



## Ocular Findings in a Harlequin Baby: A Very Rare Case

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### Abstract

Harlequin ichthyosis is a severe congenital disorder with incidence of 1 in 300000 births. Oedematous lids with ectropion, hypertelorism and sometimes poorly developed eyes are seen in Harlequin babies. Reporting such cases stimulate genetic research by developing a data base. We report a case of Harlequin baby who survived for hours to discuss ocular features and difficulty in management.

**Keywords:** Cyclopamine; Neuroblastoma; Sonic Hedgehog; CD133; CD15

### Introduction

The term collodion membrane was first introduced by Hallopeau and Watelet but first time clinically described by Perez in 1880 [1]. Harlequin ichthyosis is rare autosomal recessive (autosomal dominant and sporadic case also reported) keratinizing skin disorders characterised by severe thickening of stratum corneum with incidence rate 1 in 300000 births [2]. As per scientific literature report in 2007, 101 cases have been reported in worldwide medical literature [3]. HI also affecting eye, ear, nose, mouth and other part of body. Eye manifestations include conjunctival chemosis, blepharophimosis, hypertelorism, cracked eyelid skin, oedema of eyelid 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.

Harlequin baby means baby with body features and facial expression resemble 17th century comic actor that usually dressed in multi-coloured, 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" [4]. Harlequin ichthyosis is 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 diagnose prenatally by foetal skin biopsy, chorionic villus sampling and three-dimensional ultrasonography but in the near future pre-implantation diagnosis may be available to prevent harlequin baby. In pre-implantation diagnosis, *in vitro* fertil-

ization of normally fertile couple will undergo and then test each of embryos for harlequin ichthyosis gene, before they are transferred to mother's womb. Prenatal diagnosis is difficult, costly and not widely available but is still an opportunity for parent who already had harlequin baby.

### Aim

To support genetic research about harlequin ichthyosis for its prevention in future and help the families that affected by harlequin baby in past.

### Case Report

28 years old unbooked Muslim women with G<sub>3</sub>P<sub>2</sub> (?) was delivered baby by caesarean section at 32 weeks of gestation on 29 May 2015. Gestational age was confirmed by history of last menstruation period, per abdominal and per vaginal examinations and by ultrasonography. History of first male baby delivered at term that live healthy and second male baby revealed homemade preterm delivery who expired after 1 day.

She had no record of past pregnancy and no record of antenatal check of present pregnancy. No history of consanguinity. A baby with weight 2000 gm., length 45 cm, head circumference 35 cm and Apgar score 1/10 with heart rate less than 100/min. The baby had immediately cried after birth but due to repeated apnoeic spells baby was immediately transferred to Special Care Baby Unit. On examinations, baby covered by shiny yellow cellophane like membrane which taught the body features and gives pugilistic posture to baby (Figure 1). Baby also had cracked skin over entire body, hyperkeratosis scalp with hair loss, single umbilical artery, rudimentary ear, eclabium, ankle haematoma, fish

mouth, protruded tongue and underdeveloped nostril (Figure 2). On ocular examination, baby had extreme blepharophimosis, hypertelorism and severe oedematous eyelid with ectropion and conjunctival chemosis due to which eye was not opened for detailed ocular examinations (Figure 2). Repeated apnoeic spells lead to death after 1 hour due to cardiopulmonary arrest. Resuscitation was not possible due underdeveloped nostrils and huge protruded tongue.



**Figure 1:** Harlequin Baby Born on May 2015. Pugilistic Posture of Harlequin Baby Due to Contraction of Ichthyotic Skin.



**Figure 2:** Ocular Features in Harlequin Baby: Hypertelorism, Ectropion, Chemosis, Difficulty in Eversion of Eyelids.

## Discussion

Harlequin ichthyosis was first reported by Oliver Hart in 1750 [5]. Incidence of harlequin ichthyosis 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 in 1983 [6]. Retinoid and their derivatives prevent skin cracking and facilitate desquamation. Similar ocular findings (hypertelorism, cicatrice ectropion, complete eversion of the upper eyelids, Conjunctival prolapse limiting visualization of the globes etc.) have been noted [7]. After initial resuscitation on 40<sup>th</sup> day, this child was treated with artificial skin graft (Apligraf) to release ectropion and repeat surgery was done at 61<sup>st</sup> day. He expired due to respiratory failure at 6 months age.

The primary treatment of such case is stabilisation of ABC (airway, breathing and circulatory compromise). Sterile lubricant is used to moist and soft 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.

## Conclusion

Harlequin ichthyosis is rare genetic disease. Due to high morbidity and mortality it disturbed parent's physical, mental and social health. If a patient survives beyond neonatal period care should be taken to keep patient's eyes open to prevent amblyopia. Artificial skin grafts are useful as patient's skin condition is prohibitive. Prevention of HI occurrence in future may be possible by genetic counselling, parental education and pre-implantation diagnosis.

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