

A Case Of Harlequin Fetus With Psoriasis In His Family

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Abstract

Harlequin fetus is the most severe form of congenital ichthyosis with an incidence of one in 300.000 births. An autosomal recessive pattern of inheritance is seen in this disorder. However; a new autosomal dominant mutation may possibly be responsible. The disorder has an ominous prognosis since the neonates usually die in the first hours or days of life. We reported here; a case of harlequin fetus with a history of psoriasis in his father.

INTRODUCTION

Harlequin fetus is the most severe form of congenital ichthyosis (1). The earliest record of its description is from the Oliver Hart in 1750 (2). Since then it has been referred to as “fetal ichthyosis”, “ichthyosis intrauterina”, “keratosis diffusa fetalis”, “congenital diffuse maligna keratoma”, “malignant keratosis”, “alligator baby” (3).

The disorder has an ominous prognosis, and affected babies usually die within the first days to weeks of life (1,4). The disorder is being presented here because of its extreme rarity and a history of psoriasis in his father.

CASE REPORT

An hour-old male infant was referred to Pamukkale University Hospital. He was born at 35-36 weeks gestation to a gravida I para 0, following a normal pregnancy. The parents were not relatives. The mother was 21 years old and the father was 23 years old. His father had psoriasis diagnosed a year ago.

Physical examination revealed an infant weighing 1700 g. The patient's temperature was 35(C, pulse rate 110/min, and respiratory rate 40/min. The clinical appearance of the baby was striking. The skin was hard, thickened, waxy and yellowish in colour. It was split irregularly to reveal erythematous moist fissures. The ears were underdeveloped and rudimentary. There was severe ectropion and eclabium. The baby's cry was normal, but he was unable to suck effectively. The nose was deformed and flattened. The nostrils were only being visible after the skin removed.

There was not chonal atresia. Hairs were underdeveloped (Fig. 1).

Figure 1

Figure 1: The patient showing ectropion, eclabium and thickened skin with deep fissures



There was a small penis, undescended testes, and a rudimentary scrotum. The limbs were in a semiflexed position and had limited mobility (Fig. 2).

Figure 2

Figure 2: The patient with small testes and rudimentary scrotum



He had 60(flexion contractures at elbows and knees and no

limitation in the movement at the wrist. Restricted abduction in the hip joint. The hands and feet were edematous with claw-like fingers and toes were clenched in a flexed position. The fingers and toes were hypoplastic and ischemic. The nails were absent (Fig. 3). The rest of the physical findings were unremarkable.

Figure 3

Figure 3: Edematous hands and clenched in a flexed position



Laboratory findings included hemoglobin 15.1 g/dl, white blood cell count 10700/mm³, platelet count 192000/mm³, creatinin 0.6 mg/dl, Na 128 mEq/L, K 3.1 mEq/L, Cl 101 mEq/L, AST 9 U/L, ALT 12 U/L.

Immediately after transfer to our neonatal intensive care unit, the baby was nursed in a humidified incubator maintained at 33(C. As peripheral venous access was difficult, an umbilical venous line was set up. An extra 25% allowance was provided for fluid and calorie requirements from the first day. After taking appropriate cultures, antibiotics were commenced in order to prevent infection. Vaseline containing five percent lactic acid and local antiseptics were applied topically. Ectropion was covered with eye pads soaked in saline.

Initially progress was slow. The plate like scales split and peeled off revealing glazed and erythematous skin underneath. There were necrotic areas on the tips of the fingers. He did not tolerate oral or N/G feeding and parenteral route for nutrition was used. His temperature was 38(C on the 16th day of admission. He was investigated for possible sites of sepsis. Twenty-one days after admission he had a cardiorespiratory arrest. A post-mortem examination could not be conducted. No micro-organisms grew in the cultures.

DISCUSSION

Harlequin fetus is a rare disorder with an incidence of 1 in 300.000 births (3). The first report is from the diary of Oliver

Hart, of Charleston, South Carolina who described these features in 1750 (2). These children are at great risk during the neonatal period and often die shortly after birth from the undernourishment caused by the rigidity of the lips, and underventilation and pneumonia because of respiratory distress. Abnormal water loss through the skin and poor temperature regulation lead to risk for infection beginning in the skin, but at the same time, because of poor temperature regulation, do not show the usual signs of infection. The taut skin may restrict normal respiration (4,6). However the longest survival period reported was up to 2.5 years in a child who was treated with etretinate (7).

This disorder is reported from different ethnic groups and occurs in both sexes. There have been reports of several families with siblings affected with harlequin ichthyosis (1,8). Twins affected by harlequin ichthyosis have also been reported (8). Occurrence of consanguinity in same parents and of harlequin ichthyosis in siblings suggest an autosomal recessive pattern of inheritance of this disorder (8). However, consanguinity has not been observed in a large number of cases which makes it difficult to explain as an autosomal recessive mode of inheritance (9). A new dominant mutation may possibly be responsible for the disorder (9,10). There were not any consanguinity and sibling in our case.

Ultrastructural studies of this condition indicate that harlequin ichthyosis is a genetically heterogeneous group of disorders, with altered lamellar granules and intercellular lipids, and variations in expression and/or processing of structural protein markers of normal epidermal keratinization (10,11). Dale and Kam (9) classified harlequin ichthyosis into three different types based on their epidermal protein analysis. They suggested a possible genetic defect altering the terminal differentiation based on their observation of the lack of desquamation and retention of cornified cells in the skin. Keratohyalin granules look fairly normal in type 1, are too small to be seen by light microscopy in type 2, and are absent in type 3. In all types there are no lamellar granules; instead, there are small vesicles that lack internal structure. There is also no evidence of the lipid lamellar that from between granular and cornified cells as a result of discharge of lamellar granular contents into the intercellular space (6).

Ultrasonography and the electron microscopic examination of the fetal skin biopsy permit the possibility of prenatal diagnosis of this disorder (1,3,9).

Oral vitamin A, topical antiseptics, liquid paraffin, five

percent lactic acid lotions have been used and found effective in the treatment of congenital ichthyosis (12). Erdem (12) has reported five collodion babies treated successfully with a topical vasoline preparation which contained five percent lactic acid. Etretinate is a synthetic retinoid derived from vitamin A. Its mode of action is unknown but retinoids have been shown to control differentiation and proliferation of keratinising and non-keratinising epithelia (13). We could not find etretinate instead we used topical vasoline preparation which contained five percent lactic acid.

The development of digital gangrene, as occurred in our patient in second day, has been reported previously (14).

In this case; interestingly his father had psoriasis. The lesions of psoriasis is characterized by erythematous papules that coalesce to form plaques with sharply demarcated, irregular borders. The pathogenesis is also unknown: epidermal turnover time, however, is distinctly accelerated compared with that normal epidermis. The mode of transmission is unknown; a multifactorial type of inheritance has been proposed. There is an association with histocompatibility antigens (HLA)-BW17, -B13, -B16 and -BW37 (15).

Two unrelated boys were described in the literature who were born as collodion babies and subsequently developed non-bullous ichthyosiform erythroderma and flares of generalized sterile pustulosis, similar to generalized pustular psoriasis (16).

There is no report for the relation of harlequin ichthyosis and psoriasis. However both of them are inherited keratinization disorders. We would like to point out that there could be a relation between harlequin fetus and psoriasis.

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