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Case Report

A Rare Case of Congenital Ichthyosis, Harlequin Type

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ABSTRACT

Harlequin fetus is a rare and most severe form of congenital ichthyosis. The condition is associated with a fatal outcome. The disease affects the entire skin as well as the eyes, ears, mouth and the limbs which may be abnormally contracted. It is world's most unconquered medical challenge. Here we report one such rare case of Harlequin fetus, born to consanguineous parents with a history of previous sibling death due to the same disorder.

Keywords: Harlequin fetus, congenital ichthyosis

INTRODUCTION

Harlequin fetus is a rare keratinizing disorder. [1] The incidence is about 1 in 300,000 births. [2] The condition was first reported by Oliver Hart in 1750. [3] It is associated with a grave prognosis, as affected neonates usually die within first few days to weeks of life. The term Harlequin is derived from the newborn's facial expression and the triangular diamond shaped pattern of hyperkeratosis, which resembles the uniform of the comic actor Harlequin who is a masked clown dressed in multicoloured diamond-patterned tights. The usual pattern of inheritance is autosomal recessive, although autosomal dominant pattern may also be possibly responsible.

CASE REPORT

A female neonate weighing 1.4kg was born at 34 weeks of gestation by labour naturalis at Chengalpattu Medical College Hospital. The cause for preterm birth was oligohydramnios and preterm labour. Baby was the fourth child of a third degree consanguineous marriage. The present was an unplanned pregnancy with no proper antenatal care. The couple had lost one previous offspring in the neonatal period due to the same disorder. The other two children are normal.

On physical examination baby had a grotesque appearance. The entire body was covered with a thick parchment like membrane. There were extensive areas of hyperkeratotic scales separated by deep erythematous fissures. The facial features were distorted. The infant had bilateral severe ectropion, as there was complete eversion of eyelids with occlusion of the eyes. The lips were everted - eclabium, with the mouth being persistently open, hence the

infant was unable to suck effectively. The ears were malformed and compressed. There was a depressed nasal bridge and the hair was scanty. The limbs were edematous and there was restriction of joint mobility due to contractures. The fingers and toes were ischemic. The external genitalia were normal. (Fig 1,2,3)

Baby was nursed in a thermoregulatory environment. Umbilical catheterisation was done for venous access. Intravenous fluids and appropriate antibiotics were administered. Liquid paraffin was applied over the skin. The basic investigations were within normal limits. Ectropion was covered with eye pads and appropriate antibiotics and ophthalmic lubricants were used. Despite intensive care baby succumbed to cardiorespiratory failure at 50 hours of life.

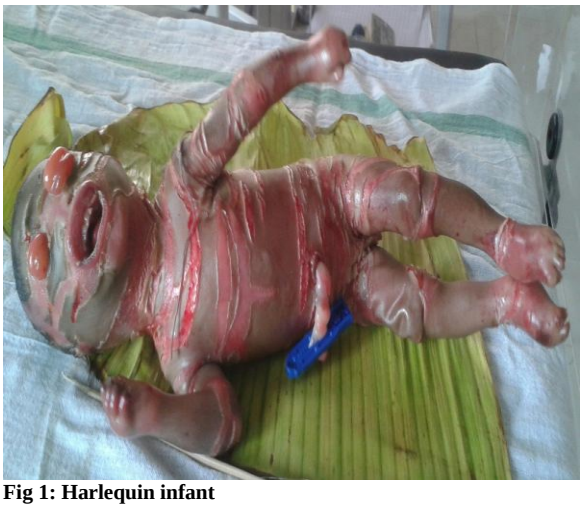

Fig 1: Harlequin infant
Large thick hyperkeratotic scales separated by deep erythematous fissures.

Fig 2: B/L ectropion & eclabium

Fig 3: Hypoplastic & ischemic digits

DISCUSSION

Harlequin fetus is a rare and lethal form of congenital ichthyosis. There is no racial or gender predilection identified. [4] Consanguinity and family history of Harlequin or other skin disorders are hallmark of the diagnosis. This also suggests an autosomal recessive mode of inheritance. [5] Premature birth is typical in Harlequin infants which add on to its complications. The condition is characterised by profound thickening of the keratin layer of skin, dense "armour"-like scale that covers the entire body. The thick cracked skin disfigures the facial features and causes constriction of digits. Facial anomalies include eclabium (eversion of the lips), ectropion (complete eversion of the eyelids with occlusion of the eyes), nasal hypoplasia and absence of external ears. [6] There are contractures of

the extremities at the joints and the fingers and toes appear hypoplastic and ischemic.

Histopathological analysis of the skin has identified abnormal or absence of lamellar bodies and absent intercellular lamellae. [7] The gene responsible for harlequin ichthyosis was traced as ABCA12 gene. This gene encodes a protein involved in the transport of epidermal lipids across cell membranes. [8] As there is a breakdown of the protective epithelial layer of skin, infants are prone for dehydration, electrolyte disturbances, sepsis, temperature dysregulation etc. Nutrition is impaired as constriction and swelling of the mouth interferes with the suck response. The thick shell of hyperkeratotic layer compromises respiration.

Treatment involves close monitoring of fluids and electrolyte status, intensive care of the skin, eyes and surveillance against infection. Research in this field suggests the administration of functional peptides with ABCA12-like properties in the management of Harlequin infants. Retinoids are often used and have a promising role when combined with good neonatal care. A survival rate of upto 8 years has been reported in children treated with Isotretinoin, acitretin and etretinate. [8]

Genetic counselling is of major importance, especially for those with a previous affected child. There are certain established features suggestive of Harlequin ichthyosis in three-dimensional antenatal ultrasound. They are as follows:

Constant sonographic findings	Indirect signs	Non specific ultrasonographic signs
Persistent large gaping mouth	"Snow flake sign"	Polyhydramnios
Dysplastic or swollen hands and feet	Intra- amniotic debris	Oligohydramnios
Aplasia of the nose	Floating membranes	
Bulging eyes		

These ultrasound markers although useful are not widely adopted. Definite prenatal diagnosis is possible by fetal skin biopsy taken at 17-20 weeks gestation.

The differential diagnosis include X linked recessive ichthyosis, Sjogren-Larsson syndrome, Dorfman - Chanarin syndrome, Neu-Laxova syndrome .

X linked recessive ichthyosis is caused by Steroid sulfatase defect. It is characterised by ichthyosis and scales all over the body. The flexural surfaces are spared. It is associated with extracutaneous manifestations like corneal opacity and cryptorchidism.

Sjogren-Larsson syndrome is characterised by a triad of ichthyosis, spastic paraplegia, mental retardation. The skin changes here involve hyperkeratosis, and they are of the ichthyosiform erythroderma type with red scaly skin.

Dorfman - Chanarin syndrome is characterised by ichthyosis along with multisystem involvement like hepatitis, cataract, sensorineural hearing loss.

Neu-Laxova syndrome is an autosomal recessive disorder with severe IUGR, ichthyosis, marked edema with skin restriction, limb deformities extreme microcephaly, cranio facial anomalies, and CNS malformations.

Our case had the classical presentation of Harlequin ichthyosis with extensive hyperkeratotic scales, deep erythematous fissures, contractures, distorted facial features and there was no systemic involvement.

CONCLUSION

Harlequin fetus is a rare type of congenital ichthyosis with high mortality. Despite supportive care Harlequin ichthyosis is associated with high mortality as affected

neonates succumb to sepsis, respiratory failure, electrolyte imbalance and poor nutrition. Although survival rates are prolonged with advanced treatment modalities, the severity of the persisting dermatosis leaves the individual with a lifetime of suffering. Prenatal DNA test would soon replace the fetal biopsy for better diagnosis. This case is reported for its rarity and for the classical clinical presentation of Harlequin fetus.

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