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Harlequin Ichthyosis – A Rare Entity in the New Born

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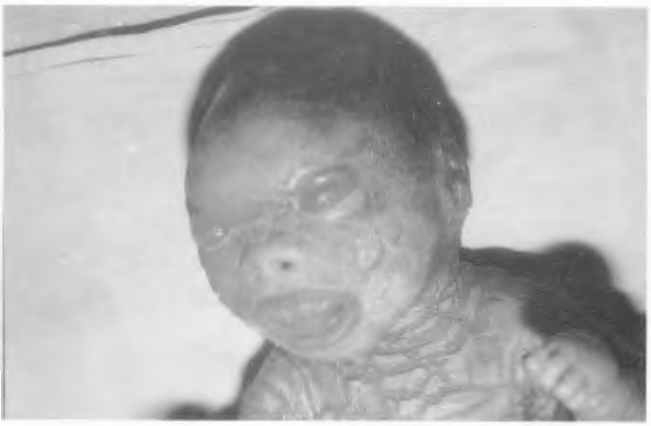
Introduction

Harlequin ichthyosis describes a severe, usually lethal erythrodermic ichthyosis that causes a distinctive and grotesque appearance at birth. The disorder is an inborn error of epidermal keratinization and is usually regarded as a form of nonbullous congenital ichthyosiform erythroderma[1]. Many cases are sporadic with apparent autosomal recessive inheritance, but others occur in consanguineous families in which there may be more than one affected child[2,3]. We describe a case of this rare entity.

Case Report

A male baby was born at our institution in May 2000 at 35 weeks of gestation by normal vaginal delivery as breech presentation. He was the second child of unrelated parents. His sibling was a female child who is alive and well. His birth weight was 2250 gm. At birth, his whole body was encased in a 'Coat of armour' like membrane. Soon after birth (within 2-3 hours), this taut, inflexible cast produced thick yellowish brown firmly adherent plaques which showed deep red fissures. He had severe ectropion and eclabium. His nose and ears were tethered and appeared rudimentary (Photograph 1) Hands and feet were encased in hard mitten like casts and clenched in a flexed position. The scalp felt boggy. The eyebrows and eyelashes were absent. Nails were present on all digits.



Photograph 1 : Showing Harlequin Ichthyosis (Face Close-up)

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He was fed regularly with a spoon and was also given supportive treatment. Paraffin based emollient and oil baths were advised. Artificial tears were instilled into his eyes and oral antibiotics were also given.

On the 10th day, the membrane dried and cracked further. The baby was discharged on request after two weeks. The infant died at 6 weeks of age at home.

Discussion

Since the original description of harlequin fetus in 1750 by the Reverend Oliver Hart, many cases have been described in the literature. Harlequin ichthyosis was invariably associated with stillbirth or neonatal death until Lawlor[4] reported a case in 1985 that survived on treatment with retinoids.

The exact nature of harlequin ichthyosis abnormality is unclear. It has been suggested that it may represent the most severe form of nonbullous congenital ichthyosiform erythroderma or lamellar ichthyosis. Another possibility is that it is a separate form of autosomal recessive ichthyosis. Finally, it may represent a phenotypic expression of several genotypes akin to the situation with a collodion baby as some cases show abnormalities in stratum corneum keratin and epidermal lipids[1].

The clinical appearance of harlequin ichthyosis is distinct. The criteria for diagnosis of harlequin ichthyosis must be clinical because inheritance is not absolutely certain and histopathologic findings are not specific. No consistent biochemical findings have yet been recognized.[3] Our patient had a clinical picture very similar to that of other reported cases.

Most of the infants reported in the literature had died in the first few days or weeks of life, with death

suspected to be singly or in combination due to septicaemia, pulmonary infections, mechanical restriction of breathing and disordered temperature regulation. Many of these infants were born prematurely which almost certainly contributes to their early death . Our case was also born preterm and died at 6 weeks of age at home and therefore the cause of death could not be ascertained.

Although, it is now possible to keep harlequin infants alive with intensive neonatal skin care and treatment with retinoids, the severity of the persisting dermatosis may result in a lifetime of suffering for the saved individuals.

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