Case 11

A 3 year-old boy.

C/C : Blisters continually appeared since birth.

P/I : He was born with blisters which limited primarily to the hands and feet.

The skin lesions had progressed to generalized blistering which finally eroded and healed with atrophic scars. Toenails became dystrophic and lost.

PHx : Full term baby with normal labor and normal development.

FHx : No family history of similar skin lesions.

P/E : A Thai boy, not pale, no jaundice

Heent normal eyes, erosion of tonque, normal teeth

Skin Milia at forehead, generalized superficial erosion with scales and crusts and atrophic scars at trunk and extremities. few bullae at right thigh. 3 toenails were absent and the others were dystrophic.

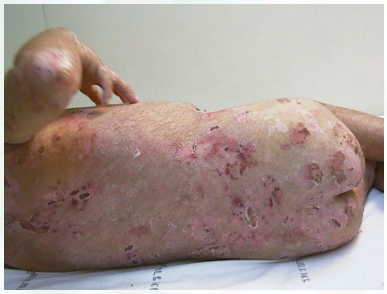

Fig. 11.1

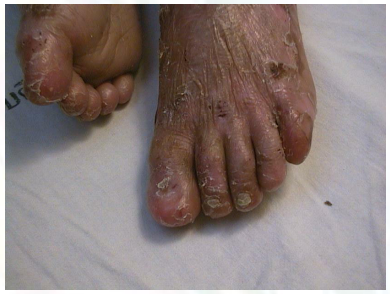

Fig. 11.2

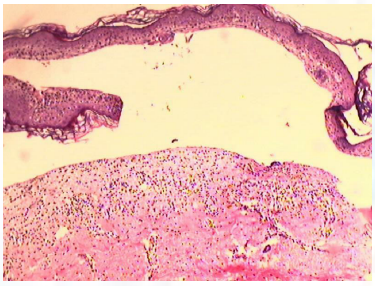

Fig. 11.3

Histopathology : Slide no. 41-1474.

Subepidermal blister
Moderately dense infiltrates of lymphocytes and neutrophils.
Fibrosis of thickened papillary dermis.

Microscopic diagnosis : Subepidermal vesicular dermatitis

EM : Pending

Diagnosis : Dystrophic epidermolysis bullosa

Treatment : Palliative treatment

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Dermtologist consultant : Penwadee Timpatanapong, M.D.

References :

1. Dunhill MGS, Rodeck CH, Richards AJ, et al. Use of type VII collagen [COL7A1] marker in prenatal diagnosis of recessive dystrophic epidermolysis bullosa. J Med Genet 1995 ; 32 : 749-50.
2. Dunhill MGS, McGrath JA, Richards AJ, et al. Clinicopathological correlations of compound heterogenous COL7A1 mutations in recessive dystrophic epidermolysis bullosa. J Invest Dermatol 1996 ; 107 : 171-7.