Lamellar ichthyosis: a case report

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Ichthyoses are divided into four groups according to clinical, histopathologic and genetic findings. Lamellar ichthyosis is one of them. The incidence of lamellar ichthyosis is believed to be approximately 1 per 100,000 to 300,000 live births. It is characterized by large, polygonal, grayish brown, and tightly adherent scales. We report a four-year-old boy with desquamative lesions since birth who had six-year-old sister with similar lesions, suggesting an autosomal recessive inheritance. His skin biopsy revealed hyperkeratosis with lamellae. There were no associated hair or neurological abnormalities. His clinical and histopathological findings were typical for isolated lamellar ichthyosis. Because of its rare occurrence, we report this case with a review of the literature.

Key words: lamellar ichthyosis, treatment, ectropion.

Lamellar ichthyosis (LI) is a rare autosomally inherited disorder of keratinization. The incidence is believed to be approximately 1 per 100,000 to 300,000 live births. Males and females are affected equally, with no ethnic preponderance. The major distinguishing feature is clinically the presence of thick hyperkeratotic scaling all over the skin surface[1,2]. Here, we discuss an uncommon case of ichthyosis and review the literature.

Case Report

A four-year-old boy suffered from desquamative lesions since birth. His past history was nonsignificant except for repair of ectropion when he was two years old. He is the product of a consanguineous marriage. His parents have two sons and one daughter. The six-year-old daughter has a similar skin problem.

On admission, his physical examination revealed a height of 102 cm (50p), weight of 15 kg (25p), and large, brown, centrally attached scales covering the entire body including the flexural folds and nonpalpable left testis (Figs. 1 and 2). All other findings on physical examination were normal. His hematological and biochemical tests were within normal limits. In skin biopsy, hyperkeratosis with lamellae was found. The patient was diagnosed as LI with clinical and histopathologic findings.

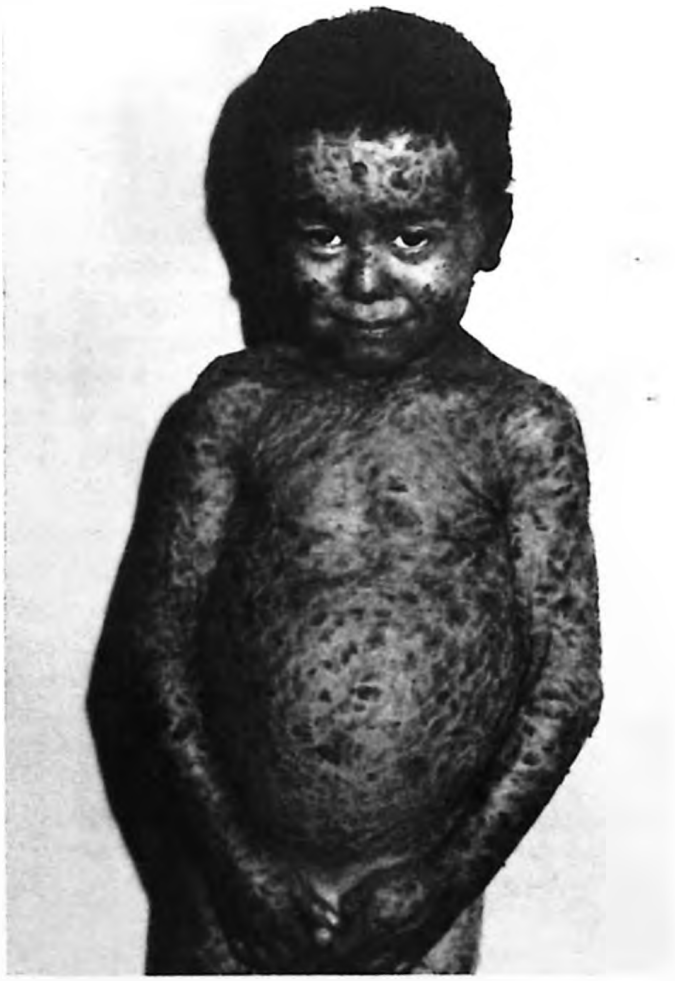

Fig. 1. General view of the patient.

Fig. 2. General view of the patient.

Discussion

The ichthyoses are a heterogeneous group of diseases that represent abnormalities in the formation and desquamation of the keratinocytes. There are four types, with different clinical, histopathologic and genetic findings (Table I). LI is one of them. LI is present at birth with collodion baby or becomes apparent soon thereafter and almost always involves the entire cutaneous surface. It is characterized by large, polygonal, grayish brown, and tightly adherent scales. Nails can be dystrophic. Palms and soles show thickening. Persistent ectropion of the eyelids is almost always present and is a helpful diagnostic sign. LI is generally inherited by an autosomal pattern[1-3].

The histologic findings of LI show proliferation, hyperkeratosis with acanthosis, hypergranulosis, and ortho- and parakeratosis. In the autosomal-dominant form, some ultrastructural similarities to the autosomal-recessive form are found, but it can be distinguished by a prominent regular keratin pattern of horny cells and the presence of only a few lipid inclusions in the corneocytes[1].

The mechanism of the disease is not yet known. Epidermal proliferation rates are normal or slightly elevated. In some families, transglutaminase 1 gene mutations have been identified as causative genetic defects[4,5]. This gene encodes the keratinocyte transglutaminase which is responsible for cross-linking epidermal proteins during formation of the stratum corneum. But the transglutaminase activity measured in some patients was within the normal range[6]. A new gene of lamellar ichthyosis was identified in a 6-cM interval on chromosome 19 during the examinations of nine large consanguineous families[3]. These results indicate that LI is a genetically heterogeneous disorder.

Alpha-hydroxy acids, urea-based creams, and propylene glycol are used for the aggressive moisturization which is the mainstay of treatment. Oral retinoids provide relief for LI, but the adverse effects of long-term maintenance therapy do not allow for prolonged use. Therefore topical retinoid appears to be an alternative approach in the treatment of lamellar ichthyosis[7]. Clinical trials with retroviral vectors expressing

Table I. Classification of Ichthyoses[2,8]

Type	Incidence	Genetics	Onset	Cause	Ectropion	Flexural affection	Prognosis
Ichthyosis vulgaris	1:250 (common)	Autosomal dominant	1-4 years	Profilaggrin expression ↓	No	No	Improves
Sex-linked ichthyosis	1: 2,000-6,000 males	Sex-linked recessive	Birth-1 year	Steroid sulfatase ↓	No	Occasional	Stable
Lamellar ichthyosis	1: 200,000-300,000	Autosomal recessive-dominant	Birth	Defect in transgluta-minase gene	Common	Always	Worsens
Epidermolytic keratosis	1: 300,000	Autosomal dominant	Birth-6 months	Defect in keratin 1 or 10	No	Always	Improves

transglutaminase 1 have been successful in the laboratory. Therefore gene therapy is the great hope of the future. Prenatal diagnosis can be done early in the first timester via chorionic villous sampling or prenatal ultrasonography. Characteristically, the ultrasonography demonstrates the collodian baby appearance during the intrauterine period. Intrauterine biopsying of the fetal skin is not useful because of false-positive and false-negative skin biopsy results[3,8].

In our case, the presence of ectropion, the persistence of skin lesions since birth, the autosomal recessive genetic pattern of inheritance as suggested by the parental consanguinity and histopathological findings revealed the diagnosis of LI. Characteristic hair findings (trichorrhexis invaginata) and neurological abnormalities which are seen in Netherton's syndrome and Sjögren-Larsson syndrome, respectively, which are associated with ichthyosis, were absent in our patient. For this reason, we considered the patient as isolated LI.

REFERENCES

1. Kolde G. Happle R. Traupe H. Autosomal-dominant lamellar ichthyosis: ultrastructural charcteristics of a new type of congenital ichthyosis. Arch Dermatol Res 1985; 278: 1-5.
2. Mansour AM, Traboulsi AI, Frangieh GT. Jarudi N. Unilateral magalocornea in lamellar ichthyosis. Ann Ophthalmol 1985; 17: 466-470.
3. Fischer J, Faure A, Bouadjar B, et al. Two new loci for autosomal recessive ichthyosis on chromosomes 3p21 and 19p12-q12 and evidence for further genetic heterogeneity. Am J Hum Gen 2000; 66: 904-913.
4. Tok J, Garzon MC, Cserhalmi-Friedman P, Lam HM, Spitz JL, Christiano AM. Identification of mutations in the transglutaminase 1 gene in lamellar ichthyosis. Exp Dermatol 1999; 8: 128-133.
5. Hennies HC, Raghunath M, Wiebe V, et al. Genetic and immunohistochemical detection of mutations inactivating the keratinocyte transglutaminase in patients with lamellar ichthyosis. Hum Genet 1998; 102: 314-318.
6. Huber M, Rettler I, Bernasconi K, Wyss M, Hohl D. Lamellar ichthyosis is genetically heterogenous-cases with normal keratinocyte transglutaminase. J Invest Dermatol 1995; 105: 653-654.
7. Steijlen PM, Reifenschweiler DO. Ramaekers FC, et al. Topical treatment of ichthyoses and Darier's disease with 13-cis-retinoic acid. A clinical and immunohistochemical study. Arch Dermatol Res 1993; 285: 221-226.
8. Shwayder T. Ichthyosis in a nutshell. Pediatr Rev 1999; 20: 5-12.