LAMELLAR ICHTHYOSIS: ONE CASE REPORT

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ABSTRACT Introduction: An ichthyosis is a heterogeneous group of skin disease characterized by generalized scaling. Lamellar ichthyosis is an autosomal recessive disorder with a mutation in the TGM 1 gene encodes the transglutaminase I protein was found. The scales reflect changes in epidermal differentiation. Lamellar ichthyosis is the one of the rarest form of ichthyosis where the infant is usually born with a collodion membrane.
Case: The ophthalmologic department consulted a 10-year-old girl with a chief complaint of generalized scales on the body since birth. Both patients also complained of sore eyes because they cannot close tightly. According to the parent, at the time of birth, the patient was enveloped by membranes where the eyes could not close tightly. The patient was also unable to breastfeed. Physical examination showed ichthyosiform scales arranged lamellar in a mosaic pattern throughout the body, Eclabium on the lips and ectropion on the eyes were found. Histopathological examination results support the diagnosis of lamellar ichthyosis. The patient was given emollient and supportive therapy. A good improvement shown by markedly diminished scales and skin stiffness was seen after six weeks of treatment.
Conclusion: Lamellar ichthyosis is a rare type of ichthyosis. Treatment aims to prevent excessive transepidermal water loss, prevent complications such as secondary infections and to reduce clinical symptoms.

KEYWORDS ectropion, emollient, lamellar ichthyosis

Introduction

Ichthyosis is a heterogeneous group of skin disorders characterized by generalized scales and dryness. Scales signify changes in epidermal differentiation. [1],[2] Clinical symptoms of Ichthyosis depend on aetiology. Ichthyosis is divided into three subgroups: Autosomal dominant ichthyosis, X-linked recessive ichthyosis and autosomal recessive ichthyosis [3].[4]

Lamellar Ichthyosis (LI) is an autosomal recessive Ichthyosis. LI is the rarest disease that appears at birth, where the newborn infant is usually encased in a collodion membrane. [1] The prevalence of LI is about 1 in 300,000 births. Ichthyosis is twice more common in men. Twenty-five percent of patients were born premature, and in fifty-one percent of cases, the siblings are also affected with LI. In some patients, the disease is limited to the trunk and extremities while in eighty percent of cases a systemic involvement is found.[4] A genetic defect in TGM 1 gene is the basis of LI, where this gene mutation is found in fifty percent of patients.2 TGM 1 gene encodes transglutaminase 1, an enzyme that is crucial in the formation of envelope cells in keratinocyte differentiation.[6]

Diagnosis is based on clinical symptoms, family history, and can be supported by morphological examination and other specialized tests. If available, molecular analysis is recommended to confirm the diagnosis. This test is also useful for family member testing and prenatal diagnosis. Routine histopathological findings in some Ichthyosis is not characteristic, often only showing only epidermal hyperplasia and varying degree of orthohyperkeratosis. In combination with typical clinical findings, the result of histopathological examination can provide important clues to Ichthyosis Vulgaris or Ichthyosis epidermolysis. [7]

Systemic and topical retinoids are beneficial in reducing the scales, unusually thick periocular squares, which can mitigate the risk of developing ectropion. Ichthyosis with ectropion can cause complications such as bulbar conjunctivitis and keratinization or keratopathy due to sun exposure.8 Conservative therapy

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in the early phase of life is highly recommended in LI with ectropion. [9],[10]

We report a case of LI in a 10-year-old female with a history of generalized scales since birth. The objective of this report is to describe the clinical features and complications of LI as well as the current treatment options.

Case

A 10-year old girl was consulted by to the dermatology department of Wahidin Sudirohusodo Hospital, Makassar, South Sulawesi, Indonesia, by the ophthalmology department with a chief complaint of generalized scales since birth. Pruritus was present which was exacerbated during perspiration and hot environment. The scales were persistent and never subsided. Pain on both eyes was also present as the patient was not able to completely close her eyes. The mother reported that the patient was born encased in a membrane. The eyes could not be closed and the patient could not breastfeed, prompting a nasogastric tube insertion and the hairs were also noted to fall out slowly. History for other medical conditions was unremarkable. No familial history was present. Consanguinity in the parents was absent. None of the siblings experienced the same condition.

The patient was born with a caesarian section with buttocks presentation and was a term. The mother consumed no medications except for vitamins obtained from the physician. During her life, the patient had only got topical oxytetracycline and hydrocortisone ointment with no significant improvement.

Physical examination revealed good general condition with mild malnutrition. Vital signs were all in normal limit. The heart, lungs, and abdomen were all within normal limit. Dermatological examination showed generalized ichthyosiform and lamellar scales with a mosaic pattern, erosions and fissures. On the scalp, thick scales and alopecia were present. Ectropion was observed in the superior, and inferior palpebrae with scales and eclabium were observed in the perioral area. Contractures were present on both extremities with swan neck deformity on the fingers of both limbs.

Based on the history taking and physical examination, the patient was diagnosed with lamellar ichthyosis with a differential diagnosis of congenital ichthyosiform erythroderma (CIE). The patient was given emollient containing capric triglyceride, glycerine, pentylene glycol, sodium carbomer, squalene, Cocos nucifera, cetirizine, vitamin B complex, fusidic acid cream for erosions on the face. The patient was also given 2% salicylic acid and 10-gram miconazole cream to be applied on the scalp.

Histopathological examination showed epidermal hyperplasia with hyperkeratosis, parakeratosis, hypergranulosis with focal hypogranulosis, and follicular plugging. The upper dermis showed perivascular lymphocytic inflammatory cells. These results were consistent with lamellar ichthyosis. A follow-up visit in the fifth month showed a marked reduction of scales and improvement in the skin stiffness.

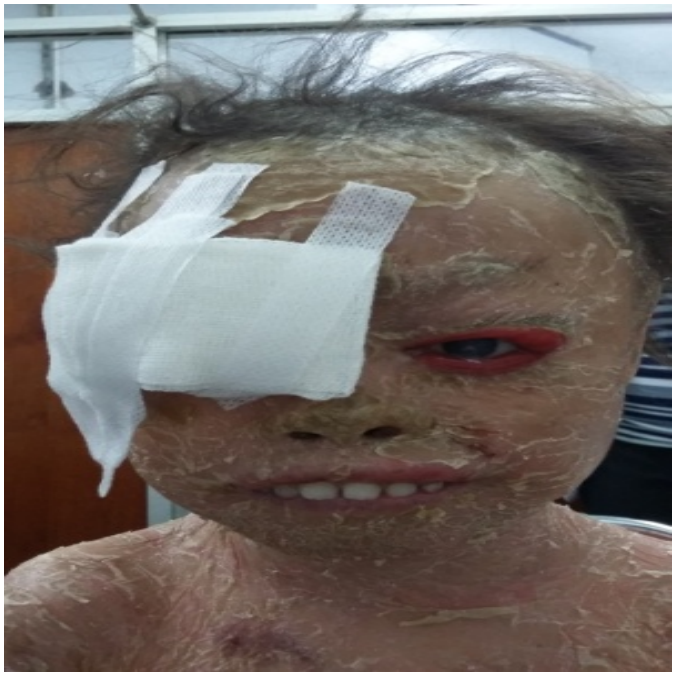


Fig.1.a.

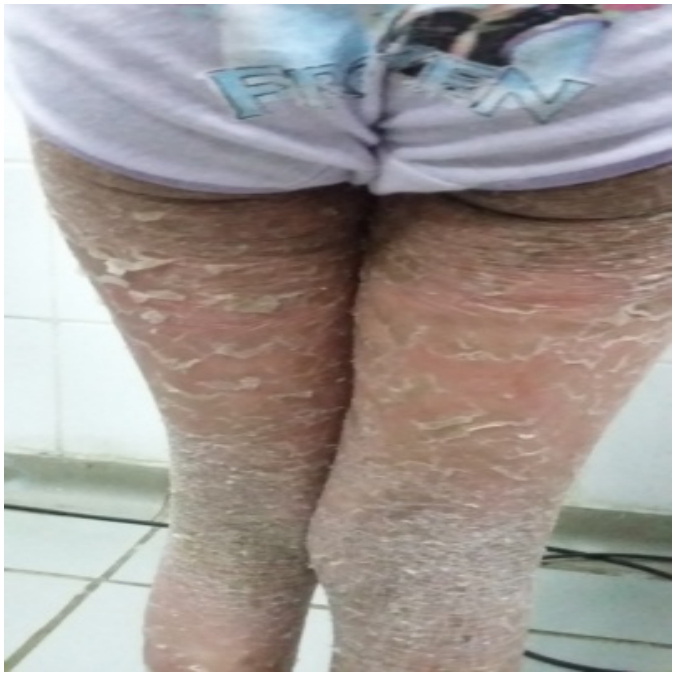


Fig.1.b.

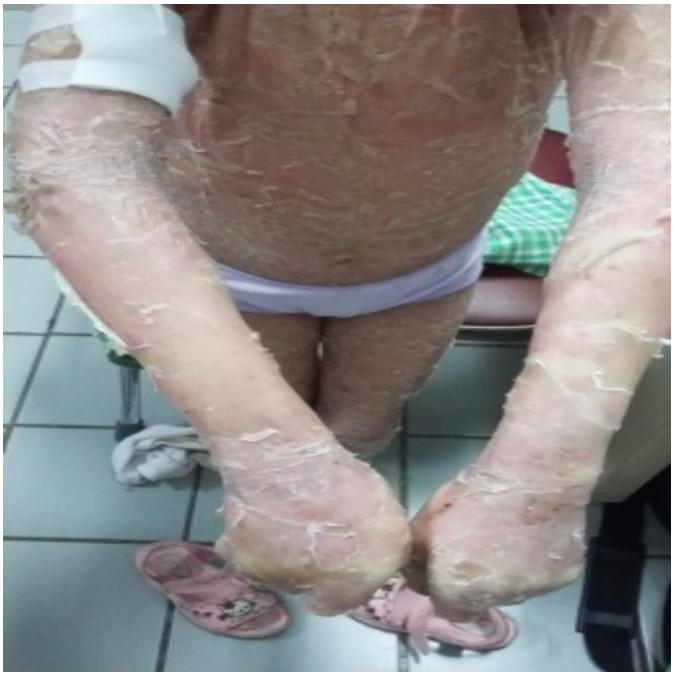
Appendix

Pre-treatment

Histopathological Examination

Post-treatment

Improvement of the scales and skin stiffness after treatment



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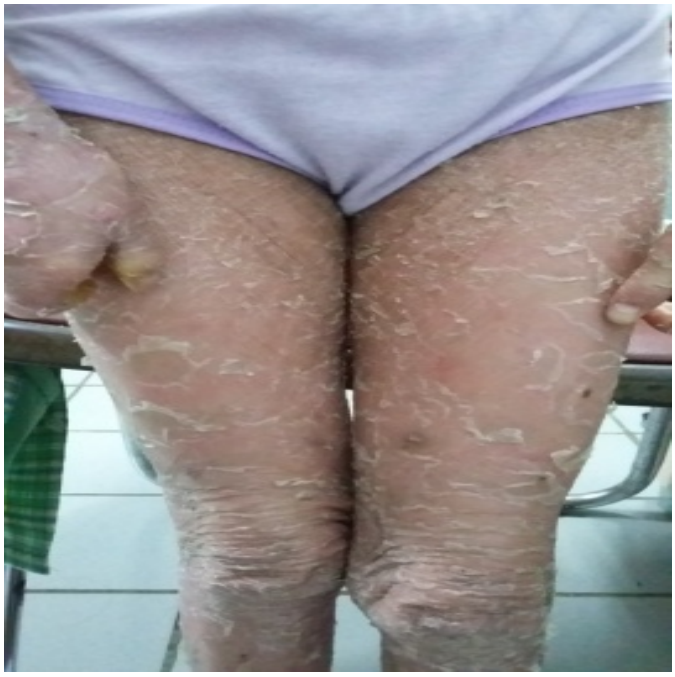

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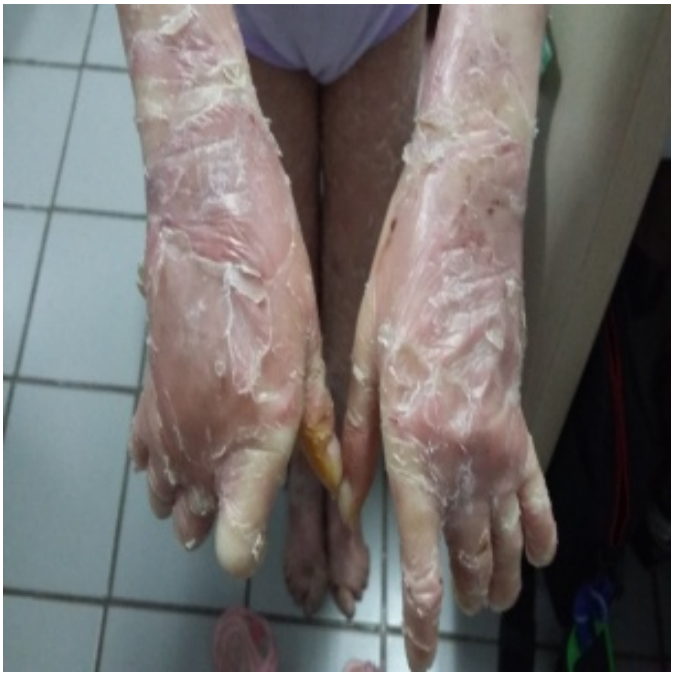

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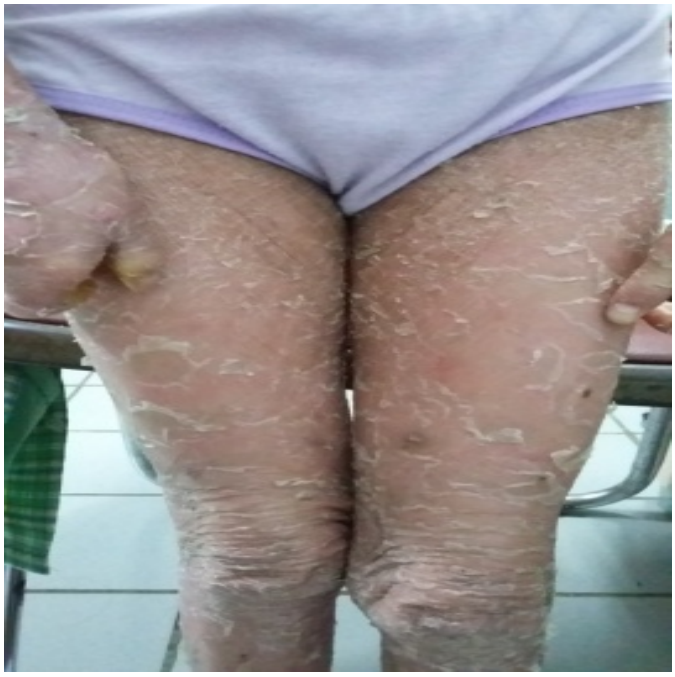

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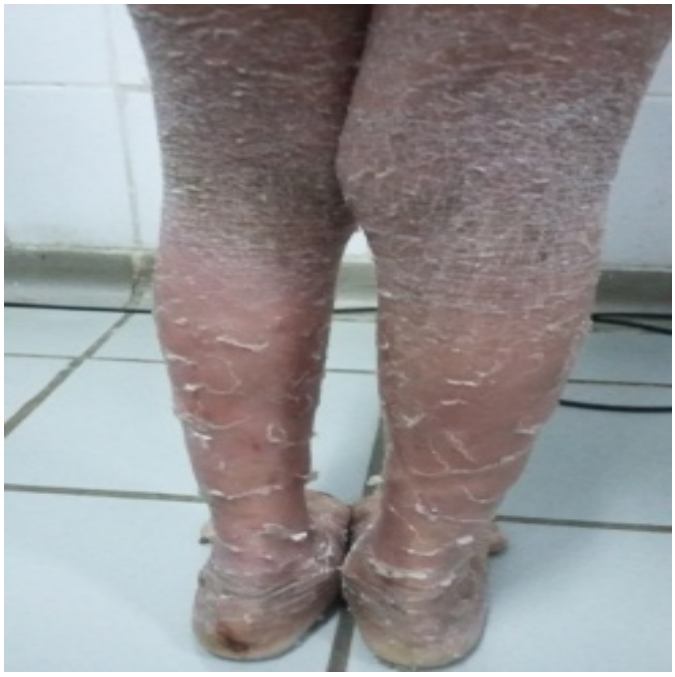
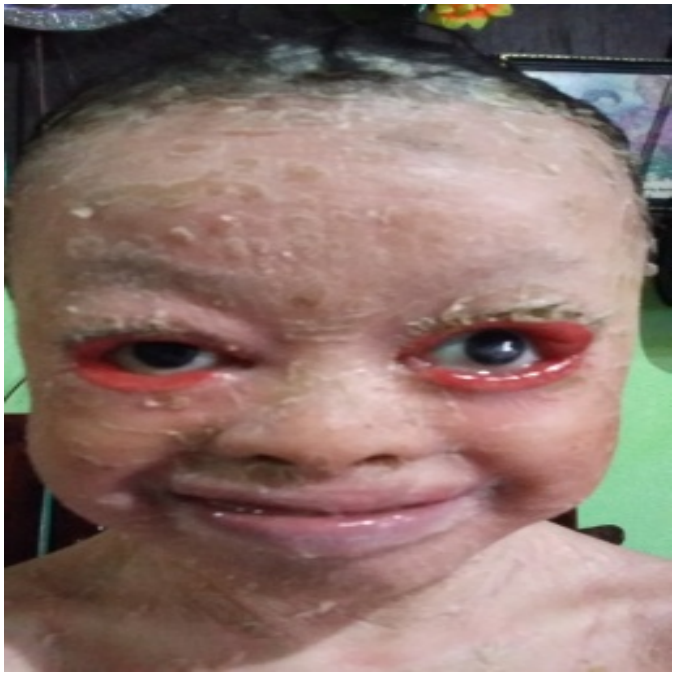


Fig.1.m.



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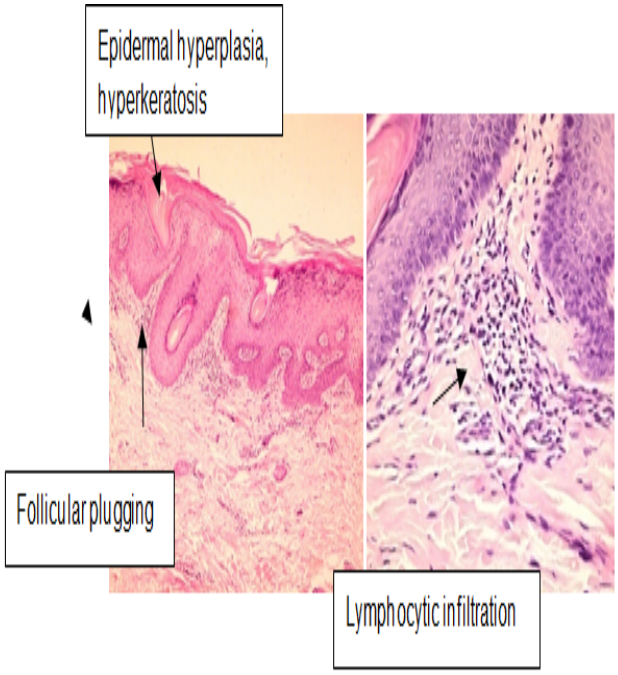
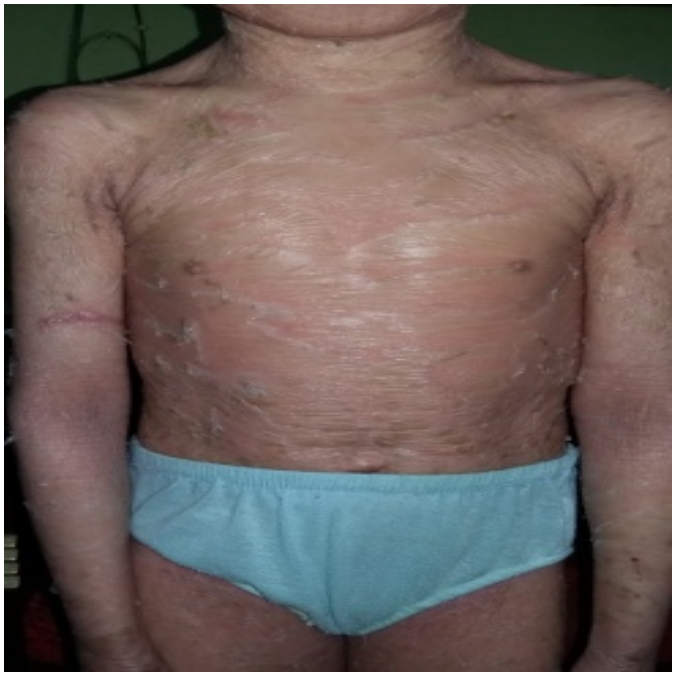


Fig.2.



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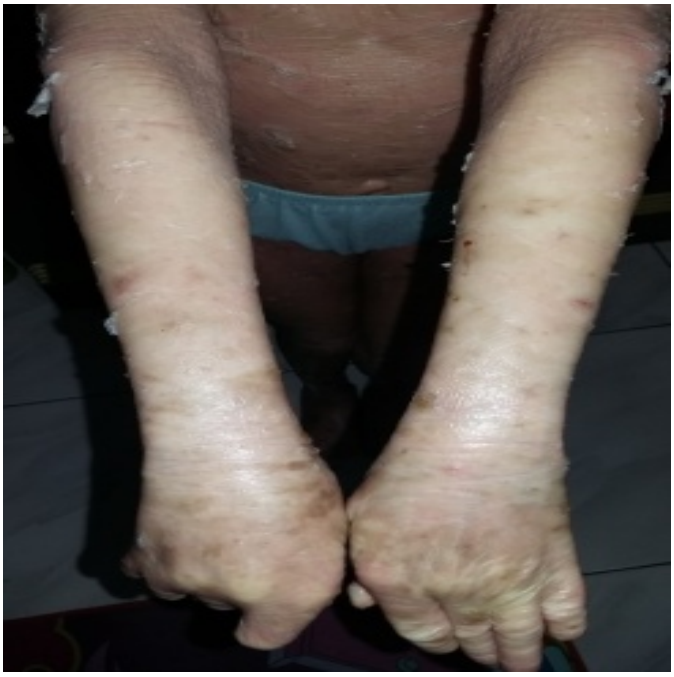

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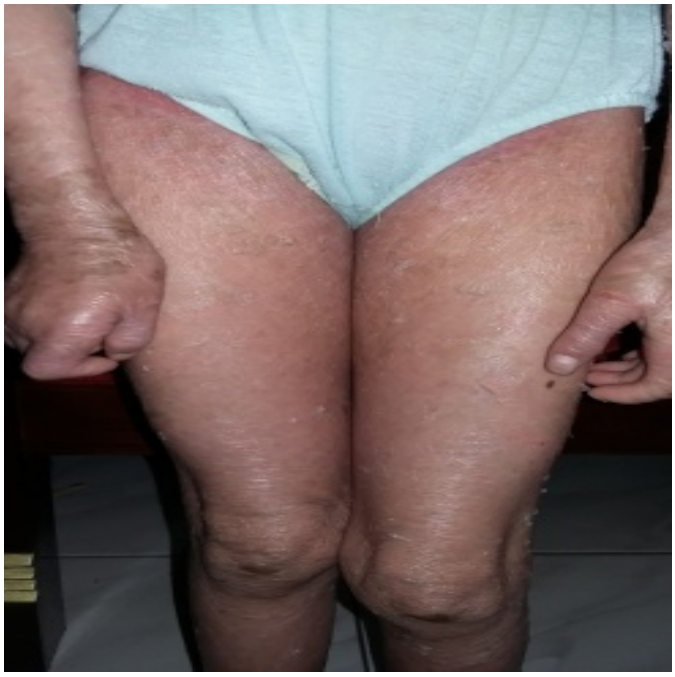

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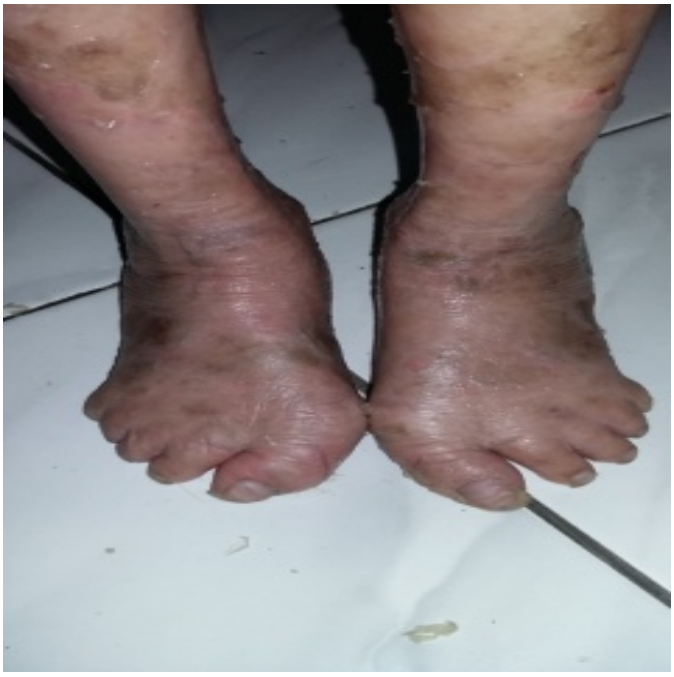

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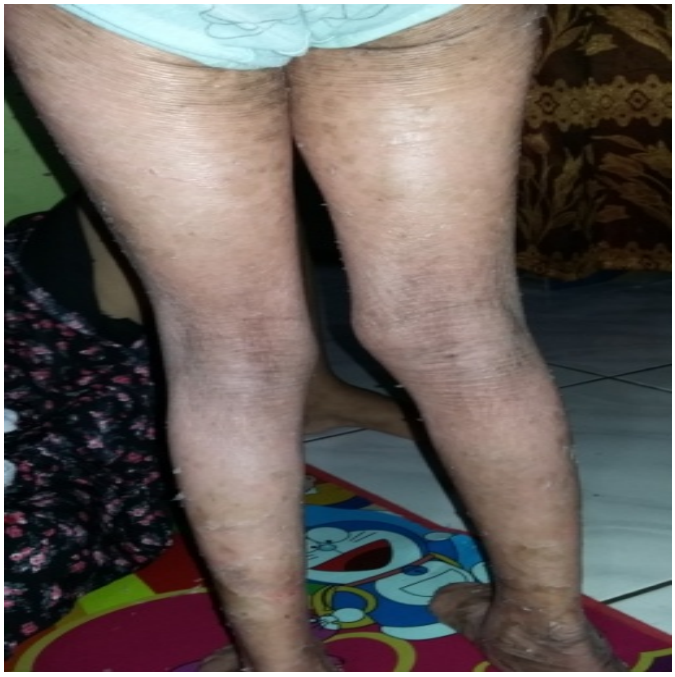

Fig.3.f.

Discussion

In this case, a 10-year-old girl came with a chief complaint of generalized scales since birth. The patient was born with a thick membrane, and the skin was dry and hard. Pruritus was sometimes present. This clinical finding is consistent with LI, which is an autosomal recessive disorder due to TGM 1 gene mutation that encodes for transglutaminase I. According to the literature, infants with LI are usually born with generalized scales since birth. They are typically born with hard parchment-like membrane during the first week of life. Initially, the skin appears to be erythematous which with time hardens and forms mosaic-pattern scales. [1,2,9] It then develops into a shiny, dry, and brownish yellow structure, resembling a parchment membrane. [2]

The physical examination showed generalized ichthyosiform lamellar scales with a mosaic pattern, fissures, and erosions. On the scalp, thick scales and alopecia were present. Ectropion was observed on both eyes as well as scales and eclabium on the perioral area. These findings are consistent with previous studies where scales in patients with LI are plate-like separated by superficial fissures. Scales denote a change in epidermal differentiation and dryness with excessive epidermal scale accumulation.2 The resulting thick stratum corneum on the scalp may lead to alopecia. Hyperkeratosis can disrupt the normal eccrine gland that causes hypohydrosis. The cutaneous changes on the eyelids cause anterior lamellar shortening with a resulting bilateral ectropion. [4],[9]

Emollient with a composition of capric triglyceride, glycerine, pentylene glycol, sodium carbomer, squalene and cocos nucifera to be applied daily in the morning and evening, cetirizine and vitamin B complex was prescribed to the patient as a symptomatic treatment. According to the literature, the management approach in LI remains supportive. Treatment aims to prevent excessive transepidermal water loss, secondary infection, and clinical symptoms. Possible topical preparations include emollients such as petrolatum, coconut oil, alpha hydroxyl acetic acid and keratolytic agents such as 0.1% retinoic acid, lactic acid, urea, a mixture of lactic acid and propylene glycol, and calcipotriol. Antihistamine was given as a symptomatic approach for the pruritus that results from the disruption of the skin barrier function and diminished sebaceous and eccrine gland function. Topical antibiotics are also widely used in LI. [1,9]

Oral retinoids are not routinely used by are recommended for neonates with severe cutaneous involvement. [2] The recommended dose ranges from 0.6-1 mg/kgBW for 6 until 16 weeks. In severe cases, topical and systemic corticosteroids may be considered. The treatment principle is hydration, moisturization, and keratolytic and is aimed at clinical improvement. [10-16]

The differential diagnosis, in this case, is congenital ichthyosiform erythroderma (CIE). CIE occurs since birth and manifests as infants covered in the shiny erythematous collodion membrane. After the membrane exfoliates, the skin remains erythematous with fine scales. In contrast to LI, ectropion, eclabium, or alopecia are rarely found in CIE. [1]

Histopathological examination showed epidermal hyperplasia with hyperkeratosis, parakeratosis, hypergranulosis with focal hypogranulosis, and follicular plugging. Perivascular lymphocytic infiltration was evident on the upper dermis. This finding is consistent with the literature, where typical LI shows hyperkeratosis, orthokeratosis with mild acanthosis, normal or mildly elevated epidermal proliferation rate.1

Conclusion and Take-Home Message

Lamellar ichthyosis is a genetically inherited disorder, and thus no definite treatment is yet to be available. Treatment is mainly symptomatic and aims to prevent excessive transepidermal water loss and secondary infections. The use of emollient, in this case, was shown to be effective in improving the skin barrier function of the patient.

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