www.jcimcr.org

JCIMCR OPEN ACCESS Journal of Clinical Images and Medical Case Reports

ISSN 2766-7820

Short Report

Open Access, Volume 2

What's your diagnosis? Harlequin ichthyosis

Giridhar Guntreddi , MD FAAP; Jayasree Vasudevan Nair, MD FAAP; Swayam Prabha Nirujogi, MD MBA

[1]Pediatric Hospitalist, SANFORD Medical Center, 1300 Anne St NW, Bemidji, MN-56601, USA.

[2]Pediatrician, Amador Pediatrics, Sutter health, 100 Misson Blvd, Jackson, CA 95642, USA.

[3]Family Medicine, Tower health, Reading Hospital, Reading, PA 19611, USA.

***Corresponding Author: Giridhar Guntreddi, MD**

Pediatric Hospitalist, SANFORD Medical Center, 1300 Anne St NW, Bemidji, MN- 56601, USA.

Tel: 9735731527

Email: guntreddig@yahoo.com

Received: Feb 20, 2021

Accepted: Mar 10, 2021

Published: Mar 12, 2021

Archived: www.jcimcr.org

Abstract

Harlequin ichthyosis is associated with high morbidity and mortality in the past. However, fetal DNA analysis, 3 D facial antenatal ultrasound, and fetal skin biopsy significantly improved the diagnostic capabilities. The current advancement in treating these babies in level 3 neonatal intensive care units with a multidisciplinary team approach and early use of oral retinoids such as Acitretin, have made the outcomes better recently. Genetic counselling and prenatal screening are of vital importance after the birth of a baby with HI. In conclusion, it is necessary for the practicing clinicians to be aware of this diagnosis and the current management protocols if they come across a similar case.

Keywords: Harlequin ichthyosis, Amniocentesis, Retinoids.

Abbreviations: HI: Harlequin ichthyosis, U/S: Ultrasound.

Short report

A preterm male neonate born at 36 weeks of gestation to a primigravida mother, of South Indian descent with no prenatal care, no consanguinity was admitted to the NICU due to striking morphological features (Fig 1). The child appeared to be encased in armor-like plates of keratotic scales separated by deep red fissures. The face had the characteristic morphological appearance with severe eversion of the eyelids (ectropion), eversion of the lips (eclabium), flattened nose and deformed ears, generalized flexion contractures, deformed digits (Fig 2)(1). A clinical diagnosis on Harlequin ichthyosis was made, which is the most severe type of congenital ichthyosis with an incidence of about 1 in 300,000 births. The mode of inheritance is autosomal recessive with mutation in the lipid transporter adenosine triphosphate binding cassette A12 (ABCA 12) located on chromosome 2. Trans epidermal fluid loss, respiratory failure, electrolyte imbalance, and infections are the common complications. Despite of the optimal intensive care support, our patient died on Day 4 of life.

Harlequin ichthyosis is typically a clinical diagnosis in affected newborns. Fetal diagnostic modalities include fetal DNA analysis, ultrasound mostly 3 D, fetal skin biopsy. Fetal DNA analysis can be offered to parents who had a previous childbirth with HI. Fetal DNA can be obtained from chorionic villous sampling or amniocentesis (2). Prenatal U/S may show signs highly suggestive of eclabium, ectropion, rudimentary ears, contractures, and dense floating particles in amniotic fluid (snowflake sign) (3). Management of HI babies are always complex and must be in level 3 NICU with multidisciplinary team approach including neonatology, dermatology,

genetics, ophthalmology, ENT, plastic surgery, orthopedics, skilled nursing and Occupational and physical therapy. High humidity (50 to 70%) incubator care (4), nutritional support through umbilical/ PICC lines, liberal use of bland emollients applied at frequent intervals, and application of lubricant ophthalmic ointments (Lacrilube) (5) are mainstays of management of affected newborns. Early use of oral retinoids became the standard care of the management of HI. Retinoid treatment should be initiated with in first 7 days of life is for all infants who can tolerate the medication. The current literature suggests that the oral retinoid Acitretin dose be between 0.5 to 1 mg/kg/day (6). Retinoids are typically able to be discontinued by 6 months of age. Harlequin ichthyosis is associated with high morbidity and mortality in the past. The current advancement in treating these babies in level 3 neonatal intensive care units with a multidisciplinary team approach and early use of oral retinoids such as Acitretin, have made the outcomes better recently. Genetic counselling and prenatal screening are of vital importance after the birth of a baby with HI. In conclusion, it is necessary for the practicing clinicians to be aware of this diagnosis and the current management protocols if they come across a similar case.

Figure 1: Preterm baby with diffuse hyperkeratotic plates, deep red fissures, limited joint mobility, clenched fists and deformed fingers.

Figure 2: Eversion of eyelids (ectropion), everted lips (eclabium), flattened nose deformed ears.

References

1. Hashem Zadeh A, Heydarian F. Harlequin ichthyosis. Acta medica Iranica. 2009; 47(1): 81-82.
2. Yanagi T, Akiyama M, Sakai K, et al. DNA based prenatal exclusion of harlequin ichthyosis. J Am Acad Dermatol. 2008; 58(4): 653-656.
3. Raj popat S, Moss C, Mellerio J, et al. Harlequin ichthyosis; A review of clinical and molecular findings in 45 cases. Arch Dermatol. 2011; 147(6): 681-686.
4. Prado R, Ellis LZ, Gamble R, Funk T, Arbuckle HA, Bruckner AL. Collodian baby; An update with a focus on practical management. J Am Acad Dermatol. 2012; 67(6): 1362-1374.
5. Edwards WH, Corner JM, Soll RF, Vermont oxford Network neonatal skincare study group. The effect of prophylactic ointment therapy on nosocomial sepsis rates and skin integrity in infants with birthweights of 501 to 1000g. Pediatrics. 2004; 113(5): 1195 -1203.
6. Singh B, Bhura M, Maheswari A, Kumar A, Singh CP, Pandey SS. Successful treatment of Harlequin ichthyosis with Acitretin. Int J Dermatol. 2001; 40(7): 472-473.