the eye area. The hands and feet may be smaller than usual, with restricted mobility of fingers and toes.

- **Breathing.** Individuals with harlequin ichthyosis often suffer from respiratory problems or even respiratory failure. This is due to the restriction of chest movement by the tight thick skin.