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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

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Eruptive Vellus Hair Cyst and Epidermoid Cyst in a Patient with Pachyonychia Congenita

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Abstract

We report the first case of pachyonychia congenita (PC) associated with both eruptive vellus hair cyst (EVHC) and epidermoid cyst. The patient is a 12-year-old Japanese girl who presented with two natal teeth at birth. She had thickening and discoloration of the fingernails and toenails, plantar hyperkeratosis, palmar-plantar hyperhidrosis and multiple cutaneous cysts. Histologic examination revealed EVHC and epidermoid cyst.

Key words: eruptive vellus hair cyst; epidermoid cyst; pachyonychia congenita

Introduction

Pachyonychia congenita (PC) is a rare genetic disorder characterized by multiple ectodermal abnormalities, classified into two major clinical subtypes. The Jadassohn-Lewandowsky type (1) involves the congenital thickening of the nails, oral leukokeratosis, palmar-plantar hyperkeratosis and hyperhidrosis. The Jackson-Lawler type (2) also has congenital thickening of the nails, palmar-plantar hyperkaratosis and hyperhidrosis but lacks the oral mucosal involvement and possesses the additional features of natal teeth and multiple cutaneous cysts such as steatocystoma multiplex (3) and epidermoid cysts (4). Herein we report the first case of PC associated with both eruptive vellus hair cyst (EVHC) and epidermoid cyst.

Case Report

The patient is a 12-year-old Japanese girl who presented two natal teeth at birth. Thickening and discoloration of the fingernails and toenails were noted during the first year of life (Fig. 1). Plantar hyperkeratosis and hyperhidrosis of the palms and soles developed after age 9 (Fig. 2). Cutaneous cysts on the scalp, forehead, axilla and lower limb developed after age 10. There were one yellowish dome shaped cyst and a couple of normal skin colored small cysts within the left axilla (Fig. 3). There was no leukokeratosis oris and no hoarseness, and the eyes were normal. Her physical and mental growth had been normal. More than 10 potassium hydroxide examinations and fungal cultures of the nails on Sabouraud's agar have revealed no fungal organisms. No family members were affected and her parents were not consanguineous. A biopsy

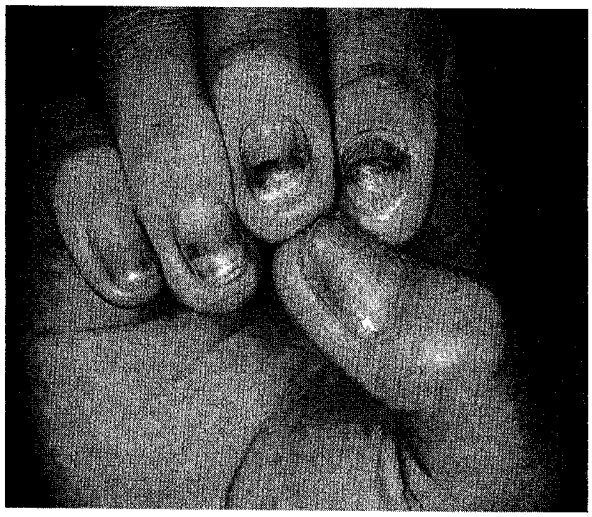

Fig. 1. Thickened fingernails

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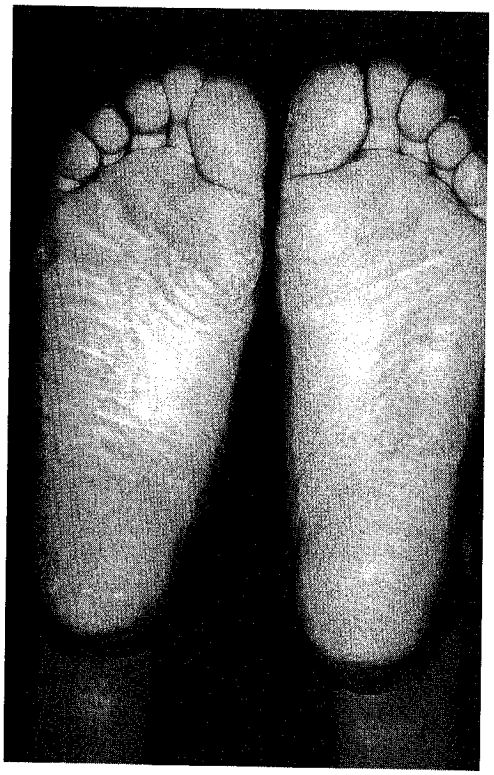

Fig. 2. Plantar hyperkeratosis.

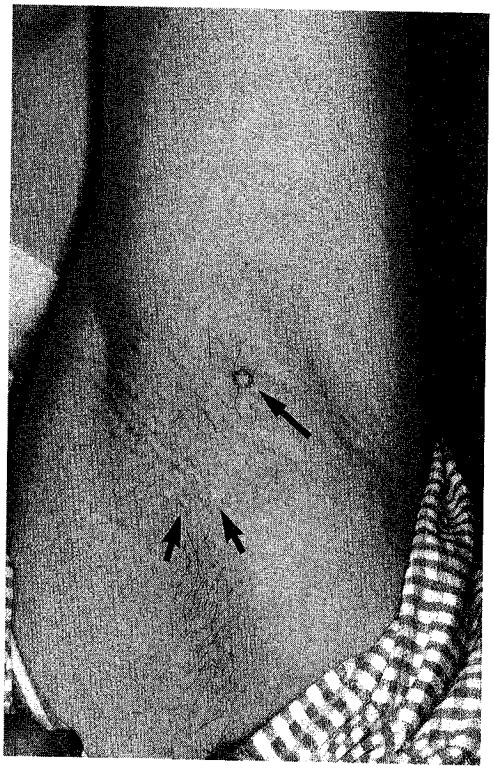

Fig. 3. One yellowish dome shaped cyst (long arrow) and a couple of normal skin colored small cysts within the left axilla (short arrows).

specimen from a normal skin colored small cyst within the left axilla showed a keratin-filled cyst in the middle dermis containing several transversely or obliquely cut portions of vellus hair (Fig. 4). The cyst was lined by squamous epithelium with focal areas of a granular layer. There was no sebaceous element. This cyst was diagnosed as EVHC. Another biopsy specimen from a yellowish dome shaped cyst within the left axilla showed a cyst lined by a squamous epithelium with a granular layer and filled with loose keratin arranged in laminated layers. In serially sectioned specimens the cyst had definite attachment to the surface of epidermis and had a telogen hair bulb extending from the lower pole towards the subcutis. This cyst was diagnosed as epidermoid cyst.

Discussion

Recently, EVHC and steatocystoma multiplex in a patient with PC has been reported (5). Also, there are several papers supporting the notion that EVHC and steatocystoma are the same entity (6–8), and they probably represent cystic hamartomas of the sebaceous duct and/or its connection to the follicular infundibulum. This point of view would explain the existence of cases of PC associated both with EVHC and with steatocystomas.

Our case is PC associated with both EVHC and epidermoid cyst. EVHC was first reported by Esterly et al. (9). The pathogenesis is unknown. A possible pathogenetic mechanism has been proposed by Esterly et al.: An initially abnormal vellus hair follicule, with keratinous plugs at the follicular infundibulum, deflects vellus hair shafts to the deeper part of the follicule, resulting in its cystic dilatation and gradual disruption of the continuity between proximal and distal follicule. The epithelium of the lumen of EVHC closely resembles the infundibular squamous epithelium in most cases and our case. EVHC may originate mainly in the infundibulum. In EVHC, cells composed of

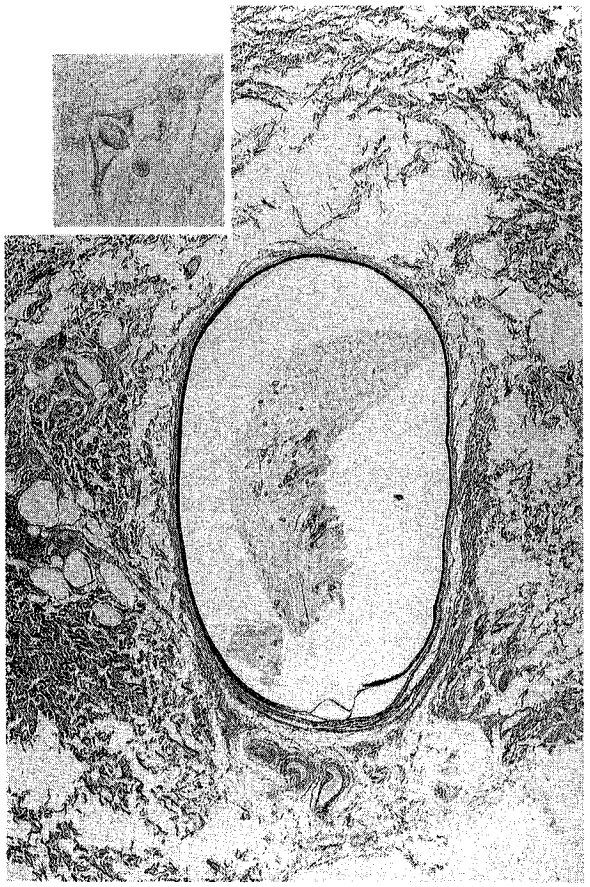

Fig. 4. Cyst containing vellus hairs lined by squamous epithelium with focal areas of a granular layer. Haematoxylin and eosin; ×20. Inset: Detail of vellus hairs. Haematoxylin and eosin; ×100.

the cyst, immunohistochemically expressed strong reaction for keratin 17 (10). Heterozygous missense mutation of keratin 17 gene have been identified in Jackson-Lawler type PC (11). We suggest that the mutation of keratin 17 gene may cause EVHC in Jackson-Lawler type PC.

It is widely assumed that most spontaneously arising epidermoid cysts are related to the follicular infundibulum. The occurrence of hybrid cysts with partially epidermal and partially trichilemmal lining favors this assumption. In regions devoid of follicles, such as palms and soles, epidermoid cysts can arise from traumatic implantation of epidermis into the dermis or subcutis. In our case, epidermoid cysts were found on the scalp, axilla and lower limb. In serially sectioned specimens, the epidermoid cyst had definite attachment to the surface epidermis and had a telogen hair bulb extending from the lower pole towards the subcutis. This histopathological finding proves that the epidermoid cysts in our case arise in the follicular infundibulum. We think that both EVHC and epidermoid cysts in our case result from a cystic change in the follicular infundibulum.

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