Chapter 2 Abnormalities of Keratinization

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Acrokeratoses

Acrokeratosis Verruciformis

This rare disease was first reported by Hopf in 1931 [1].

Clinical Presentation. The eruption appears at birth, in infancy, or in childhood. The lesions consist of skin-colored verrucous or lichenoid papules, typically affecting the dorsa of the hands and fingers. The dorsa of the feet, the palms and soles, and flexors of the fingers, wrists, and forearms may be involved. Occasionally, other sites may be affected. The palmar and plantar lesions appear as translucent punctae. The nails may be white and thickened (Figs. 2.1–2.4).

Pathology and Pathophysiology. There is hyperkeratosis, hypergranulosis, and acanthosis. There is also papillomatosis, which is frequently associated with circumscribed elevations of the epidermis resembling church spires [2]. The latter feature is quite typical of this condition. There is no vacuolization of epidermal cells. The dermis is unremarkable. The association of acrokeratosis verruciformis and keratosis follicularis is so frequent that the former may be considered a forme fruste of the latter [3]. However, the occurrence of acrokeratosis verruciformis in families with Darier disease [4] may point to a coincidental association.

Inheritance. Autosomal dominant [4].

Treatment. If the lesions are few, superficial destruction with electrodesiccation or liquid nitrogen may be attempted. Otherwise there is no specific treatment.

Van den Bosch Syndrome

Van den Bosch described this syndrome in 1959 [5]. It comprises acrokeratosis verruciformis, anhidrosis, skeletal deformity, mental deficiency, and choroaderemia. This syndrome is inherited as X-linked recessive.

References


Erythrokeratodermias

Erythrokeratodermia Variabilis

(Mendes da Costa Syndrome, Keratosis Rubra Figurata, Erythrokeratodermia Figurata Variabilis)

Since 1925, when Mendes da Costa [1] described this rare disorder, many reports have appeared attesting to its two distinctive components, erythema and hyperkeratosis [2, 3].

Clinical Presentation. The disease starts in infancy. There are hyperkeratotic plaques, which are usually persistent, involving the extremities, buttocks, and face. These plaques have sharp margins and may be circinate, polycyclic, or have other configurations. The erythodermic patches are also sharply demarcated, confluent, and varying in size, location, and duration. The appearance of these patches may be associated with environmental and emotional influences. The erythemas are transient and may last for hours or days. Occasionally, fixed hyperkeratotic patches are superimposed on the erythematous areas. There are no associated changes in the hair, nails, or internal organs.

Pathology and Pathophysiology. The involved skin shows orthokeratotic hyperkeratosis, moderate to marked acanthosis and papillomatosis, and a normal granular cell layer. The epidermis has a saw-toothed appearance. The vessels in the papillary dermis are straight, prominent, and surrounded by a modest inflammatory infiltrate [4, 5]. There is an increase in the amounts of hydrolytic enzymes in the epidermis [4, 5]. Ultrastructurally, there are increased numbers of unmyelinated nerves in the papillary dermis and a pronounced reduction of keratinosomes in the epidermis [5]. Autoradiographic studies show normal mitotic indices [6]. All of these findings confirm that erythrokerato-