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DYSTROPHIC EPIDERMOLYSIS BULLOSA (COCKAYNE-TOURAINE TYPE)

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Dystrophic epidermolysis bullosa of Cockayne-Touraine type in a 37-year-old male was diagnosed on the basis of genetic transmission, clinical findings and histopathological evidence. Nine members of his family including himself in 4 generations suffered from the disease.

Key words : Epidermolysis bullosa dystrophica, Cockayne-Touraine type.

Epidermolysis bullosa is now classified on the basis of clinical patterns, genetic transmission, laboratory and electronmicroscopic findings.[1] Two varieties of the dystrophic type of disorder are documented, (a) Cockayne-Touraine type, and (b) Pasini or albopapuloid type. The common features of Cockayne-Touraine type are : early onset, blisters localised to the extremities, healing with milia and hypertrophic scars, and dystrophic or absent nails. Occasionally, healing results in hyperkeratosis of the skin. Mouth lesions are uncommon and teeth are normal. In the Pasini variant, blisters are more extensive, but later get localised to the hands and feet, elbows and knees, healing occurs with atrophic scars, and spontaneous appearance of albopapuloid lesions on the trunk without preceding blisters is a characteristic feature.

Case Report

A 37-year-old male had been having recurring blisters on the upper and lower extremities since childhood. He was the second of 9 siblings (6 brothers and 3 sisters). Four of them including the patient himself, had similar skin lesions. On tracing the family tree, the disease was found to have manifested in 4 generations involving 8 males and 1 female including one son of the proband (Fig. 1). In all the affected members,

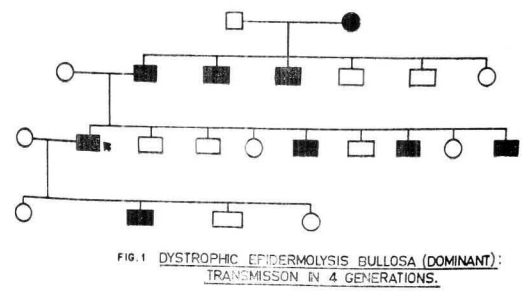

Fig. 1. Dystrophic epidermolysis bullosa (dominant) : transmission in 4 generations.

the disease was localised to the extremities only. General and systemic examination revealed no abnormality. The blisters were present on the knees, elbows and legs with depigmentation and hypertrophic scarring. Milia were present in some healed lesions. Hyperkeratotic, skin-coloured discrete papules were seen on the elbows, forearms, dorsa of hands, feet and legs. A few papular lesions were scaly and excoriated. On the trunk and buttocks, multiple hypopigmented, healed, atrophic scars were detected. Toe nails showed onycholysis, while left finger nails were thickened, pigmented and lustureless. Mucosal surfaces were normal.

Routine blood, urine and stools examination were normal. Skin biopsy from a blister revealed subepidermal bulla with a mononuclear cell infiltrate in the dermis. Another biopsy from

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a hyperkeratotic papule also showed subepidermal bulla. Sections stained for amyloid were negative. For want of facilities, the lesion could not be studied ultrastructurally.

Comments

Genetic transmission, clinical pattern and histopathological findings suggest the diagnosis of dominant dystrophic epidermolysis bullosa in our case. General physical and mental development were normal and hair and teeth were unaffected in this variety.[2] The clinical findings are more representative of Cockayne-Touraine type of disease.

Enamel hypoplasia has been described as an occasional feature of dominant dystrophic EB.[3,4] However, in our case, the teeth were normal.

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