HARLEQUIN BABY

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INTRODUCTION

Harlequin ichthyosis is a severe and very rare erythrodermic ichthyosis listed as Disorder of Cornification DOC -7 [1, 2]. It heralds clinically as distinct monstrous appearance of baby at birth. Defective keratin expression and lipid deposition has been observed histologically [3, 4]. Skin pattern and lethality incites the label of harlequin i.e. Venomous snake from genus Elaps [5], however survival is now possible [6, 7]. Many cases are sporadic with apparent autosomal recessive inheritance, others occur in consanguineous families. More than 100 cases have been reported in literature [8].

CASE REPORT

A multi-parous female from low financial family of Sialkot, reported in Emergency Room, CMH Hyderabad with Labour pains at 28th week of gestation. Ante-natal Sonography revealed breech lie of fetus and cleft palate. At Labour room she delivered a female baby with Apgar score 6/10, weight 1.8 Kg, exhibiting dysmorphic features of boggy scalp, eclabium, cleft palate, rudimentary nose and ears, closed eyes, mitten cased hands and feet deformities and cracked, deeply fissured skin covering the whole body (fig.1). Rest of systemic examination was unremarkable. Baby was diagnosed as Harlequin and nursed in humidified incubator at intensive care unit. Temperature of 32-33oc was maintained, topical care by application of white soft paraffin and liquid paraffin was provided, feeding was done using nasogastric tube. Feeding requirement was increased with passing days. On fourth day she developed foul smell so blood sample and skin swab for culture & sensitivity were ordered. In the meantime she was given a warm saline bath to eradicate the foul smell and on the same day suspension Co-amoxiclave was commenced. However she remained afebrile and no growth of organisms after 48 hours of incubation was observed. On 7th day her intake reduced and the condition gradually deteriorated. Despite adequate measures she expired at the age of 10 days. The couple was consanguineous. Baby was the fourth of other normal siblings with no history of ichthyosis, skin disorders, intrauterine or neonatal deaths in the family. No interventional prenatal procedure (e.g., amniocentesis, and fetal skin biopsy) was performed.

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DISCUSSION

Harlequin ichthyosis is a lethal devastating most probably autosomal recessive disorder [2] characterized by an extremely thickened keratin layer of skin that is cracked, fissured & there are deformed facial and acral features [8]. The term Harlequin derives from the newborn's facial expression and the triangular and diamond-shaped pattern of hyperkeratotic plaques. The newborn's mouth is pulled wide open, mimicking a clown's smile [9]. The first such report is from the diary of Rev. Oliver Hart, of Charleston, South Carolina, who described these features in 1750 [2]. Frequency is approximately 1 in 300,000 children [8]. These children are at great risk during the neonatal period and often dies shortly after birth. Abnormal water loss through the skin and poor temperature regulation lead to risk of fluid and electrolyte imbalance and are also at risk for infection beginning in the skin. However, because of poor temperature regulation, do not show the usual signs & symptoms of infection [10]. Normal respiration may be restricted by the taut skin [11]. The skin of those who survive the newborn period usually resembles the skin of those with a severe phenotype of Cogenital Ichthyosiform Erythrodermac. [12]. Because no mutation has been identified, molecular genetic diagnosis is currently impossible. Prenatal Sonography diagnosis has been described, with findings of a persistently open mouth, echogenic amniotic fluid, and fixed flexion of the extremities [9,10]. Amniotic fluid samples obtained as early as 17 weeks' gestation have demonstrated hyperkeratosis and abnormal lipid droplets within the cornified cell [8]. Prenatal diagnosis is also possible by fetal skin biopsy

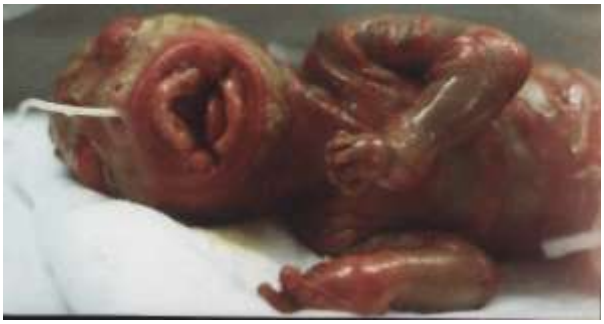

Fig 1: Harlequin Baby

[6,13,14]. Advances in neonatal intensive care together with facilitating desquamation with judicious use of systemic retinoid therapy have led to improvements in survival [6,15]. The newborn may require intensive care with fluid and electrolyte monitoring. The newborn with extensive fissuring is prone to bacterial infection and sepsis, and carefully chosen topical and systemic antibiotics can minimize the extent of infection [14].

CONCLUSION

The facade of the neonate can be dreadful to parents and health care providers. Detailed genetic counseling and prenatal diagnosis of this disorder may help prepare them. Psychological support to the family especially the mother is essential to prevent psychosocial trauma.

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