Harlequin Baby (A Case Report)

Mohammad Jafar Golalipour, Sakine Mohamadian and Elham Mobasher

In this article, a full-term male neonate with Harlequin fetus has been reported. It was born from a 20 year old mother by the Caesarean operation. In the clinical examination, broad, thick and hard hyperkeratotic disks were observed all troughs the body with extended gashes among them in the skin. The deformity of the face, the eyes server ectropion, the orbital chenosis and the flattening of the nose and the ears were among other disorders to be observed. The lips were overturned. There were no nails or hairs to be seen. The movements of the joint were limited. Hands and legs, in particular in the extremities were on a state of fixed gangrene and seemed to be ischemic. The patient died in the third day of hospitalization due to indicators of respiratory infections and the sepsis.

Key words: Ichthyosis, skin, hyperkeratosis, harlequin fetus

Mohammad Jafar Golalipour
Gorgan Congenital Malformations Research Center,
Gorgan University of Medical Sciences,
P.O. Box 49175-553
Gorgan, Iran
Tel./fax: +981714425165, 4421660

1Department of Embryology and Histology,
Gorgan Congenital Malformations Research Center,
Gorgan University of Medical Sciences, Gorgan, Iran
2Department of Pediatrics, Gorgan University of Medical Sciences,
3Department of Obstetric and Gynecology,
Gorgan Congenital Malformation Research Center,
Gorgan University of Medical Sciences, Gorgan Iran
INTRODUCTION

Harlequin type ichthyosis is a rare genetic skin disorder characterized by massive thick skin plates that usually produce distorted field features and often deformities in other parts of the body (Vijayaragavan et al., 1990; Akiyama et al., 1996).

It was first described by Hart in 1730 on the basis of familial occurrences, the mode of inheritance is autosomal recessive (Vijayaragavan et al., 1990; Watson and Mabec, 1995). It is usually fatal in the first few days of life but the development of one such fetus to age of 30 months is reported (Watson and Mabec, 1995; Prasad et al., 1994; Singalavannja et al., 1998).

We report a case of harlequin ichthyosis, which we believe is the first case in our province.

Case report: A full term male baby was delivered with 1 and 5 min apgar score 6 and 7 via cesarean section at Dezuri hospital in Gorgan, North of Iran. This baby was the first child of consanguineous marriage of healthy parents. There was no family history of ichthyosis. The pregnancy of the 20 years old mother was safe till she went into labor. She didn’t take any medication during pregnancy period.

Examination: A full term male baby without respiratory distress was born via cesarean section. The causes of cesarean surgery were cephalo pelvic disproportion and fetal face presentation. He was born with 37 cm head circumference, 52 cm height, 3800 g weight. In physical exams, he had normal heart, lungs and abdomen. The whole body was covered by hard hyperkeratotic skin disk, cracked skin similar homy plates over the entire body, disfiguring his facial features (Fig. 1).

Beyond the skin manifestation the baby had a severe ectropion and chemosis obscure the orbits. The nose and ears were flattened lend and the lips were everted and gaping. His nails and hair were absent, joint mobility was restricted and the hands and feet appeared fixed and ischemic (Fig. 1).

We diagnosed harlequin fetus for this patient based on consanguineous marriage of parents, characteristic of skin appearance, eyes, ears and extremities abnormalities. The patient was transferred into humidified heated incubator. Initial therapy began with a high fluid intake to avoid dehydration, antiseptic solution and antibiotic therapy.

At 3 day of his life, some clinical symptoms and signs such as grunting, respiratory distress and sepsis appeared. The patient died in third day of his life, his parents did not allow us for pathologic biopsy, autopsy and further investigation.

Fig. 1: The child with Harlequin ichthyosis after birth

DISCUSSION

Harlequin fetus is a rare manifestation of severe congenital ichthyosis. The clinical features of harlequin fetus are characteristic. Some infants have an abnormality of keratinization and others have a disorder of epidermal lipid metabolism (Vijayaragavan et al., 1990; Akiyama et al., 1996; Watson and Mabec, 1995).

The skin is dry and hard, cracked in places, markedly thickened, rigid and homy plate of skin over the body, disfiguring the facial appearance and constricting the digits. Hands and feet are usually crumpled and digits may be necrosed. There are ectropion, flattening of the ears and nose and fixation of lips. The membrane crinkles with initial respiratory efforts and shortly after birth begin to desquamate large sheets (Vijayaragavan et al., 1990; Akiyama et al., 1996; Watson, Mabec L.M, 1995 and Bianca et al., 2003).

The main problems faced by these babies are mechanical obstruction of respiration and feeding (Vijayaragavan et al., 1990; Akiyama et al., 1996).

Diagnosis is essentially clinical, supplemented by histological findings (Soares and Waghantikar, 1989; Baden et al., 1982).
Prenatal diagnosis has been accomplished by fetal biopsy and sonographic finding at 16-21 weeks of gestational age (Watson, Mabee LM, 1995).

In 1994 some studies on amniocentesis between 16-21 week of gestational age identified specific histological changes such keratinized and with increased abnormal growth and fat drop in affected fetus (Watson WJ, Mabee LM Jr, 1995; Elias S et al., 1980).

Treatment includes adequate humidification use of emollients, adequate hydration and maintenance of temperature, prevention of infection. Clinical improvement can be induced by oral retinoid but the prognosis remain grave (Prasad et al., 1994; Baden et al., 1982).

Neonatal morbidity and mortality may be due to cutaneous infection aspiration, (pneumonia squamous material) or hypernatremic dehydration from excessive transcutaneous fluid losses due to increased skin permeability (Vijayaragavan et al., 1990; Lawlor and Peiris, 1985; Nelson, 2004).

Prognosis is poor and survival of affected infants beyond the first year of life is uncommon (Singalavanija S et al., 1998; Nelson, 2004).

REFERENCES


