



# Journal of Medical Sciences

ISSN 1682-4474

**science**  
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*JMS (ISSN 1682-4474) is an International, peer-reviewed scientific journal that publishes original article in experimental & clinical medicine and related disciplines such as molecular biology, biochemistry, genetics, biophysics, bio-and medical technology. JMS is issued eight times per year on paper and in electronic format.*

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J. Med. Sci., 7 (5): 926-928  
1st July, 2007

## Harlequin Baby (A Case Report)

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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 chemosis 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

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## INTRODUCTION

Harlequin type ichthyosis is a rare genetic skin disorder characterized by massive thick skin plates that usually produce distorted facial 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 1750 on the basis of familial occurrences, the mode of inheritance is autosomal recessive (Vijayaragavan *et al.*, 1990; Watson and Mabee, 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 Mabee, 1995; Prasad *et al.*, 1994; Singalavanija *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 Dezyani 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 horny 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 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 to 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 Mabee, 1995).

The skin is dry and hard, cracked in places, markedly thickened, rigid and horny 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 cracks with initial respiratory efforts and shortly after birth begin to desquamate large sheets (Vijayaragavan *et al.*, 1990; Akiyama *et al.*, 1996; Watson, Mabee LM, 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 Waghlikar, 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).

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