Familial Kyrle's disease: a case report

Kyrle's disease (KD) is a rare focal keratinization disorder of unknown etiology, with no sex or racial predilection.[1] The hereditary nature of the disease has been disputed.[2] Characteristically, KD is said not to involve mucous membranes, plantar, or palmar surfaces.[3] We report a rare case of KD with mucosal and palmo-plantar involvement in multiple family members.

Case report

A 30-year-old man presented with multiple, asymptomatic skin lesions, which started at the age of 5 over the shins, progressed to involve the forearms, upper back, palms, and soles. There was history of similar lesions in several other family members spanning over four generations.

Dermatological examination revealed 1–20 mm greyish verrucous papules and nodules distributed bilaterally over the patient's arms, forearms, palms, upper back, thighs, legs, and soles. Most lesions showed tightly adherent scales, with few showing hyperkeratotic masses extruding from the center. Early lesions were skin colored, pin-head sized, hyperkeratotic papules. Healed areas showed atrophic depressed scars with spotty hyperpigmentation. New lesions emerged around old scars. Palms showed hyperkeratotic papules near wrists and pits along palmar creases. Soles showed large hyperkeratotic nodules with central horny mass and hyperkeratotic plaques, mainly over weight-bearing areas.

Ophthalmic examination revealed conjunctival dryness, pseudo-pterygium medially, palpebral adhesions, and ectropion uvea. Nodular lesions were seen at the limbus, extending from 7 o'clock to 9 o'clock position on the right eye and 3 o'clock to 5 o'clock position on the left eye. Fundoscopy was normal. Visual acuity was decreased (6/24) in right eye. Oral cavity showed chronic generalized periodontitis with spontaneous loss of few teeth (Fig. 1). Hairs and nails were normal. Similar skin lesions with involvement of palms, soles, eye changes, and dental anomalies were also seen in two siblings of our patient and their children (Fig. 2).

Except for an absolute eosinophil count of 720 per mm^3 (10%), other laboratory tests such as blood biochemistry, urinalysis, chest X-ray, and liver and renal functions were normal.

Histopathology of skin lesion revealed irregular acanthosis with parakeratotic plug containing basophilic debris invaginating into the epidermis. The plug penetrated the dermis at the deepest point of invagination. Dyskeratotic keratinization was seen extending to the basal cell zone beneath the plug. Dermis showed moderate amount of chronic inflammatory infiltration. Staining for elastic tissue and collagen was negative. Biopsy of conjunctival lesion showed hyperkeratosis, areas of parakeratosis, acanthosis, and islands of dyskeratotic cells in the epithelium. The subepithelium showed dense chronic inflammatory cells. Corneal biopsy revealed hyperplastic squamous epithelium with mild parakeratosis and acanthosis. The subepithelium showed mixed inflammatory cells at focal points.

Figure 1 Ocular involvement showing bilateral limbal nodular lesions with dental anomalies and palmar lesions

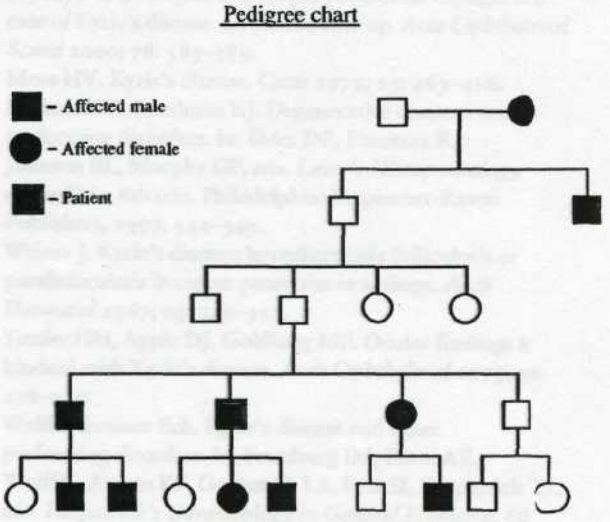

Figure 2 Pedigree chart showing five generations

Discussion

Kyrle's disease is a chronic keratinization disorder characterized by transepidermal elimination of abnormal keratin.[4] It predominantly affects young adults, mainly in the third decade, but has also been reported in children.[5,6] Its disputed hereditary nature notwithstanding, KD has been reported in siblings[5] and in a family of three generations,[6] suggesting a possible genetic background, with either an autosomal dominant[6] or autosomal recessive[7] inheritance.

Our patient had characteristic hyperkeratotic papules and nodules distributed mainly over his extremities with

histopathological features highly suggestive of KD. In his family comprising 22 members spread through five generations, nine had history of similar skin lesions with involvement of the palms and soles, highly suggestive of an autosomal dominant mode of inheritance.

The following differential diagnoses were considered and excluded accordingly:

1 Perforating folliculitis (PF) shows discrete, 2–8 mm follicular keratotic papules distributed on hairy parts of extremities and histopathologically characterized by keratotic plug, basophilic debris, and brightly staining eosinophilic elastic fibers in the dilated hair follicle. It was excluded in our case by the presence of palmar, plantar, and ocular lesions and histopathology.

2 Reactive perforating collagenosis (RPC) shows 5–10 mm umblicated keratotic papules distributed over hands, forearms, trunk, and face and histopathologically reveals thin epidermis with a cup-shaped depression plugged with parakeratotic keratin, basophilic collagen, and numerous pyknotic inflammatory cell nuclei.

3 Elastosis perforans serpiginosa (EPS) shows 2–3 mm keratotic papules in an annular/circinate configuration distributed over nape and sides of neck, face, and upper extremities. Histopathology reveals plugged, winding, tortuous, or straight canals with a focal increase of elastic tissue in dermal papillae.[2,7] RPC and EPS are excluded in our patient based on distribution and morphology of lesions, histopathological features and negative collagen/elastic tissue staining in the plug.

Only two reports of ocular changes with KD exist in literature, although Carter and Constantine[1] stated that KD does not involve mucous membranes. Alyahya *et al.*[2] reported the first case of KD with conjunctival and corneal involvement. Tessler[6] reported posterior subcapsular cataract and bilateral subepithelial anterior stromal scarring of cornea in 3 out of 13 family members affected with KD.

Although characteristically KD is said not to involve plantar or palmar surfaces, there are reports of circumscribed palmo-plantar hyperkeratosis[8] and hyperkeratosis punctata of palmar creases[9] in KD.

Skin lesions with clinical and histopathological similarities to KD, associated with diabetes mellitus, chronic hepatic disease, internal malignancies, congestive heart disease, or renal failure are termed "acquired perforating dermatoses."[10]

In the present case, several interesting and unique features have been observed. Our series showed involvement of cornea and conjunctiva, palms and soles with dental anamolies in all affected family members in an autosomal dominant pattern.

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