Harlequin ichthyosis

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Citation

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Abstract

We describe a case of Harlequin Ichthyosis

CASE REPORT

A 24 years aged 3 rd gravida mother gave birth to a male, 34 wks, appropriate for gestational age baby with birth weight1.7kg, length 41.5cm. and head circumference 30.4cm. Mother had history of previous two foetal loss, one at 12 wks and another at 16 wks of pregnancy. There was no history of consanguinity. The baby had markedly thickened, adherent skin like “coat of armour “. The skin was cracked with deep red fissures almost all over the body. There was ectropion and both eyes were obscured by everted and edematous eyelids. There was eclabium and baby was unable to suck breast. Head appeared microcephalic . Nose and ears appeared rudimentary because they were tethered with the thickened skin. There was depressed bridge of nose. There were contracture of both upper and lower limbs. Digits of the hands and feet were partially obscured by thickened and edematous skin of palms and soles. From the above clinical features a diagnosis of Harlequin Ichthyosis (HI) was made(Fig - 1). Karyotyping was not done in this patient. The baby died on 8th day of his age due to septicemia.

DISCUSSION

HI is a severe usually fatal congenital erythrodermic ichthyosis. [1] .Incidence of this condition is about 1 in 300000. Most cases of HI are sporadic with autosomal recessive inheritance. This condition is characterized by plaques, measuring up to 4 or 5 cms on a side, that have a diamond-like configuration resembling the suit of a harlequin clown. There is also ectropion, eclabium and contracture of limbs. The skin development is altered in utero, hyperkeratosis of the hair canal occurs in the second trimester & characteristic ultrastructural abnormalities are expressed in the affected foetal epidermis. [2,3]

There is defect in keratin expression and epidermal lipid deposition in HI. Skin morphological abnormalities reflect a defect of lipid content of lamellar granules: absent or abnormal lamellar granules in the granular cells, lipid droplets in the stratum corneum & a lack of extracellular lipid lamellae have been reported in harlequin foetus. [4] There is abnormal although variable keratin and filaggrin expression suggestive of genetic heterogeneity. [5] Large intragenic deletion & frameshift deletions has been found at ABCA12 gene, in 11 of 12 screened individual with HI in a study by Teruki Yanagi et al. [6,7] Three subtypes have been detected. [1,5] In type 2 & 3, profilaggrin can not be converted to filaggrin because of block in post translational processing. There is defective dephosphorylation secondary to abnormal protein phosphatase activity. In type 2 HI abnormal expression of protein phosphatase 2 Ac was found. Low reactivity was also seen in type 1 cases. Now early prenatal diagnosis is possible by direct sequence analysis & restriction enzyme digestion analysis usuing foetal genomic DNA from amniotic fluid cells at 16 wks of gestation. [7]

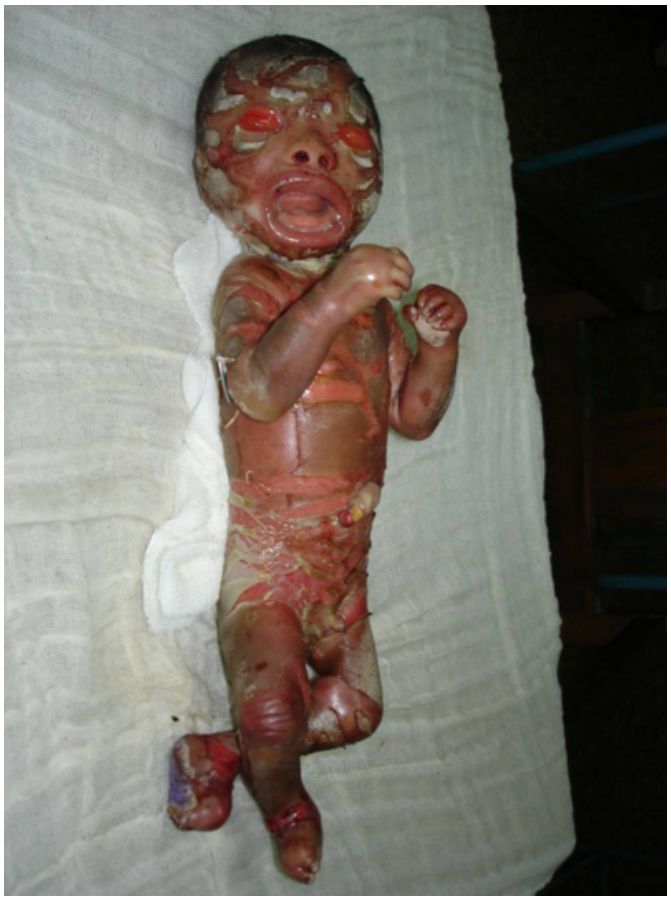
Differential diagnosis should be done from collodion baby and from restrictive dermopathy. Collodion baby is a less severe form of icthyosis than HI. In restrictive dermopathy there is skeletal and facial abnormalities. [1]

HI affected neonates usually die within few days because of feeding problems, bacterial infections, and/or respiratory difficulties. There is transepidermal water loss, impaired thermal regulation & increased risk of secondary infection. With the wider availability of neonatal intensive care facilities & benefits from oral retinoids, some of the babies with HI are surviving beyond neonatal periods. .Though, this is usually associated with still birth or early neonatal death but survivors at 9 Yrs of age were reported to be of normal intelligence and has discontinued retinoid therapy. [1,8] A well

coordinated multidisciplinary approach is required to manage such babies. Management of HI involves , care of the skin, eyes & fluid-electrolyte balance and surveillance against infections. Constant support to & counseling of parents is also essential. In survivors the disease comes to resemble severe nonbullous congenital ichthyosiform erythroderma. [8]

Figure 1

Figure 1: Baby with Harlequin Ichthyosis.



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