CASE REPORT

Mucocutaneous dyskeratosis with periodontal destruction and premature tooth loss

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We report the case of a 16-month-old boy who presented an exuberant erythematous gingival swelling and severe tooth mobility. Radiographic examination confirmed alveolar bone loss, and gingival biopsy showed epithelium containing numerous dyskeratotic cells. Because of feeding difficulties, the enlarged gingival tissue and involved teeth were removed. One year later, similar problems were encountered during the eruption of the deciduous second molars. The patient also exhibited papular skin lesions. Histopathologic features on biopsies of the skin and oral lesions were similar. The oral and cutaneous lesions presented by this patient were similar to those described by From et al. in 1978 in a father and son, reported as dyskeratosis benigna intraepithelialis mucosae et cutis hereditaria—the sole report in the English language. To avoid confusion with hereditary benign intraepithelial dyskeratosis (Witkop–von Sallmann syndrome) we have renamed the condition as mucocutaneous dyskeratosis with periodontal destruction and tooth loss. (Oral Surg Oral Med Oral Pathol Oral Radiol 2012;113:254-259)

Dyskeratosis is characterized by premature single-cell keratinization, usually caused by alterations in cellular desmosomal proteins or cytokeratins. Dyskeratotic cells are isolated round cells, with pyknotic dark-staining nuclei and clear or eosinophilic cytoplasm, seen mainly in the spinous layer, in areas of acantholysis or in various mucocutaneous conditions, including Darier disease, warty dyskeratoma, hereditary benign intraepithelial dyskeratosis (Witkop–von Sallmann syndrome), pachyonychia congenita, and hereditary mucoepithelial dysplasia. Typically, the oral lesions in these disorders show hyperkeratotic white papules or plaques, mainly seen on the palate, tongue, and buccal mucosa.

From et al.[1] described a new entity entitled dyskeratosis benigna intraepithelialis mucosae et cutis hereditaria. That report, the only one to date in the English language, described a father and son who had benign intraepithelial dyskeratosis affecting skin, bulbar conjunctiva, and oral mucosa. This condition differed from other dyskeratotic diseases, in that oral involvement was mainly restricted to the tooth-bearing areas, causing premature tooth loss. It is the only dyskeratotic condition reported causing tooth loss. We describe a similar case, but without ocular involvement and with a sporadic presentation. We consider mucocutaneous dyskeratosis with periodontal destruction and premature tooth loss a more appropriate nomenclature for this condition.

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A white male child aged 16 months was referred for evaluation of a slow-growing gingival swelling and tooth mobility. The gingival swelling was first noticed when the child was 7 months old and the mandibular central incisors were erupting. There was no consanguinity, with both parents aged 24 years old at the time of conception. The patient was the third of 3 siblings and was delivered with no complications. The patient had normal development, with no problems of vision and hearing or any other ectodermal manifestations or internal organ abnormalities. Height, weight, and skull circumference were normal for his age.

Intraoral examination revealed an edematous and erythematous gingival enlargement partially or totally covering the crowns of the deciduous teeth, all of which showed grade III mobility. The gingival surface showed diffuse or punctate pale-yellowish areas suggesting deposition of fibrin or keratin (Figure 1, *A*). Periapical radiographs showed pronounced alveolar bone resorption.

The clinical findings initially suggested a diagnosis of Langerhans cell histiocytosis. Complete blood count was within normal limits. Incisional biopsy was performed under general anesthesia. Histopathology showed a hyperplastic epithelium with hyperparakeratosis, acanthosis, and dyskeratosis characterized by suprabasal scattered single-cell keratinization (Figure 1, *B*). The unusual hyperkeratosis showed

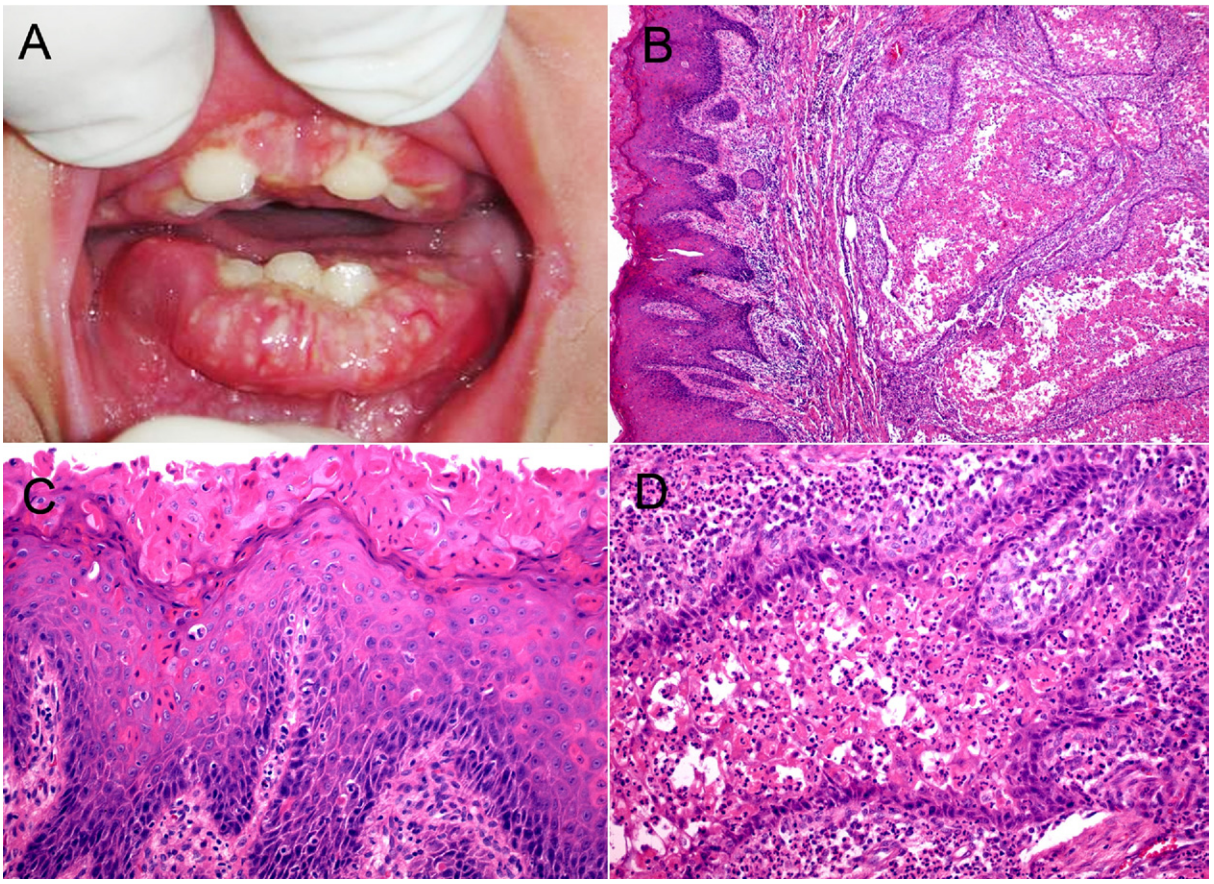

Figure. 1. **A,** Erythematous gingival enlargement with pale-yellowish areas, partially or totally covering the crowns of the erupted teeth, which showed severe mobility. **B-D,** Oral epithelium displaying pronounced single-cell keratinization in the suprabasal layers (hematoxylin-eosin [HE], **B** ×25, **C** ×200). Acantholytic cystic epithelial islands permeated by mixed inflammatory infiltrate in the connective tissue (HE, **B** ×25, **D** ×200).

focal areas containing numerous dyskeratotic cells (Figure 1, *C*). No dysplasia was seen. In the connective tissue were islands of dyskeratotic and nondyskeratotic acantholytic epithelium permeated by a mixed inflammatory infiltrate, forming pseudocystic areas containing numerous neutrophils (Figure 1, *B* and *D*). The focal areas of hyperkeratosis in the superficial epithelium and the inflamed pseudocystic structures in the connective tissue probably corresponded to the clinical appearance of pale-yellowish areas on the gingival surface. The microscopic differential diagnosis therefore included a dyskeratotic mucosal or mucocutaneous disorder with oral involvement, with no definitive diagnosis.

Because the mother had difficulties feeding the child, all erupted deciduous teeth and the enlarged gingival tissues were removed. The resulting edentulous alveolar process healed with clinically normal mucosa.

One year later, a similar gingival enlargement developed around the deciduous second molars and an erupted lower permanent incisor. Angular cheilitis was then also observed (Figure 2, *A*). The mother reported that the child had also developed recurrent episodes of red, suppurating, and itching papular eruptions on the skin of the lower limbs, buttocks, abdomen, and back, which during healing became brownish. Physical examination revealed numerous papular skin lesions showing central keratotic plugs, not exceeding 10 mm in diameter, mainly on the lower limbs, with multiple brownish macules on the back and buttocks, probably due to postinflammatory hyperpigmentation (Figure 3, *A* and *B*). Circumscribed hyperkeratotic palmoplantar lesions were also noted (Figure 3, *D*). New lesions developed in areas of trauma, characterizing the Koebner phenomenon, as seen on the ankle, where the skin was in contact with sandals. Panoramic radiography revealed that the anterior mandibular permanent teeth buds were displaced from their original position and associated with severe alveolar bone loss and surrounding radiolucent areas. The second deciduous molars were "floating," without any bone support (Figure 2, *B*). A biopsy from a skin lesion presented histopathologic features similar to those seen previously in the gingiva (Figure 3, *C*).

Based on the clinical and histopathologic findings, as well as the skin involvement, the diagnosis of mucocutaneous dyskeratosis with periodontal destruction and tooth loss (likely the same disease first described by From et al.[1] as dyskeratosis benigna intraepithelialis mucosae et cutis hereditaria) was made. However, ocular involvement was