# HARLEQUIN BABY

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The most severe form of ichthysosis congenita is designated as ' Harlequin-Baby ' because in this condition, the skin has a horrid resemblance to the ragged garments of a circus clown. In the gravest form of ichthyosis congenita the child may be still born. This is a rare lesion in any part of the world and most of the reports of this lesion are only single case reports. Athavale (1961) and also Manshramani et al (1965) reported cases of Ichthyosis congenita. Full autopsy reports are very few on these cases. Lattuda and Parker (1951) reported an autopsy study. Pathology of the skin in this lesion has been studied by many and Ormsby and Montgomery (1954) described the pathological change extensively. we had occasion to do a complete autopsy on a ' Harlequin ' foetus and the changes observed are given below.

*Case*: A dead born female baby was brought to the hospital by the mother because of the peculiarity of the baby. The mother had a normal delivery and no other history was available,

The baby weighed 2.1 kgs, There was incomplete flexion of all limbs and all joints. The skin of the whole body was parchment like, yellowish, thick and rigid. wide red cracks were present in the skin giving the fetus a tigroid appearance (Fig. 1) The palms and soles were rounded and cracked by the hyperkeratinised process, The nostrils and opening of the ears were almost plugged by hyperkeratinised non desquamating skin. There was microphthalmia and eversion of eyelids. No eyebrows were seen. The mouth was small and both the lips were everted. Very few hair could be seen on the scalp. Anal opening was almost closed by the keratotic process. The fresh fetus presented an awesome picture.

*Autopsy*: Heart (13 gm.), lungs (Rt. 23 gm. Lt. 18 gm), liver (70 gm.) spleen (6 gm), kidney (7 gm. each), adrenals (3 gm. each), thymus (4 gm.), were all normal. No macroscopic abnormality could be made out internally. Macroscopic change in the skin were as noted before.

*Histopathology:* None of the organs showed any pathologic change. No metaplastic changes could be seen in the tracheal and bronchial lining. Kidney was normal and the lining of the pelvis showed increase in the number of cell layers (11 to 12 layers) but no keratinization was seen. Similarly the lining of the ureter showed increase in cell layers but no kerantinization. Bladder mucosa uterus, cervix' vagina, thyroid, adrenal, liver and spleen were all normal. Muscle of the heart and diaphragm were also normal.

Multiple full thickness sections of the skin were taken from sole and dorsum of foot, leg, thigh, abdomen, chest, neck, face, scalp, back, arm, forearm, palm and dorsum of hand.

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The constant feature in all the areas was hyperkeratosis. There was gross heaping up of the keratin without any desquamation. There was no para-keratosis. There was not much difference in the amount of keratosis of skin in different parts of the body. The cracking of the keratin was up to the level of the dermis in places but stops short of the malphigian layer in some areas (Fig 2). No stratum lucidum could be made out. Stratum granulosum was not present in the sole of the foot, abdomen, palm, back and scalp. Stratum granulosum was present in the dorsum of foot. leg, thigh, chest and forearms. There was varying degrees of thinning of the malphigian layer. There was dovetailing of the rete pegs and papillae (Fig. 3). The rete pegs were needle shaped in some areas. The cells of the basal layer showed swelling and vacuolation here and there.

*Adnexae of the skin*: Sweat glands were normal and present in all the sections studied. There was no atrophy. The only noticeable change was mild dilatation of the sweat glands. There was no keratinization of the glands. Sebacious glands showed variable changes. They were atrophic over the leg, abdomen chest and scalp. The glands were smaller. In most of the areas where hair ought to be present there was keratotic plugging of the hair follicle and as a result of this the sebacious gland also showed atrophy (Figs. 4 & 5). In some areas all that remined was a keratotic plug with a few sebacious cells around. The only area in the body where a few hair could be made out was scalp.

The connective tissue and the blood vessels in the dermis did uot show any change. In the padillae there were dilated capillaries and no inflammatory cells.

*Comment*: Pathological features of ichthyosis congenita has been described (Stowens, 1959; Potter, 1957, Lever. 1954, Ormsby & Montgomery, 1954). Hyperkeratosis with inadequate shedding is the prominent feature described by all. Absence of sweat glands (Stowens, 1959; Potter 1957) to an increase in sweat glands (Ormsby and Montgomery 1954) has been described. Atrophy of the pilosebacious appartus due to keratotic plugging of hair follicles was described by all. Absence of stratum granulosum has been stressed by Ormsby and Montgomery (1954). They also mention that this is an example where horn layer is formed without the intervention of keratohyalin contrary to the belief that granular layer is essential for keratinization. Lever (1954) differentiates ichthyosis congenita and ichthyosis vulgaris by the presence of stratum granulosum in the former condition. Williamson (1934) mentions a case where thyroid was absent. Changes in the adrenals have also been described (Ormsby and Montgomery 1954).

The constant pathological feature was hyperkeratosis and non-sheding of the keratin. As a result cracking of the heaped up keratin occurred. The presence or absence of stratum granulosum was variable. Probably it is present in some areas and not present in other areas. Sweat glands were invariably

present and normal and the pilosebacious apparatus showed hyperkeratotic plugging. The controversial nature of some of tne findings in literature like presence and absence of stratum granulosum and sweat glands is probably due to not taking enough number of sections from all parts of the body. Except for the hyperkeratosis and non-shedding of keratin there is no pathological feature which is consistent in all the parts of the skin and we are in conformity with Bloom and Goodfried (1962) that unless we know the normal factors of keratinization and shedding of kereatin we may not know the pathogenesis of this lesion.

We thought that the lining epithelium of the respisatory tract or the genitouriary tract might show keratinization but they were essentially normal except for an increase in the number of layer of cells in the pelvis of the kindney and ureter. No changes were seen either in thyroid or adrenals.

Various theories have been put fourth to explain this condition such as due to absence of amniotic fluid (Kebrer), endocrinal disturbances (Ormsby and Montgomery, 1954), persistence of the "epitrichial layer" (Bowen 1895) familial in nature (Briceno-Maaz, 1963, Bloom and Goodfried 1962. But none of the above explains the peculiar change in the skin of the newborn. Unless we know more about the normal process of keratinization and shedding of the keratin flakes we may not be able to understand the nondesquamative hyperkeratinization seen in this condition.

*Summary*: An autopsy study of Harlequin fetus is presented.

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