

Sudden Infant Death Syndrome with Harlequin Fetus

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SUMMARY

The harlequin fetus, a severe variant of ichthyosis, occurs rarely, and these babies die within the first few days of life. Early retinoid therapy may improve the disorder and help increase survival rates. The exact cause of the sudden infant death syndrome of the suckling is not known and the incidence approximately is 0.1-0.3 %. In general, these babies looked well and healthy at the time of the sleeping but were found dead in their bed in the morning. We report a harlequin fetus with sudden infant death syndrome.

Key Words: Harlequin Fetus, Acitretin, Sudden Infant Death Syndrome

Harlequin Fetus ile Ani Bebek Ölümü Sendromu Birlikteliği

ÖZET

Harlequin fetus ihtiyozisin şiddetli bir varyantı olup, bu hastalar genellikle yaşamın ilk günlerinde kaybedilirler. Erken retinoid tedavisi iyileşme ve yaşama oranını arttırabilir. Ani bebek ölümü sendromu genellikle sağlıklı görünen, sabahları yataklarında ölmüş olarak bulunan ve ölüm nedeni saptanamayan bebeklerdir. Yaklaşık %0.1-0.3 oranında görülür. Bu olgu raporunda harlequin fetus ile birlikte ani bebek ölümü sendromu olabileceği düşünülen bir olgu sunulmaktadır.

Anahtar Kelimeler: Harlequin Fetus, Acitretin, Ani Bebek Ölümü Sendromu

INTRODUCTION

Harlequin fetus is an extremely severe congenital form of ichthyosis, characterized by hyperkeratotic plates covering the entire body, ectropion, eclabium, poorly developed ears, and contractures of the hands and feet at the birth (1). Affected infants rarely survive after the first several weeks of the life (2). Reported cases were variable, associated with stillbirth or early neonatal death due to causes such as prematurity, respiratory compromise, infection, hypothermia, and dehydration (3). Early retinoid therapy may improve the disorder and help increase survival rates. We report a harlequin fetus with sudden infant death syndrome (SIDS) in the 6th month of the life.

CASE REPORT

An 18-hours-old newborn girl was transferred to our hospital for unusual appearance. She was born by vaginal delivery in the 38th week of gestation. The parents were first degree relatives and their previous born child was healthy. In the family history, the grand mother had given birth to three boys with harlequin fetus who had died within a few days after birth.

In physical examination, her weight was 3100 grs (50-75th percentile), length 52 cms (50-75th percentile), and head circumference 33 cms (50-75th percentile). She had firm, plate-like skin with fissures, contractures of all digits were present. (Fig. 1). The baby's

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activity and reflexes were normal. Marked ectropion, eclabium, pseudocontractures, and hypoplastic nails were other remarkable findings. The newborn was nursed in a highly humidified incubator and fed through a nasogastric tube in the neonatal intensive care unit. Body temperature and water electrolyte balance were closely monitored. Topical emollients were applied. Care of the eyes was consisted of the application of a topical ophthalmic antibiotic guttata.



Figure 1. Patient at 1 day of age had hyperkeratotic yellow scales, erythema, ectropion, eclabium, and contractures digits held

Electron microscopy of the skin section revealed hyperkeratosis, abnormal giant mitochondria, crystallizes of mitochondria, lipid drops and vacuolisation of cytoplasm (Fig. 2). Full blood counts, serum electrolytes, liver function tests, serum lipids were within normal limits. Tandem mass was normal. Radiologic survey was performed within the first week of birth and showed no abnormalities. Echocardiographic findings were within normal limits. After taking blood and skin cultures, antibiotics (cefazolin 100 mgs/kg/day and gentamicine 5 mgs/kg/day) were given order to prevent infection. Oral acitretin (0.75 mg/kg body weight/day) was given 2 days after birth. After 48 hours *S. hemolyticus* was isolated in the blood and skin cultures. Cephazolin and gentamycine were stopped, vancomycine 60 mg/kg day and amikacin 15 mg/kg day were given. The skin showed clinically apparent improvement after about 1 month of therapy (Fig. 3). The baby

was kept in the neonatal unit until 5 weeks of age and then was discharged.

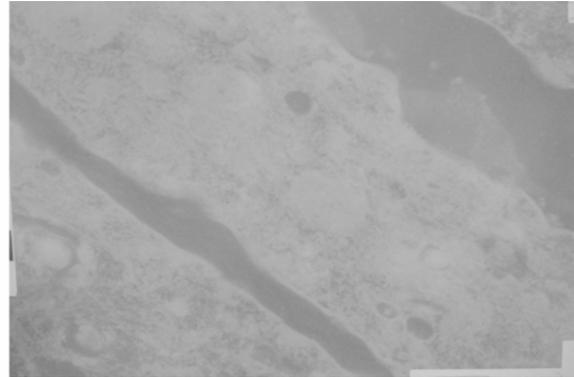


Figure 2. Electron micrograph ; skin section of epidermis ; (Uranyl acetate – lead citrate, x3000) Hyperkeratosis, abnormal giant mitochondria, crystallizes of mitochondria, lipid drops and vacuolization of cytoplasm.



Figure 3. Apperance the patient at 6 week of age

DISCUSSION

Harlequin fetus is the most severe form of congenital ichthyosis with an incidence of 1 out of 300,000 births (4). Harlequin ichthyosis is characterized by a profound thickening of the keratin layer in fetal skin. Most affected infants die within the first few days of the life (5, 6). Only a few children with harlequin ichthyosis have survived beyond the first year of the life so far (7). All of the previous cases received retinoids (etretinate or isotretinoin) for short periods or intermittently. The present child received acitretin (0.75 mg/kg /day), a good response was seen within 2-3 weeks, and she has been on continuous acitretin therapy for 6 months. Lacour et al⁸. recommended a

starting dose of acitretin of 0.5-0.75 mg/kg/day. Most of the long-term survivors of harlequin ichthyosis in the literature had been given oral retinoids early in life. In presented case acitretin therapy was started at the second day of the life. Eclabium, ectropion and skin fissures were improved at the end of the second week of the retinoid therapy. During 6 months follow up period, blood chemistry tests were performed monthly and they were found within normal limits.

Sudden infant death syndrome is defined as the sudden death of an infant under age 1 year remains unexplained after a thorough case investigation, including performance of a complete autopsy, examination of the death scene, and review of the clinical history (9). In general, these babies look well and healthy at the time of the sleeping but are found dead at their bed in the morning. In presented case child was put to sleep in supine position, when the mother tried to breast-feed the child again, she found him dead, after four hours. Skin findings of the patient improved with acitretin therapy. Also the exact cause of the dead is not known, SIDS may be the cause of the dead.

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