Indian J. Dermatol. Venereol. Leprol. 1989; 55 : 61-63

HARLEQUIN ICHTHYOSIS

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Autopsies were performed on two cases of harlequin ichthyosis, with typical clinical and microscopic features. Both died of lung compromise. Neutral fat in the stratum corneum could be demonstrated in one. This feature has been documented in only one previous case report. Routine laboratory investigations were within normal limits.

Key words : Harlequin, Ichthyosis, Autopsy.

Harlequin foetus is the most severe form of congenital ichthyosis. The name harlequin refers to the clown-like appearance of the face, and the resemblance of the skin lesions to the traditional costume of the harlequin. It is a very rare disorder; an accurate incidence is difficult to ascertain because of lack of documentation, and confusion with other forms of ichthyosis.[1] The pathogenesis is unknown; it is inherited as an autosomal recessive disease, and the lesions are present at birth. There is a paucity of cases with parental consanguinity.[1]

Case Reports

Case 1

An autopsy was performed on a 2-month-old male, born of a first degree consanguinous marriage. The gross appearance was classical. The entire body, including the flexural creases, was covered with a thick dark-brown horny cuirass, which spared only the palms and the soles. There were deep intervening fissures. All the four limbs were in flexion, due to the extreme inelasticity of the skin. The face was grotesque, with rudimentary ears, bilateral ectropion with exposure keratitis, absence of eyebrows and eyelashes, eclabium, a broad nose with patent nares, and furrows about the mouth and chin (Fig. 1).

Microscopic features (Fig. 2) were also typical, with marked hyperkeratosis; at places the stratum corneum was upto twenty times as thick as the stratum Malpighii. The granular layer was composed of a single layer of cells and contained keratohyalin granules. There was no appreciable parakeratosis or papillomatosis. Hair follicles were normal.

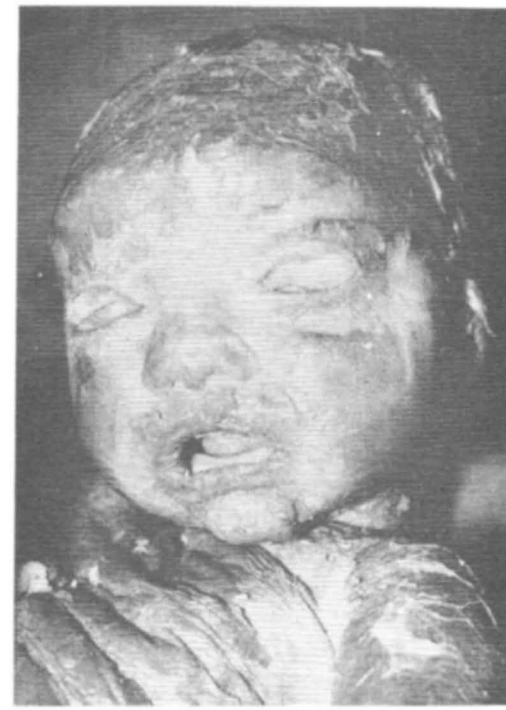

Fig. 1. The face in harlequin ichthyosis.

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Fig. 2. The characteristic marked hyperkeratosis, with a thin granular layer, and a sparse upper dermal lymphocytic infiltrate.

There was a sparse lymphocytic infiltrate in the upper dermis.

Routine laboratory investigations were within normal limits. A Sudan III stain for fat was positive in the stratum corneum. Examination of the viscera at autopsy revealed no other congenital anomaly. The cause of death was a haemorrhagic interstitial pneumonitis.

Case 2

An autopsy was performed on a 2-day-old female. There was no history of consanguinity. The gross and microscopic findings were identical to the foregoing, the only additional feature being the presence of stunted digits involving all the four limbs (Fig. 3). At autopsy, both ovaries were rudimentary. There was no other congenital abnormality. The stain for fat, which requires the use of fresh tissue, was unfortunately not done at the time. Routine laboratory investigations were normal. The baby died of a haemorrhagic bronchopneumonia.

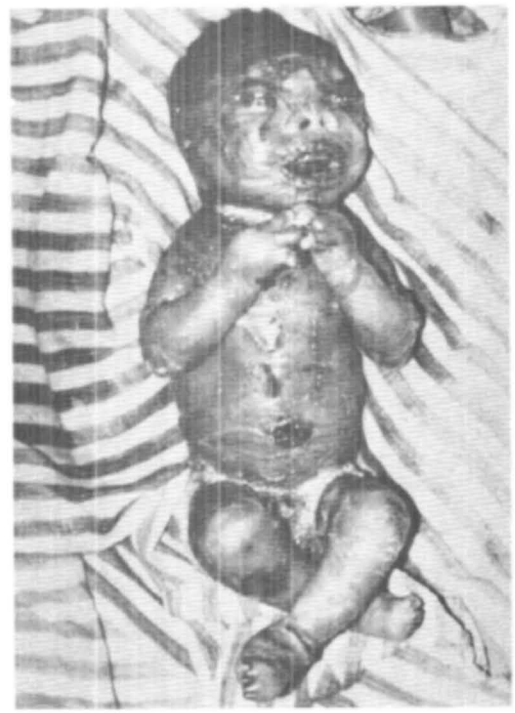

Fig. 3. Harlequin foetus with stunted digits involving all the four limbs.

Comments

The diagnosis of harlequin foetus is no great problem. The striking appearance at birth coupled with the fairly characteristic histopathology is pathognomonic.[1-5] There have however, been studies demonstrating the variable nature of the keratinisation pattern, which suggest that this disorder may have different causes.[3] Stunted digits, as seen in case 2, are an occasional feature, and are due to the pressure atrophy caused by thick hyperkeratotic tissue.[1] The significance of rudimentary ovaries is not known. The only previous documentation of fat positivity in the stratum corneum was by Buxman et al[2] in 1979. They postulated the cause to be a defect in the intraepidermal lipid metabolism. The value of demonstrating fat in the stratum corneum as a diagnostic confirmation, needs to be established. As is invariable,[1] routine

laboratory investigations were normal in both our cases. Affected infants usually die within two months of birth, though a case of exceptional longevity, in which the baby survived nine months, has been reported.[2] The cause of death[2] in these babies is generally attributed to lung compromise, sepsis from deep fissures, or pneumonia from mechanical restriction of breathing, sometimes combined with aspiration of abnormal scales into the lungs before birth. Dehydration because of the abnormal skin, and inadequate feeding due to inability to suckle properly, are inevitable complications.

Both our cases were in hospital for only two days before death intervened. Both received only supportive treatment. No treatment is of known benefit. While lubrication and protection from infection may prolong life, the palliative value of oral retinoids has not been established.[1] Intrauterine diagnosis by foetoscopy and foetal skin biopsy is possible,[6] and should be of great value in prenatal counselling.

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