Case 16

A 25-year-old Thai woman from Patumthani

Chief complaint: white scaly erythematous skin on the face, scalp, trunk and extremities since birth.

Present illness: The patient has fine white scale erythematous skin without bullous lesion on the face, scalp, trunk and extremities since she was born.The only treatment she received was topical medication from a community hospital. She had decrease sweat and experienced flushing on her face in hot climate.

Past history: She was a full term baby, normal ANC.

Family history: She is the only child in her family. No history of the congenital skin disorder in her family

Physical examination (at present):
A woman with normal growth and development
Skin examination revealed palmoplantar hyperkeratosis and desquamation
No ectropian or eclabium. Her hair, nails and teeth appeared normal (Figure 16.1, Figure 16.2)

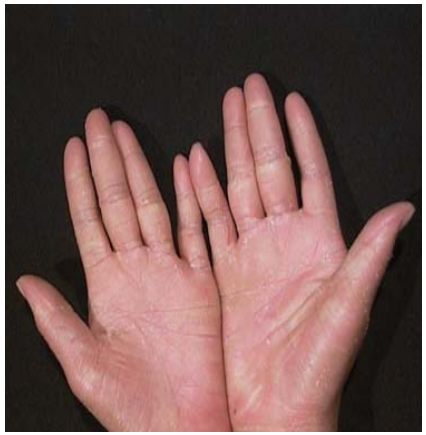
Fig. 16.1

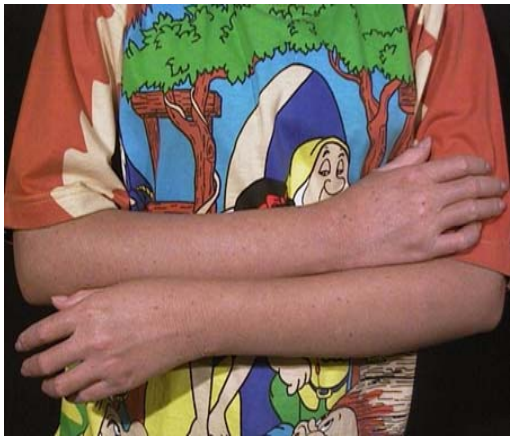
Fig. 16.2

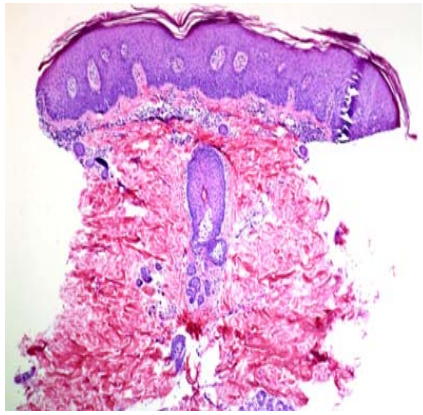
Fig. 16.3

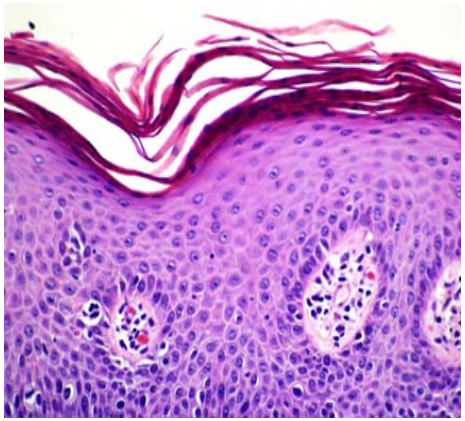
Fig. 16.4

Histopathology: (S42-0258A, B) (Fig. 16.3, 16.4)
- Orthokeratosis, focal parakeratosis with hypogranulosis and psoriasiform epidermal hyperplasia
- Superficial perivascular infiltrate of lymphocytes in the upper dermis

Diagnosis: Nonbullous congenital ichthyosiform erythroderma
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Discussion:

Nonbullous congenital ichthyosiform erythroderma is one form of severe congenital ichthyosis. Most of the patients inherit the disease in an autosomal recessive trait[1]. This type of congenital ichthyosis has been maped to at least four different genetic loci at chromosomes 14q12 (TGM1), 17p13.1 (ALOXE3; ALOX12B), 5q33.3 (ichthyin) and 19p13.1 –p13.2[2].

An effected child is frequently born as a collodion baby. In the classic CIE, the entire body is covered in erythrodermic skin with fine white scales without blister formation[3]. Mild ectropian, eclabium or alopecia are common and the palms and soles are hyperkeratotic. Hypoplasia of nasal and auricular cartilage, secondary nail dystrophy and onychomycosis may be seen, usually neither extracutaneous symptoms nor other congenital anomalies are seen. The obstruction of sweat ducts and pores results in hypohidrosis and heat intolerance.[4]

Histopathologic examination show focal or extensive parakeratosis, hypergranulosis and the epidermal acanthosis but are non-diagnostic. Other diagnostic test is molecular testing which is limited to families with known TGM1, ALOXE3, ALOX12B and ichthyin defects.

The aim of therapy is symptomatic, the correction of the epidermal function and prevention of complication, by focusing on hydration, lubrication and keratolysis. Topical treatment with retinoids and vitamin D may be effective but can be irritating in some patients. Systemic retinoids are useful, leading to significant physical and psychosocial benefits, including decrease the severity of scale, improvement of heat tolerance, sweating, ectropion and a decrease in the tendency towards future ectropion development[5, 6].

Reference

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