Case Report / Olgu Sunumu

DOI: 10.4274/cayd.78941
J Pediatr Emerg Intensive Care Med 2018;5:134-139

Newborn with Harlequin Ichthyosis and the Nursing Care

Harlequin İktiyozisli Yenidoğan ve Hemşirelik Bakımı

Serap Torun[1], Handan Demiroğlu[2]

[1]Çukurova University Faculty of Health Sciences, Department of Nursing, Adana, Turkey
[2]Başkent University, Adana Dr. Turgut Noyan Research and Practice Hospital, Division of Nursing, Adana, Turkey

Abstract

Harlequin ichthyosis is the severest form of non-bullous ichthyosis, which is quite uncommon in newborns, and is usually characterized by fatal extreme keratinization of the skin. Because of the facial appearance of newborn, which resembles clown, the disease is called as harlequin -one of the comic characters of Italian street theatre. The present case report concerning a newborn with harlequin ichthyosis, which is an uncommon condition requiring intensive care, aimed to guide the care process by discussing the experiences over the nursing care period. Within the scope of nursing process, the data were collected, the diagnoses were identified, interventions were planned and implemented, and the outcomes of these implementations were evaluated. The nursing diagnoses were made based on the diagnosis list established by the North American Nursing Diagnosis Association. Nursing process was performed within the scope of the activities of daily living. Life expectancy of the patient, who was expected to die within a few days after birth, was extended by efficient and preventive nursing care, but these implementations have not been sufficient to prolong survival. Specific to this case, we can say that death could be postponed by nursing care. Palliative and preventive care is recommended when harlequin ichthyosis is encountered.
Keywords: Care, Harlequin, nursing period, case, medical ethics, newborn

Öz

"Harlequin" iktiyozis yenidoğanlarda, çok nadir görülen nonbülloz iktiyozisin en ciddi formu olup genellikle ölümcül seyreden cildin aşırı keratinleşmesi ile karakterize olan bir hastalıktır. Harlequinli yenidoğanın yüz ifadesi İtalyan sokak tiyatrosunda yer alan komedi karakterinden olan palyaçoya benzediği için bu isim verilmiştir. Nadir görülen ve yoğun bakım gerektiren bir olgu olan Harlequin iktiyozisli yenidoğanı konu alan bu olgunun hemşirelik bakım süreci deneyimleri paylaşılarak bakım sürecine rehberlik etmesi amaçlanmıştır. Hemşirelik süreci kapsamında veriler toplanmış, tanılar belirlenmiş, girişimler planlanmış, uygulanmış ve uygulamaların sonuçları değerlendirilmiştir. Hemşirelik tanıların belirlenmesinde North American Nursing Diagnosis Association tanıları kullanılmıştır. Hemşirelik süreci günlük yaşam aktiviteleri kapsamında uygulanmıştır. Hastaya verilen koruyucu ve etkili hemşirelik bakımı ile doğumdan sonra birkaç gün içinde kaybedilmesi beklenen hastanın beklenen yaşam süresi uzatılmış, ancak yapılan uygulamalar hayatta kalışı sağlamakta yeterli olmamıştır. Etkin hemşirelik bakımı ile bu olgu özelinde ölümün ötelenebildiğini söylemek olanaklıdır. Harlequin ile karşılaşıldığında palyatif ve koruyucu içerikli bakım sunulması önerilmektedir.
Anahtar Kelimeler: Bakım, Harlequin, hemşirelik süreci, olgu, tıbbi etik, yenidoğan

Introduction

The skin is the largest organ of the body and protects the body from physical injury, water loss and harmful microorganisms by serving as a barrier against external environment.[1] The cornified layer (stratum corneum) of the skin, which is found in the epidermis, consists of two compartments: keratin-rich matrix and lipid-rich matrix.[2,3] While keratin-rich corneocytes perform the water holding by providing skin elasticity, the lipid-rich matrix establishes a barrier against transepidermal water loss.[2] Protein- and keratin-containing cells (corneocytes) in the stratum corneum provide elasticity of the skin and keep the skin moisten by preventing evaporation of water.[3] It has been reported that an abnormality in the lamellar granules (stratum corneum), which play an important role in desquamation, was the main problem in all types of ichthyosis.[2-4]

Address for Correspondence/Yazışma Adresi: Handan Demiroğlu MD, Başkent University, Adana Dr. Turgut Noyan Research and Practice Hospital, Division of Nursing, Adana, Turkey
E-mail: handankuscu2006@hotmail.com **ORCID ID:** orcid.org/000-0001-6875-0148
Received/Geliş Tarihi: 29.09.2017 **Accepted/Kabul Tarihi:** 27.10.2018

Ichthyosis occurs due to hyperproliferation of the stratum corneum as in epidermolytic hyperkeratosis or due to abnormal removal of the corneocytes from the skin because of as in the lamellar ichthyosis[2]. The types of ichthyosis are distinguished from each other by different clinical and genetic features.[2]

Harlequin is a "clown-like" comedy character in Italian street theater. Facial appearance of infants born with this anomaly resembles this character; this is why the disease is called as harlequin ichtiosis.[5] Appearance of the skin of newborns with harlequin ichthyosis is characterized by erythematous diamond-shaped fissures and deep, thick, hyperkeratotic, brilliant-white plaques.[6] The substantial proportion of the skin is broken into diamond-shaped pieces by deep fissures.[7] This abnormal structure of the skin results from extreme keratinization.[5] The skin that becomes rigid because of extreme keratinization (10-fold thicker than the normal) makes movement difficult, and thereby deep fissures occur on this non-elastic skin.[5-7] It may be fatal in many newborns if the fissures are infected.[8] Ectropion (extroversion of the eye-lids) is encountered in infants born with this disorder. This may lead to ocular bleeding or injury during labor. In the lips, eclabium (lip's turning outwards) is seen; the lips are retracted through the skin surface and looks like a clown's smile. The ears and nose are not developed or formed. The arms, feet and fingers are either smaller than the normal or so deformed that do not allow flexion. Polydactyly is not uncommon.[5-7] These babies develop respiratory failure and respiratory tract infections because of both prematurity and inadequate expansion of the chest, which results from non-elastic skin over the chest.[6]

Newborns with ichthyosis are quite susceptible to heat changes as their armored skin prevents normal heat loss; hence, hyperthermia appears to be the most common symptom. Moreover, dehydration as well is a common sign because of impaired water-holding property of the skin.[5-8] Such patients usually die in the first days of their lives due to severe fluid loss, impaired heat balance, and sepsis.[9]

The present case report concerning a newborn with harlequin ichthyosis, which is an uncommon case requiring intensive care, aimed to guide the care process by discussing the experiences over the course of nursing care period.

Within the scope of nursing process, the data were collected, diagnoses were identified, interventions were planned and implemented, and the outcomes of these implementations were evaluated. The nursing diagnoses were made based on the diagnosis list established by the North American Nursing Diagnosis Association (NANDA). Nursing process was performed within the scope of the activities of daily living.

Case

Nursing Data Collected from the Newborn with Harlequin Ichthyosis

The baby boy, who was born to a 32-year-old mother by cesarean section as the second live birth from her fifth pregnancy (34 weeks of gestation), was diagnosed at birth with harlequin ichthyosis. Of the mother, who had a history of consanguineous marriage (maternal cousin) and five pregnancies in total, the first baby was born with Down syndrome, the second and third pregnancies ended in spontaneous abortion, whereas the fourth pregnancy had been terminated because of harlequin ichthyosis on the 28th week of gestation. During her last pregnancy, the doctor who followed the mother gave information about the situation of current fetus, but the family insisted on giving birth. When the family faced with the appearance of the baby, they refused him and preferred not to establish a bond as they assumed that he will die. They gave consent for any implementation that would be necessary over the course of care process. Consent of the family was obtained also to present the care process as a case report.

Baby Y was born with the body weight of 2450 g, height of 42.5 cm and head circumference of 34 cm. His body temperature was 36 °C, heart rate was 117/min, respiratory rate was 64/min, and arterial blood pressure was 67/44 mmHg. On his physical examination, armor-like hyperkeratinized areas and fine deep fissures were observed over the whole body surface together with severe eclabium and ectropion. It was observed that fine fissures have been accompanied by bleeding, the hair was invisible because of massive keratin, the ears were underdeveloped (rudimentary), and eyelids and lips had mucous structure. There were deformities in the fingers and toes, which were not in their usual posture. Testicles and penis were also hyper keratinized and had similar appearance to the overall skin surface (Figure 1, 2).

Based on these findings, the newborn was diagnosed with "harlequin ichthyosis" and admitted to the neonatal intensive care unit for treatment and monitoring.

Implementations of Nursing Care Process for this Case

Nursing care process for Baby Y, who has been monitored in the intensive care unit, lasted 51 days. Within nursing care process consisting of palliative and preventive care, data were collected from the patient, nursing diagnoses were made, interventions were planned and implemented, and the outcomes were evaluated. Within the scope of this process, the diagnoses listed by the NANDA were used to make the nursing diagnoses.[10]

The major problem encountered in the first days in Baby Y was impaired tissue integrity. Given that such patients generally die in the first days of life due to sepsis, dehydration and impaired thermoregulation,[11-14] Baby Y was kept in a humidified incubator. Every day, he was bathed with warm water using pH 5.5 shower gel containing linoleic acid. His whole body was moistened every 3 hours applying a moisturizer. His position was changed every two hours to reduce the pressure on the skin. Adequate fluid replacement was performed to hydrate the patient. It was determined that the keratin layer and bleeding from the fissures on the newborn's skin remained stable during interventions performed for skin integrity, however, skin integrity has gradually impaired day by day because of natural course of the disease.

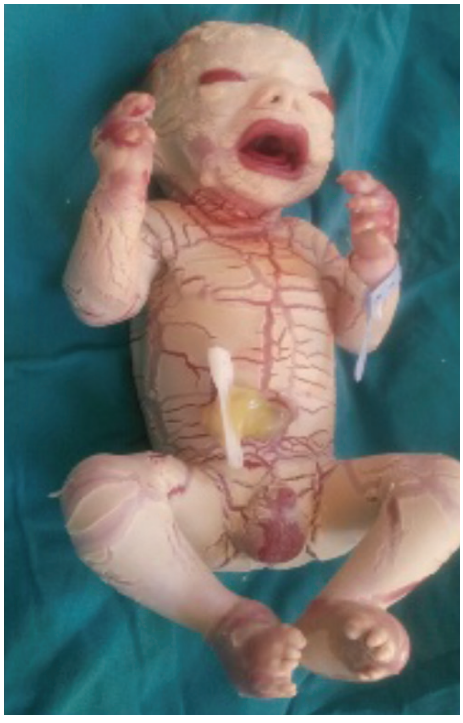

Figure 1. The first pictures after birth of the newborn with Harlequin ichthyosis

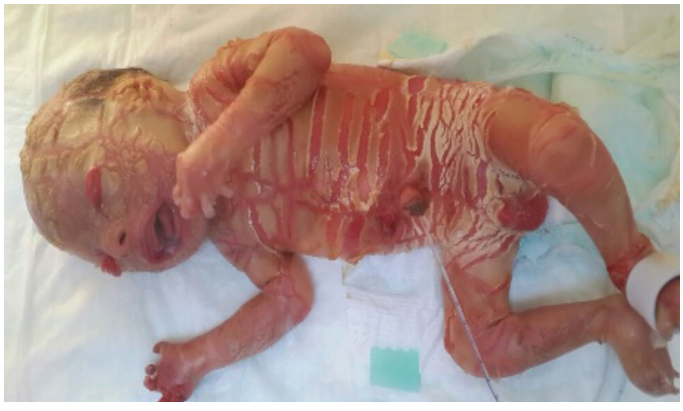

Figure 2. The picture approximately 10 days after birth of the newborn with Harlequin ichthyosis

Another nursing diagnosis was deficient fluid volume resulting from impaired tissue integrity. In order to maintain fluid and electrolyte balance in the body, intravenous fluid replacement was performed daily upon the physician's request; all intake and output and the body weight were recorded, and the infant was monitored for dehydration.

There was oral feeding problem as the infant could not suck the bottle because of eclabium (extraversion of the lips); hence, the diagnosis ineffective infant feeding pattern was made. Nursing interventions were performed to provide the infant with food necessary/adequate for his age and requirements. It was determined that body weight of the infant, which was 2450 g at birth, increased to 3770 g after one month. The level of albumin was 2.97 g/dL, Ca^{++} was 7.9 mg/dL and hemoglobin was 9.4 g/dL. Based on these data, considering protein loss due to plasma leakage out of the skin, the amount of dietary protein was increased.

Detecting impaired oral mucous membrane, which resulted from impaired oral feeding and xerostomia (dry mouth) due to eclabium, it was targeted to provide optimal oral hygiene for the baby. Within the scope of nursing care, harmful plaques were removed, oral cavity was controlled frequently, and oral care was performed regularly to prevent secondary infections. Despite the fact that the lips were frequently moistened at 2-hour intervals, the baby developed aphtha on the 25th day of care process, for which therapeutic interventions were performed.

Depending on the diagnosis of the risk of ineffective breathing pattern, which resulted from adequate expansion of the chest due to impaired skin elasticity, breathing was monitored frequently, SPO_2 was also monitored, and aspiration was performed when necessary in order to maintain effective breathing pattern. Oxygen treatment was initially performed using hood, and then it was performed via free-flow oxygen inside the incubator. Vital signs were closely monitored, skin color and warmth were checked, and pulse oximeter monitoring was performed. SPO_2 was determined to be 93% after the oxygen therapy given by hood at a dose of 2 Lt/min. Thereafter, whether free-flow oxygen therapy given inside the incubator provided sufficient breathing was monitored via oxygen saturation, which was within the normal limits indicating the presence of adequate oxygen in the system. Close monitoring continued for any problem that might occur.

Since the hyper keratinized skin of the newborn, who was at risk for imbalanced body temperature, hindered normal heat loss, it was aimed to keep the body temperature within the normal limits (36-37.2 °C). During nursing care process, the patient was hydrated based on the measurement of intake and output, he was monitored for the risk factors

for hyperthermia, and preventive measures were taken; the inner heat of the incubator was kept constant and vital signs were monitored and recorded every three hours. Body temperature of the newborn ranged between 36.9 °C and 37.0 °C. Appropriate body temperature could be maintained for approximately 4 days. The body temperature of the newborn was 37.8 °C (sub-febrile) on the 5th day of hospitalization, and then increased to 38.8 °C on day 14. Considering sepsis, broad-spectrum antibiotic therapy was started, however, growth was determined in the blood and wound cultures, which have been taken on the 11th and 24th days.

Adequate fluid replacement was targeted for the newborn who developed hyperthermia. Intake and output of the newborn were measured, inner heat of the incubator was regulated, and the newborn was bathed with warm distilled water.

Considering that the newborn had acute pain because of skin exfoliation and deep fissures due to impaired skin elasticity, it was targeted to minimize his pain. We thought the newborn's pain level assessed according to the pain scale would not give favorable results; hence, the baby was monitored closely and the alterations in his behavior, as well as the rhythm and intensity of crying, were used to assess pain. Analgesics ordered by the physician were administered and their effects were monitored. We tried to reduce the pain by moistening the skin.

In order to prevent risk for bleeding from deep fissures found on the skin, the skin was moistened every day at 3-hour intervals and bleeding profile was monitored. Catheter insertion sites were checked. The baby's hemoglobin and thrombocyte values were within the normal limits. However, blood leak was observed from catheter insertion site and the hemoglobin values decreased (8.60 h/dL) over the course of monitoring period.

Together with hyperthermia, the baby was diagnosed with infection, which, in the beginning, has been considered as a risk because of deep fissures and skin exfoliation. In the beginning of nursing care process, in order to prevent infection, aseptic principals were followed, the baby was monitored for localized and systemic signs and symptoms of infection, and he was admitted to an isolated room to minimize his risk of exposing to microorganisms. Umbilical catheter and then central catheter were inserted to use for treatment and to monitor the results of blood analyses. Thereafter, an orogastric tube was inserted for feeding.[15] Aseptic principals were followed for every intervention and care process. Despite the aseptic measures and isolation, the newborn developed hyperthermia on the 5th day of hospitalization. Considering sepsis, broad-spectrum antibiotic therapy was started. Positive growth was determined in the blood and wound cultures, which were taken on the 11th and 24th days. The treatment consisted of intravenous fluid replacement and antibiotic therapy.

Determining ineffective coping family due to family's not fulfilling the roles and responsibilities regarding the newborn with harlequin ichthyosis, it was targeted to support the family for the family members to be involved in the newborn's care process and for the parents to express their emotions about the situation. The father expressed desperate and aggressive behavior when he first saw the newborn and no communication could have been established. The mother wanted to see the newborn on 16.04.2015. The diagnosis of impaired parent/infant/child attachment was made due to the interruption of communication process, which is based on trust-love and attention, secondary to the newborn's disease. The parents were offered to involve in the newborn's care process for they can cuddle the newborn, smile, and establish a relationship. However, the father refused receiving any information and education, and mother did not want to see the baby because of health problems. As they have not visited thereafter, it is possible to say that the baby has been left by his parents. Unlike the case reported by Koocheck et al.[15], the family did not accept the baby in this case.

Despite the precise treatment-care process, hyperthermia was encountered more frequently in the next days. Unfortunately, the newborn died on the 51st day due to infection as was mentioned in the literature.[8]

Discussion

The present case includes significant ethical issues concerning the beginning and end of life. Harlequin ichthyosis is a fatal keratinization disorder passed with autosomal recessive inheritance, which occurs due to ABCA12 gene mutation -the very uncommon and severest form of non-bullous ichthyosis.[16-18] Although one of each 500 individuals carries a recessive gene for this disorder, the probability of marriage of two non-kin individuals having this gene is 1/250 000.[7] This is reported to be seen roughly one in a million. This case was presented considering the benefits of discussing the care process in the academic literature as it is a rarely encountered condition.

Prenatal diagnosis of genetically inherited fatal diseases is of great importance.[12] Today, genetic mutations leading to this disease can be detected by molecular methods, and it is possible to provide families with pre-conception counseling.[16] The parents of the present case had been informed about the necessity of termination of pregnancy,

however, they preferred giving birth. It is possible to say that the parents estranged themselves from the baby due to their conscientious responsibility and ambivalent emotions because they have not thought the problem would be so serious. The Baby Y's being left by his parents, emotional depression experienced by the parents, and trauma caused by frustrated dreams and expectations about the baby as the result of maintenance of pregnancy emphasize the necessity of supportive therapy for the family. When diagnosed during the antenatal period, the family should be informed in extreme detail, even using visual materials, what situations they may face with after birth. In the present case, the information given to the family has not been reinforced by visual materials. Although making decision to terminate the pregnancy is on the initiative of family when the diagnosis is made during antenatal period, Baby Y would have not been given birth and not lived for 51 days, which could be considered as suffering, if the situation has been explained using visual materials. Palliative and preventive care in harlequin cases is recommended within the context of respect to life in the event the baby is abandoned by the family after birth.

On the other hand, a criticism that the nursing care has been unnecessarily performed from the ethical aspect may be in question. Herein, we discuss an individual, who has got the chance of living by being given birth. Moreover, questions like "why the methods such as not treating or not feeding that are among the arguments about the end of life have not been implemented?" or "is it reasonable to postpone death, which is the expected end?" may arise. Each member of the nursing care team believes in the sanctity of life and the necessity of protecting life and has preferred the principle of 'do no harm'. Health care team, in their own way, has tried to keep the patient's quality of life at the intended level by means of palliative care. Optimum palliative care was provided for the patient and it was waited for spontaneous end of life as he was a newborn, unable to express himself, left by his parents, and had no authorized legal representative.

Patients with harlequin ichthyosis need to be followed in full-equipped intensive care units and by multidisciplinary approach.[19] The majority of patients dies on the first day of life, but can rarely survive with severe ichthyosis and neurological abnormalities until the further stages of infancy.[16-21] Life expectancy for Baby Y, who received efficient preventive and nursing care, was prolonged and the interventions allowed him to survive for 51 days.

This paper has depicted the nursing diagnoses specific to the present case, which have been frequently encountered over the course of nursing care process. Health care professionals are recommended to discuss their cases in the academic literature for the elimination of problems that may be experienced during caring process of such rarely encountered cases.

Ethics

Informed Consent: Consent of the family was obtained also to present the care process as a case report.

Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: S.T., H.D., Concept: S.T., H.D., Design: S.T., H.D., Data Collection or Processing: H.D., Analysis or Interpretation: S.T., H.D., Literature Search: S.T., H.D., Writing: S.T., H.D.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

References

1. Ivich,J M. Ichthyosis in the neonatal setting. Clinical Issues in Neonatal Care. 2015;15:253-60.
2. Karaduman A. Kalıtsal keratinizasyon bozuklukları. Türkderm-Deri Hastalıkları ve Frengi Arşivi Dergisi. 2011;45:73-80.
3. Takeichi T, Akiyama M. Inherited ichthyosis: Non-syndromic forms. J Dermatol. 2016;43:242-51.
4. Milner ME, O'Guin WM, Holbrook KA, Dale BA. Abnormal lamellar granules in Harlequin Ichthyosis. J Invest Dermatol. 1992;99:824-9.
5. Lucas P, Legendre L, Pauwels C, Mazereeuw-Hautier J. Harlequin phenomenon associated with neurological abnormalities: A case report. Ann Dermatol Venereol. 2016;143:369-71.
6. Swati R, Liji SD, Manisha MB, Mandeep SB, Gautham A. Harlequin Ichthyosis: Prenatal diagnosis of a rare yet severe genetic dermatosis. J Clin Diagn Res. 2015;9:QD04-6.
7. Butragueño Laiseca L, Vázquez López M, Polo A. Harlequin Syndrome In A Paediatric Patient: A Diagnostic Challenge. Neurologia. 2018;33:478-80.
8. Jilumudi UB. Harlequin ichthyosis: A medico legal case report & review of literature with peculiar findings in autopsy. J Forensic Leg Med 2012;19:352-4.
9. Akiyama M. The Pathogenesis of severe congenital ichthyosis of the neonate. J Dermatol Sci. 1999;21:96-104.
10. Capernito Moyer LJ. Handbook of Nursing Diagnosis (13th Edition). Lippincott Williams & Wilkins. 2010. (cev)Erdemir, F. Hemsirelik Tanıları El Kitabı. Nobel Kitabevi, İstanbul, 2012.
11. Devika P, Marla J, Tor S. Harlequın ichtiyosis. J Am Acad Dermatol. 2015;72:200.
12. Sharma D, Gupta B, Shastri S, Pandita A, Pawar S. Collodion baby with TGM1 gene mutation. Int Med Case Rep J. 2015;8:205-8.
13. Schmitt GJ, Furtat Marques T, Trevisol DJ, Schuelter-Trevisol F. Congenital Ichthyosis: A case report. Clin Pediatr (Phila). 2015;54:598-600.

14. Salehin S, Azizimoghadam A, Abdollahimohammad A, Babaeipour-Divshali M. Harlequin ichthyosis: Case report. J Res Med Sci. 2013;18:1004-5.

15. Koochek A, Choate KA, Milstone LM. Harlequin ichthyosis: neonatal management and identification of a new ABCA12 mutation. Pediatr Dermatol. 2014;31:e63-4.

16. Darmstadt GL, Sidbury R. Disorders of keratinization. In: Behrman RE, Kliegman RM, Jenson HB, (eds). Nelson Textbook of Pediatrics. (17th ed). Philadelphia: WB Saunders Company, 2004:2200-4.

17. Erdeve SS, Turkoglu O, Can O, Astarci HM, Tiras U. Harlequin iktiyozis olgu sunumu. Gülhane Tıp Dergisi. 2007;49:52-4.

18. Akiyama M, Sugiyama-Nakagiri Y, Sakai K, McMillan JR, Goto M, et al. Mutations in lipid transporter ABCA12 in harlequin ichthyosis and functional recovery by corrective gene transfer. J Clin Invest. 2005;115:1777-84.

19. Çapan K. Harlequin iktiyozis: Aynı ailede üçüncü olgu. Türk Pediatri Arşivi. 2014;49:269-71.

20. Richard G, Choate K, Milstone L, Bale S. Management of icthyosis and related conditions gene based diagnosis and emerging gene based therapy. Dermatol Ther. 2013;26:55-68.

21. Harvey HB, Shaw MG, Morrell DS. Perinatal management of harlequin ichthyosis: a case report and literature review. J Perinatol. 2010;30:66-72.