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CASE REPORT

Ichthyosis Fetalis*

by

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Abstract

A severe variety of ichthyosis fetalis or Harlequin fetus is reported with a brief review of the literature. It seemed that our case, Tadjuddin's (Jakarta), and Wong Hock Boon's (Singapore) assure us that the Harlequin fetus can also be seen in the tropics and in all races where the ichthyosis gene is present.

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Introduction

The severe form of ichthyosis fetalis or Harlequin fetus is a remarkable and very rare genetic disease of the skin which developes intrauterine in the ... and fifth month of gestation. The first description of this condition is probably that of the Reverend Oliver Hart of Charlestown, South Carolina, U.S.A., who in 1750 recorded in his diary the description of a newborn infant with this severe type of skin disease. His description, which bears repetition, fits in our case perfectly and is an excellent piece of clinical reporting (Esterly, 1968) :

"The skin was dry and hard and seemed to be cracked in many places, somewhat resembling the scale of a fish. The mouth was large and round and wide open. It has no external nose but two holes where the nose should have been. The eyes appeared to be lumps of coagulated blood, turned out, about the bigness of a plum, ghastly to be held. It has no external ears but holes where the ears should be. The hand and feet appearing swollen, were crumpt up and felt hard. The back part of the head was much open. It made a strange kind of voice, very low, which I cannot describe. It lived about eight and forty hours and was alive when I saw it."

Most descriptions of the condition which are similar to the case mentioned above are from white infants. Jelliffe (1970) described that the severe form of ichthyosis has never been seen in the tropics. Tadjuddin and Hendarto (1967) described a case of a Javanese female infant; Wong Hock Boon (1967) assured that the condition could be seen in all races.

Case report

B., a 3½-hour-old Indonesian female infant, was admitted to the Department of Child Health because of her 'very strange'-looking skin. The child was born at home and the youngest of 6 children. One elder brother and one elder sister died at the age of 3 and 5 days respectively, but their skin had no abnormalities. Another elder sister was said to have a similar skin condition, but was born dead, while other siblings had normal skins. Both parents were from Sumatra (tropical area); the father was 30 years old, and the mother 29. Both looked healthy and normal; they are cousins.

Physical examination on admission revealed a female newborn with a body weight of 2,400 gm., length 42 cm., and temperature 36° C. The skin over the whole body was thick, yellowish-white, and exfoliated, and both wrists and ankles looked erythematous. On palpation the skin felt somewhat dry, scaly and hard. The skin on the body folds was fissured; the cracks were 2 and 3 mm. deep in some places. The underlying skin appeared red and not grossly abnormal in the cracks, there was some blood-stained ooze in these areas. The abdominal wall was severely affected, while the skin of the back was less thickened. The nose was rather flat, the

ears were deformed and completely covered with 2 to 3 mm. thick horny yellowish scales. The external meatuses were also occluded. Both hands and feet were claw-like. Her wrists and ankles were covered with a bullaelike lesion which felt tense and hard.

Ectropion of the upper and lower eyelids was so severe that there was complete eversion of both lids. The mouth was shaped like an O due to hyperkeratosis. The infant lay with both arms flexed at the shoulders and elbows and the fingers were claw-like. The thighs and knees were also flexed. This extreme flexion was due to the very thickened, inelastic skin (Figure 1). Bands of hyperkeratotic epidermis were present on several fingers and toes, causing deformities. The patient's progress was marked by extension of fissuring and some loss of large flakes of hyperkeratotic skin, leaving a somewhat reddish, granular skin underneath. This 'normal-looking' skin persisted until death (Figures 2 & 3). Radiological examination of the heart, lungs, and bones revealed no abnormalities. The patient was given milk by nasogastric tube. She passed meconium on the day of admission, that is, at about 18 hours of life, and thereafter she passed one to two stools per day.

The child died on the 4th day of hospitalization. During her stay she had not passed any urine. Her body weight had increased from 2,400 gm. to 2,540 gm. and her temperature remained 36° C. On the 3rd day of hospitalization necrosis developed on the distal phalanges of her hands and feet. Necropsy was not performed because permission was not granted by her parents. Skin biopsy revealed only a thick keratine layer, because it was done too superficially. Intravenous pyelogram could not be done.

Discussion

The name of Harlequin fetus is applied to the infant, either because of the clownlike appearance of the face with the round O of the mouth or may be of the deep fissuring of the skin, often with diamond-shaped or triangular plaques on the trunk and limbs resembling the traditional costume of a Harlequin. Ichthyosis is a relatively uncommon skin condition that has significant ocular manifestations (Sever et al., 1968; Jay et al., 1968; Katowitz et al., 1974). Classification has been made by many authors, based on genetic and clinical criteria (Wells and Kerr, 1965) and histopathological findings. For convenience, we subdivide it into 4 types :

1. Autosomal dominant ichthyosis vulgaris.
2. Congenital autosomal dominant ichthyosis, with a generalized bullous form and a localized non-bullous form (ichthyosis hystrix).
3. Sex-linked recessive ichthyosis vulgaris (the rarest).
4. Congenital autosomal recessive ichthyosis, with a severe 'Harlequin' ty-

pe and a less severe 'lamellar' type (congenital ichthyosiform erythrodermal non-bullous type) (Etserly, 1968).

All these 4 types have in common symptoms of dryness of the skin with variable amounts of profuse scaling. Only in the autosomal recessive type does ectropion of the lids and conjunctival changes develop. The presence of familial cases and consanguinity among the parents or some of the subjects strongly suggest that the condition is inherited as an autosomal recessive trait. Our case had the typical clinical findings, epidermal histopathology (not complete) and course.

Pathological studies of the skin showed marked thickening of the stratum corneum, but no other distinctive changes. Electron microscopic studies of the epidermis showed the presence of intracellular filaments which were attached to the cell wall. X-ray diffraction analysis of the horny layer revealed the presence of a cross-beta-fibrous protein rather than the usual alpha-protein. The epithelial surface of other organs appeared normal. The thymus gland was involuted and showed changes in the corpuscle (Craig et al., 1970).

Abnormalities were also observed in the conversion of cartilage to bone (Craig et al., 1970). Kessel and Friedlander (1956) reported their Harlequin fetus with lower nephron nephrosis at necropsy. Baden et al. (1975) studied the stratum corneum of different types of ichthyosis. An alpha X-ray diffraction pattern and the solubility of the alpha fibrous protein of different types of ichthyosis appeared to be the same as in the normal stratum corneum. Sodium dodecyl sulfate (SDS) — polyacrylamide electrophoresis of the fibrous protein showed variable patterns in different types of ichthyosis, while amino acid analysis of the protein was quite similar to that of the normal stratum corneum. These data suggest that the fibrous protein in ichthyosis is abnormal, but further studies on the individual polypeptide chain are necessary to rule out more subtle differences (Baden et al., 1975).

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FIG. 1 : *Harlequin foetus. Multiple fissures are separated by thick scales. There is eversion of the lids and the fish-mouth appearance of the lips is characteristic.*

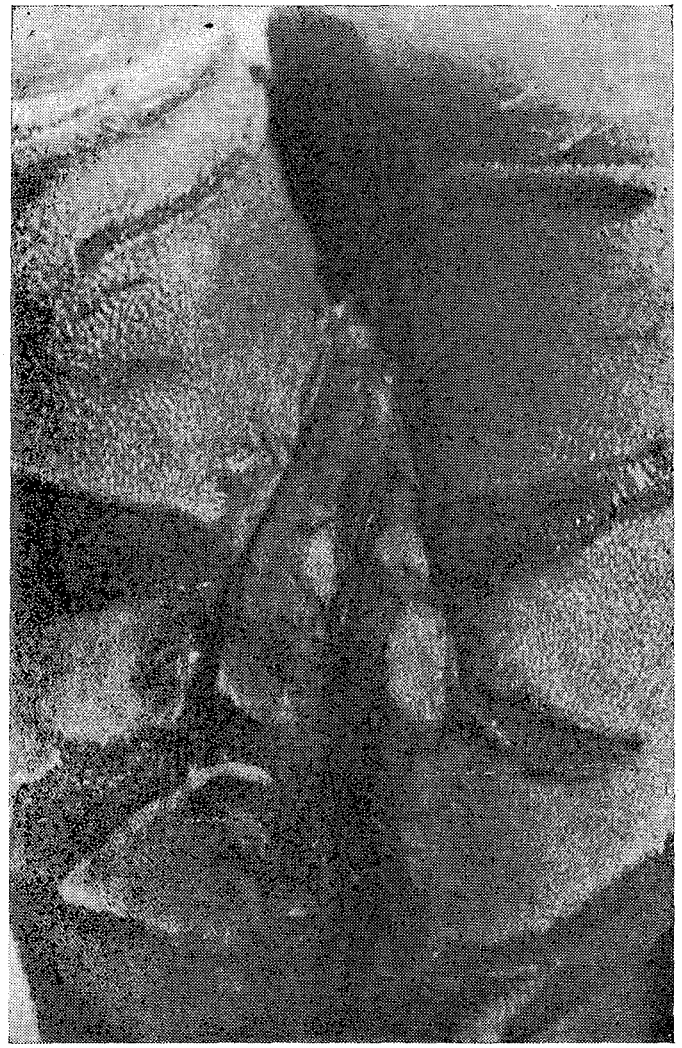


FIG. 2 : *The same patient from another view.*

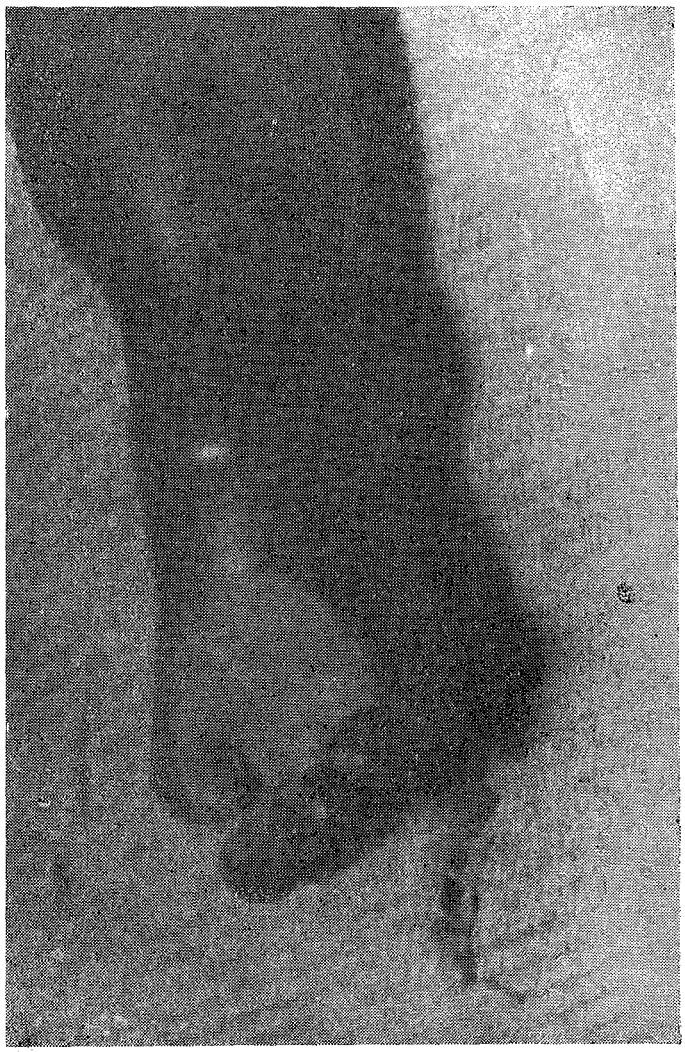
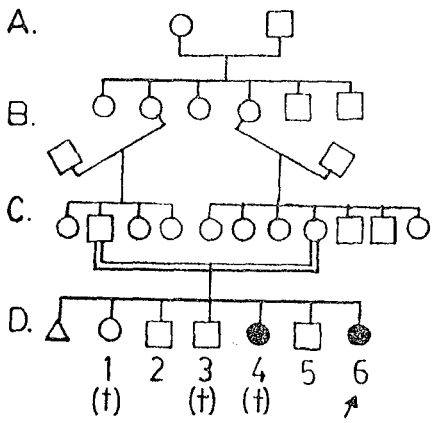


FIG. 3 : *Claw-like hand of the same patient.*

PEDIGREE OF THE CASE



△. abortion

D_4. the skin was said to be similar to D_6. but born dead.

consanguin marriage.

D_6. proband

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