JOURNAL OF
CONTEMPORARY MEDICINE

DOI:10.16899/jcm.1478881
J Contemp Med 2024;14(3):158-159

Case Report / Olgu sunumu

Oral Retinoid Treatment in a Newborn with Lamellar Ichthyosis

Lamellar İktiyozisli Yenidoğanda Oral Retinoid Tedavisi

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Abstract

Ichthyosis is a heterogeneous group of conification disorders characterized by hyperkeratosis involving the skin. One form of ichthyosis, lamellar ichthyosis (LI), is autosomal recessively inherited with an incidence of 1/300,000. The diagnosis is based on clinical findings, skin biopsy and genetic analysis. Treatment includes moisturizing the skin, reducing fluid loss and keratolytics. Oral retinoids have been found to be very successful in the treatment of LI. In this case, a premature newborn diagnosed with LI is presented because of its rarity and good response to oral retinoid treatment.

Keywords: Ichthyosis, lamellar ichthyosis, oral retinoids

Öz

İktiyozis, deriyi tutan hiperkeratoz ile karakterize heterojen bir grup konifikasyon bozukluğudur. İktiyozisin bir formu olan lamellar iktiyozis (LI), 1/300.000 insidans ile otozomal resesif kalıtılır. Tanı klinik bulgulara, deri biyopsisine ve genetik analize dayanır. Tedavi, cildin nemlendirilmesini, sıvı kaybının azaltılmasını ve keratolitikleri içerir. Oral retinoidler LI tedavisinde çok başarılı bulunmuştur. Bu olguda, nadir görülmesi ve oral retinoid tedavisine iyi yanıt vermesi nedeniyle LI tanısı konulan bir prematüre yenidoğan sunulmuştur.

Anahtar Kelimeler: İktiyozis, lamellar iktiyozis, oral retinoidler

INTRODUCTION

Ichthyosis is a group of clinically and etiologically heterogeneous cornification disorders characterized by diffuse crusting of the skin. It may be congenital or acquired. Congenital forms include lamellar ichthyosis (LI), non bullous congenital ichthyosiform erythroderma, and "harlequin ichthyosis".[1] In this case report, we present a patient with Harlequin ichthyosis who had severe skin involvement at birth and was treated with oral acitretin.

CASE REPORT

Male baby was born via spontaneous vaginal delivery from the 3rd pregnancy of a 21-year-old mother. His birth weight was 2720 gr (%75-90 Percentile) at term (34 weeks). Postnatal follow-up was performed in the neonatal intensive care unit. Dermatologic examination revealed a membrane-covered appearance, fissures between the membranes and occasional bleeding in the form of leakage. There were pseudocontracture in the fingers and toes, fish mouth appearance and ectropion was due to loss of skin elasticity (**Figure 1**). There were marked contractures at the extremity ends, especially in the fingers. Other system examinations were normal. Routine blood tests, cranial and abdominal ultrasonographic evaluation were normal. Echocardiographic findings were normal. Skin care was applied with sterile vaseline and oral feeding was started on the 2nd day. In collaboration with pediatric specialists, it was decided to start oral acitretin treatment. Family consent was obtained and 1 mg/kg oral acitretin was started. Retinoid treatment was planned to be continued by decreasing the dose in a six-month period. Weekly biochemistry tests were continued in terms of treatment side effects. Calcipotriol and vaseline were started for topical skin care. Topical retinoic acid and 10% topical urea were applied for thick plaque lesions. Fusidic acid was applied for areas with suspected infection. Fever and elevated C-reactive protein (CRP) were not detected during follow-up. The membranes on the skin of the patient, which were present at birth, started to fall off on postnatal day 10 (**Figure 2**). Informed consent was obtained from the family for this case.

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Received *(Geliş Tarihi)*: 06.05.2024 **Accepted *(Kabul Tarihi)*:** 17.05.2024

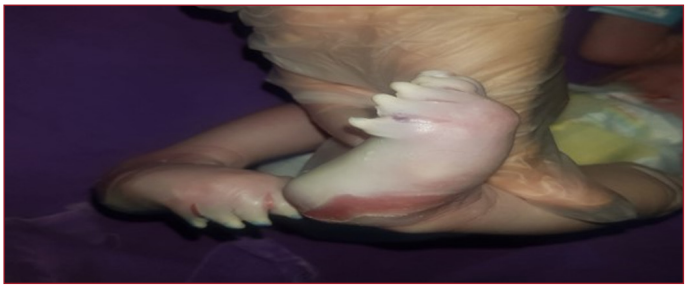

Figure 1. Membrane structure on the skin and rudimentary fingers after birth

Figure 2. Large, thick hyperkeratotic squames separated by erythematous fissures in a 34-week premature neonate with lamellar ichthyosis.

DISCUSSION

"Ichth" means fish in Latin. Ichthyosis refers to a group of cornification disorders characterized by skin lesions covered with fish scale-like squames. During neonatal period, ichthyosis may be observed in two different clinical forms including collodion baby and Harlequin ichthyosis.[2] In Harlequin ichthyosis, the skin is covered with plaques and deep cracks like hard armor. There is ectropion and eclabium and flattening in the ears and nose. Hair and nails are not yet developed. Adhesions and necrosis of the fingertips can be observed on the hands and feet. Limited joint movements may be observed due to the hard skin structure.[3] Failure to ensure skin integrity can disrupt the skin's barrier function and may result in infection, heat and fluid loss. Bacterial infections, heat loss and dehydration may develop because the skin cannot fulfill its functional barrier function and maintain its integrity. In the past years, two siblings of collodion babies who died as a result of hypernatremic dehydration have been reported.[4] Erythroderma is very little or absent in classic LI. There may be findings ranging from mild increased striation on the palms and soles of the feet to keratoderma. Lips and mucous membranes are not affected, but nearby skin areas may be involved. Ectrapion is almost always seen and is a diagnostic finding. Eclabion, hypohidrosis and alopecia may accompany other findings.[5] In our patient, parchment-like, lamellar peeling was present on whole body including palms and soles at birth. No oral mucosa involvement and erythroderma were observed and diagnosis of lamellar ichthyosis was made via clinical findings. During treatment, moisturization of the skin, prevention of fluid loss and keratolytic agents are used as a principle.[2] Lotions, creams, ointments and oils are used for skin moisturization and creams containing 5% lactic or glycolic acid, 10-40% urea and 0.1% retinoic acid are used for keratolytic treatment. Ophthalmologic care is also required for ectrapion in patients with ichthyosis.[2] The patient who was evaluated by dermatology and ophthalmology consultant physicians was started on eau distillate and vaseline for skin moisturization, artificial tears and antibiotic eye drops for the eyes. He was monitored with fluid and electrolyte therapy appropriate for her weight and age. Systemic retinoic acid (0.5-1 mg/kg/day) treatment has been reported to provide dramatic benefit in severe forms of ichthyosis including LI and congenital ichthyosiform erythroderma.[6]

CONCLUSION

Lamellar ichthyosis is a dramatic picture for the family and the physician, which may progress with fatal complications. It should be kept in mind that a successful neonatal intensive care process with a multidisciplinary approach and retinoic acid treatment may lead to long-term survival.

ETHICAL DECLARATIONS

Informed Consent: All patients signed the free and informed consent form.

Referee Evaluation Process: Externally peer-reviewed.

Conflict of Interest Statement: The authors have no conflicts of interest to declare.

Financial Disclosure: No financial support was received from any organisation in this study.

Author Contributions: All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

Note: This case was presented as a poster presentation at the 27th National Dermatology Congress (October 16 - 20, 2018, Antalya) (PS-086).

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