Harlequin Ichthyosis

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Abstract- It is an autosomal recessive, and occasionally autosomal dominant mutant extremely rare disorder with only 100 reported case in literature. This fatal disorder occur in both sexes and all races. In most circumstances the newborn die soon after birth Also it is known as harlequin fetus, alligator baby or keratosis diffusa fatalis.

Because of its rarity, we report 2 cases of this disorder, here.

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Key words: Harlequin Ichthyosis, neonate, ectropion

Case Reports

The first patient was a 2000 gram female neonate born prematurely on 32 weeks of gestation by normal vaginal delivery. Her APGAR score was 8-9. Her parents were cousins. Her mother have a history of 3 abortions in 12 to 16 weeks of gestation but 2 alive children who are well. On physical exam the baby was erythrodermic and her body was covered by thick yellow- brown plaques separated by red fissures which could easily bleed (Figure 1). Severe ectropion was observed. She had hypoplastic external ears and eclabium as well. Her hands and feet were covered by a membrane with hypoplasia of digits nails. She sufferd from respiratory distress and she was very ill. She admitted in neonatal intensive care unit and antibiotics and other conservative management started. She died 3 days after birth.

The second one was a 2300 gram weight term female neonate.

She was erythrodermic and her body was covered by thick, grooved and yellow-brown plaques separated by some areas of erythematous skin. Severe ectropion and eclabium were observed.

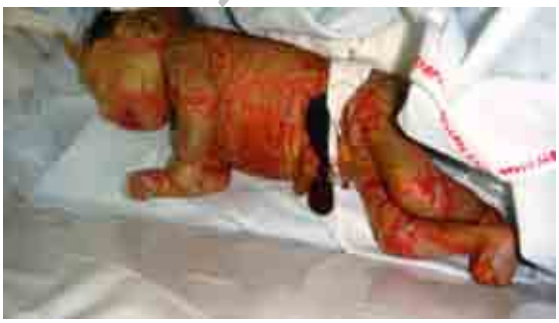

Figure 1.

Figure 2.

The hands and feet were covered with a membrane with restricted and hypoplastic toes (Figure 2).

The reflexes were normal and she had no feeding difficulty or respiratory distress. She managed conservatively.

Discussion

In 1750 Reverned Oliver Hart reported the first case who suffered from thickened and cracked skin over the whole body (4). It is a fatal disorder in which neonate die in few days after birth (5,6).

Very raely, after starting retinoids, some infants can survive a few months or years (7).

Harlequin ichthyosis presents at birth with severely thickened and grooved skin and scaling over the entire of body. It is associated with abnormal facial appearance including nasal hypoplasia, eclabimu, and lack of

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external ears. Other signs are ectropion, hypoplastic digits including absence of nails and restricted joint mobility (1,5-8).

These babies are at serious risks including hypothermia and hyperthermia, dehydration, respiratory difficulty and hypoventilation, poor feeding, hypernatremia, seizures and skin infection (1,9).

Biopsy of fetal skin can confirm the diagnosis at 20 to 23 weeks of gestation (8,10).

Postnatally, the diagnosis can be made clinically (1,9).

There is a report from India in which 3 siblings including 2 males and a female with this disorder suggesting dominant mutation cause a genetic defect in pathogenesis There was not any consanguinity between parents (11).

All of cases died within 2 days after birth.

In another report from Iran (kerman) a 1400 gram 33 week male newborn involved with this disorder. His parents were first cousin He died after a few days after birth (12).

Considering the consanguinity between parents in our case can lead us to an autosomal recessive inheritance. But pay attention to a history of 3 abortions in her mother can emerge a possibility of an autosomal dominant mutation route in inheritance.

Since It is a lethal disorder in which most of patients die early after birth, one measures is to diagnosis soon during pregnancy.

By Sonography, detecting the characteristic mouth shape at 17 weeks of gestation can conduct to diagnosis. Other clues that help to an early diagnosis are a positive history of this disorder in other sibling(s), consanguinity between parents and other keratotic cutaneous disorder in other children.

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