Website : www.ijomcr.net
Email :ijomcr@gmail.com

Indexed Journal

International Journal Of Medical Case Reports

Harlequin Baby- A Case Report

Authors

***Dr Sanjoy Chowdhury[1], Dr Hitesh Patel[2], Dr Pratik Bhosale[3], Nilanjan Chowdhury[4]**

[1]Joint Director, Medical & Health Services, Bokaro Steel City Jharkand. [2,3]DNB Residents, Bokaro General Hospital, Jharkhand, India. [4]Final MBBS student Sikkim Manipal Institute of Medical Sciences, India.

Corresponding Author

***Dr Sanjoy Chowdhury**

Joint Director, Medical & Health Services, Bokaro Steel City Jharkand. India

ABSTRACT

Disorders of cornification (ichthyosis) are a primarily group of genetic conditions which are characterized clinically by scaling and histopathologically by hyperkeratosis. Collodion baby (ichthyosis) and harlequin babies are types of these disorders. These disorders may cause considerable disfigurement in affected children and source of a great psychological stress to caretakers. Early diagnosis is helpful to predict prognosis and to provide supportive management for the patients and families. According to Various case reports the mortality of collodion babies varies from 9 to 45% with the highest mortality rates seen in harlequin ichthyosis. We present here a rather rare case report of a harlequin baby who expired after 1 hour of birth. This case also highlights difficulties the attending pediatrician may face during resuscitation of a harlequin baby. Protruding tongue and various facial anomalies may make intubation impossible for the purpose of resuscitation.

KEYWORDS: Harlequin ichthyosis, Collodion baby, ABCA12

Introduction:

The term collodion membrane was first introduced by Hallopeau and Watelet[1] but first time clinically described by Perez in 1880. Harlequin ichthyosis is rare autosomal recessive (autosomal dominant and sporadic cases also reported). Keratinizing skin disorders are characterized by severe thickening of stratum corneum with incidence rate of 1 in 300000 births[2]. As per scientific reports in 2007, 101 cases have been reported in worldwide medical litrature[3]. HI also affects eye, ear, nose, mouth and other parts of body. Eye manifestations include conjunctival chemosis, blepharophimosis, hypertelorism, cracked eyelid skin, oedema of eyelids and ectropion that may leave eyes open and cause infections. Till date big challenging questions to medical science is how to prevent harlequin baby and how to reduce mortality after delivery of harlequin baby[4]. These babies are prone to develop dehydration due to increased transdermal water loss[5]. Usually the deaths of these babies occur in early neonatal period due to sepsis, electrolyte imbalance, hypoventilation, malnutrition and dehydration but some of them may live longer[6].

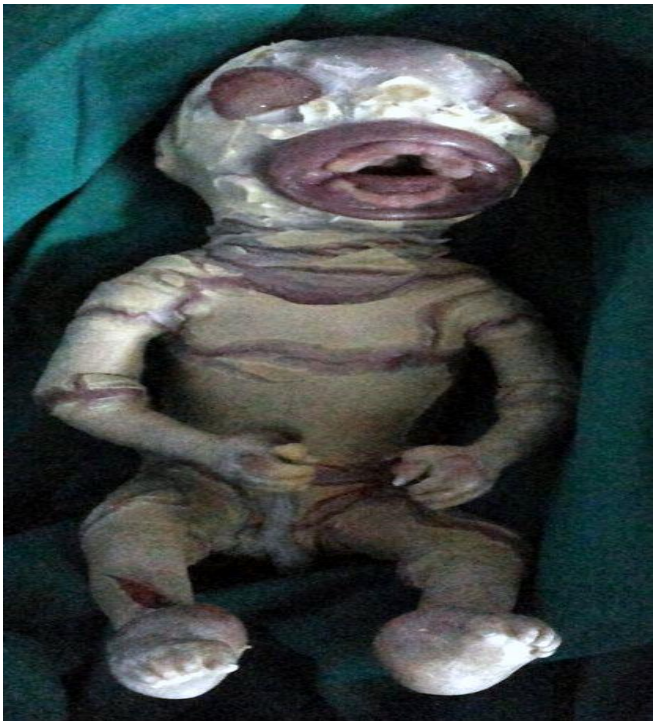
Harlequin baby means baby with body features and facial expression resembling 17thcentury comic actor that usually used to dress in multicolored, masked and diamond patterned tights and term collodion baby means phenotype that can be characterized by a yellow, shiny, tight parchment-like membrane stretched over the skin. Observers may sometimes use the descriptor "dipped in hot wax"[7]. Harlequin ichthyosis is a genetic disease due to mutation in ABCA12 gene (adenosine triphosphate binding cascade) on chromosome 2q35 thought to be involved in transport of lipid that act as protective barrier against pathogen. Harlequin ichthyosis cannot be prevented but can be diagnosed prenatally by fetal skin biopsy, chorionic villus sampling and three-dimensional ultrasonography which may give parents a choice about whether or not to continue pregnancy with an affected child. In the near future gene therapy, PCR techniques and Preimplantation diagnostic techniques may make it quite possible to do implantation of an unaffected embryo so as to prevent harlequin ichthyosis in couples who already have a history of giving birth to an affected child.

Case Report

A third gravid, 28 years old unbooked Muslim woman gave birth to a preterm male child by caesarean section. At the time of delivery, ie on 29 may 2015, the gestational maturity of the newborn was 32 weeks. Gestational age was confirmed by history of last menstruation period, per abdominal and per vaginal examinations and by ultrasonography. Family history revealed that the first child was male baby delivered at term and was live and healthy while second was also male baby delivered prematurely by normal vaginal delivery at home. The baby expired on day 2. She had no documented record of past pregnancy and no record of antenatal checkup of present pregnancy. There was no history of consanguinity.

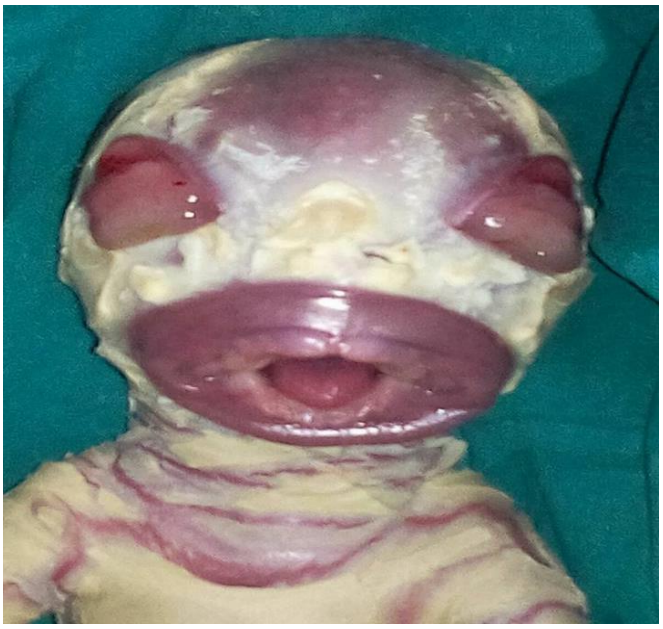
The baby's birth weight was 2000 gm., length 45 cm, head circumference was 35 cm.The baby had immediately cried after birth but due to repeated apneic spells baby was immediately transferred to SCBU. On examinations, baby was covered by shiny yellow cellophane like membrane which taught the body features and gave pugilistic posture to baby (figure 1).

Fig 1 : Pugilistic posture due to contracture of underdeveloped cellophane like membrane is visible.



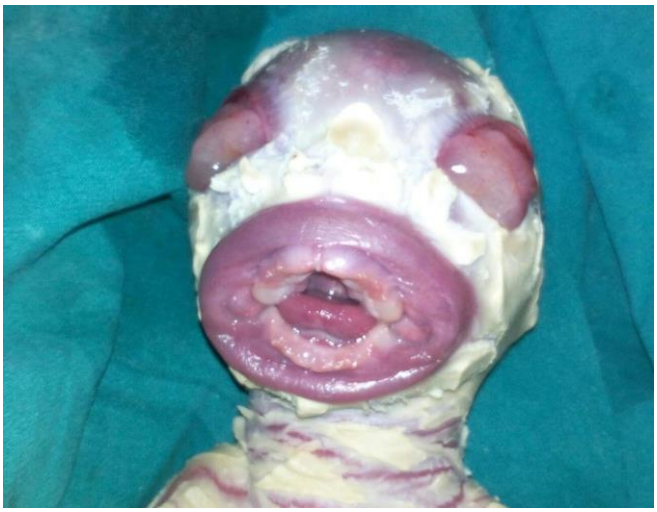
Baby also had cracked skin over entire body, hyperkeratosis of scalp with hair loss, single umbilical artery, rudimentary ears, eclabium, ankle haematoma, fish mouth, protruded tongue and underdeveloped nostrils (Figure 2).

Figure 2:Protruded tongue,rudimentary ear and nostrils with absent scalp hair



On ocular examination, baby had extreme blepharophimosis, hypertelorism and severe edematous eyelids with ectropion and conjunctival chemosis due to which a proper ocular examination couldn't be undertaken. (Figure 3).

Figure 3: Harlequin eye feature.



Repeated apneic spells lead to death after 1 hour due to cardiopulmonary arrest. Resuscitation was not possible due underdeveloped nostrils and huge protruded tongue.

Discussion

Harlequin icthyosis was first reported by Oliver hart in 1750[8]. Incidence of harlequin icthyosis is 1 in 300,000 and it is associated with a grave prognosis, as affected neonates usually die within first few days to weeks of life. HI reported in different ethnic groups and in both sex. The risk to future pregnancy is 25% and carrier rate is 1:225 individual. Antenatal diagnosis was first reported by Blanchet-Bardon et al[9] in 1983. Retinoid and their derivatives prevent skin cracking and facilitate desquamation. The primary treatment of such case is stabilisation of ABC (airway, breathing and circulatory compromise). Sterile lubricant is used to keep skin moist and soften skin that facilitates desquamation. For medication and fluid management umbilical vein is best option. The eyes are protected from exposure due to ectropion by artificial tear and antibiotics. The ectropion is treated in later stage by surgery[10].

Conclusion:

Harlequin icthyosis is rare genetic disease. Due to severe disfigurment and high mortality it disturbs parents' physical, mental and social health. Prevention of recurrence of HI in couples already having an affected child has become possible due to advancement in antenatal diagnostic techniques, PCR, Genetic studies and preimplantation techniques.

Conflict Of Interest : None

Acknowledgement :

Departments of Obstetrics and Gynaecology, Paediatrics, Dermatology, Bokaro General Hospital, Bokaro Steel city, Jharkhand.

REFERENCES

1. D. van Gysel, R. L. P. Lijnen, S. S. Moekti, P. C. J. De Laat, and A. P. Oranje, "Collodion baby: a follow-up study of 17 cases," Journal of the European Academy of Dermatology and Venereology, vol. 16, no. 5, pp. 472–475, 2002.

2. Bianca S, Ingegnosi C, Bonaffini F. Harlequin foetus. J Postgrad Med 2003; 49(1):81–2.

3. Kelsell DP, Norgett EE, Unsworth H, et al. Mutations in ABCA12 underlies the severe congenital skin disease harlequin icthyosis. Am J Hum Genet 2005; 76(5):794–803.

4. Hovnanian A. Harlequin ichthyosis unmasked: A defect of lipid transport. J Clin Invest. 2005;115:1708–10.

5. Taïeb, A. and Labrèze, C. (2002), Collodion baby: what's new. Journal of the European Academy of Dermatology and Venereology, 16: 436–437. doi: 10.1046/j.1468-3083.2002.00478.

6. Hazuku T, Yamada K, Imaizumi M, Ikebe T, Shinoda K, Nakatsuka K, et al. Unusual protrusion of conjunctiva in two neonates with Harlequin Ichthyosis. Case Rep Ophthalmol.2011;2:73–7

7. M. R. Judge, "Collodion baby and Harlequin icthyosis," in Textbook of Pediatric Dermatology, J. Harper, A. Oranje, and N. Prose, Eds., pp. 118–125, Blackwell, Malden, Mass, USA, 2nd edition, 2006.

8. Gurses D, Kilic L, Baskan M. A case of Harlequin fetus with psoriasis in his family. The Internet Journal of Pediatrics and Neonatology 2001; 2 (1): 1-7.

9. Akiyama M, Suzumori K, Shimizu H. Prenatal diagnosis of harlequin icthyosis by the examination of keratinized hair canals and amniotic fluid cells at 19 weeks estimated gestational age. Prenat Diagn 1999; 19(2):167–1.

10. Craiglow, Brittany G. "Ichthyosis in the Newborn." *Seminars in perinatology*37.1 (2013): 26–31. *PMC*. Web. 18 Sept. 2015.