

SPOT DIAGNOSIS (IMAGE GALLERY)



SKIN LESIONS AT BIRTH

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A term male baby was born vaginally with birth weight of 2.9 kg. Baby cried immediately after birth and was shifted to nursery in view of significant skin lesion all over his body. Baby was born of non-consanguineous marriage. Mother's antenatal

history was uneventful but she did not have regular antenatal visits. On examination, as shown in Fig 1, greenish thickened skin layer was present all over his body with multiple deep red cracks throughout, splitting the layer. Severe ectropion was present, mouth was open and fish mouth in appearance, and ears were flat and folded. Hands and feet were swollen and deformed, with fingers short and flexed. Whole body was in slight flexion due to tautness of the skin layer, along with limitation of movements.

What is the diagnosis?

Harlequin ichthyosis. It is an extremely rare and serious congenital ichthyosis with strikingly odd features. Autosomal recessive inheritance is seen. Mutations involving the ABCA12 lipid transporter gene are the cause. (1,2) Infants at birth are encased in a markedly thickened, hard stratum corneum. Soon after birth this cracks, resulting in deep red transverse and longitudinal fissures separating thick yellow plates of skin. The normal facial features are severely affected, with distortion of the lips (eclabion), eyelids (ectropion), ears, and nostrils, along with edematous hands and feet. (2,3) Harlequin ichthyosis was almost fatal in the past. However, infants can survive if good neonatal care with appropriate skin specific care is provided. Infrequently, it may present with a milder phenotype. Impaired skin barrier function is responsible for all further complications. Increased fluid and heat loss can lead to dehydration, electrolyte imbalance, disrupted thermoregulation, sepsis and malnutrition. Due to the membrane, these babies have restricted movements, which results in difficulty in feeding, along with impaired ventilation, hypoxia and pneumonia. Constricting bands of skin can even compromise perfusion and lead to peripheral edema and ischemia. (3) Management includes providing a humidified, temperature controlled environment, daily bathing with water, and liberal applications of emollients such as petrolatum based products, along with treatment of infections. Oral retinoids help by accelerating shedding of the thick skin layer and fasten recovery. (3,4).

References

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