Familial Atrophoderma Vermiculata Associated with Epidermal Cysts

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TAtrophoderma vermiculata (AV) is a rare disease which normally occurs on the face and cheeks. In consists of numerous, closely-crowded, atrophic pits, with a bilateral distribution, producing a reticulate appearance. Familial occurrence has been noted, as well as in combination with other congenital anomalies.

Two patients, a proband and his maternal aunt, presented with the characteristic changes of facial multiple pits. A clinical diagnosis of AV was determined. In addition, multiple subcutaneous nodules were noted. Four nodules were removed and identified as epidermal cysts. A family history revealed that the proband's mother, maternal grandfather, maternal uncle, two maternal aunts and two older sisters had also suffered from similar facial skin lesions and multiple subcutaneous nodules. An autosomal dominant mode of transmission was indicative from this family tree. We report a rare case of AV associated with multiple epidermal cysts found in eight members of a family. (Ann Dermatol 17(2) 102~105, 2005)

Key Words: Atrophoderma vermiculata (AV), Autosomal dominant, Epidermal cysts

INTRODUCTION

Atrophoderma vermiculata (AV) refers to a form of keratosis pilaris atrophicans, in which the cheeks are predominantly involved. AV was first described by Unna[1] in 1894 under the name of ulerythema acneiforme. Subsequently, numerous terms have been used for this condition, including acne vermoulante, atrophodermia reticulata symmetrica faciei, folliculitis ulerythematosa reticulata, folliculitis atrophicans reticulata and honeycomb atrophy[2-4]. AV usually presents as an isolated skin defect with no associated medical conditions. Rarely it may be associated with epidermal cysts, folliculitis decalvans, leukokeratosis oris, Down's syndrome, a congenital heart disorder or neurofibromatosis[5-7]. To our knowledge, only one case of AV, which occurred in a patient with congenital heart disease, has been reported in the Korean literature[6]. Herein, we present two cases of AV associated with epidermal cysts in a nephew and a maternal aunt.

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CASE REPORT

Case 1

A 26-year-old man presented with irregular, atrophic scars on the face. The lesions had first been noted 10 years earlier, and had progressively worsened over the years. There was no prior history of papules, acne or trauma. During the same period of time, subcutaneous nodules on the chest, arms and groin had appeared. Physical examination showed marked, pitted scarring of the forehead, cheeks, nose and preauricualr areas. Some were confluent with a cribriform appearance, while others were solitary (Fig. 1). There was no involvement of

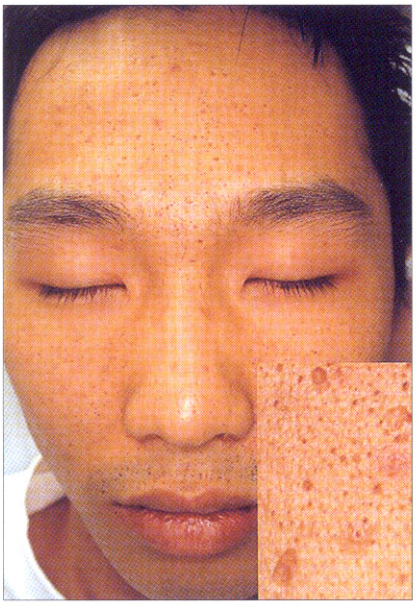

Fig. 1. Irregular, depressed, pit-like depressions on the forehead, both cheeks and nose (Case 1).

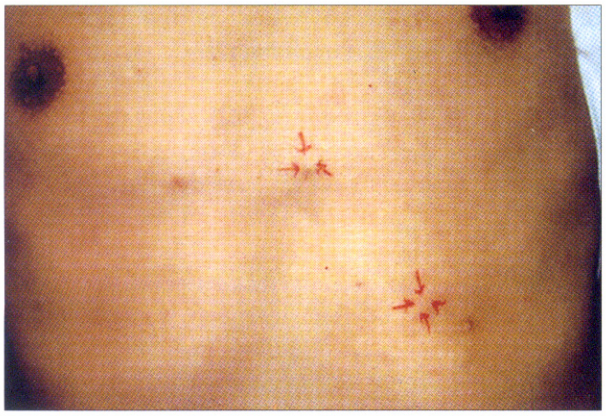

Fig. 2. Multiple, subcutaneous nodules on the chest (Case 1).

the eyebrows. A clinical diagnosis of AV was determined. More than 5 subcutaneous nodules were also discovered, and 2 of the nodules on the arm and chest were histologically identified as epidermal cysts (Fig. 2). No heart disease, hypotrichosis, basal cell carcinoma or other skin abnormalities were detected. Results of routine laboratory tests and findings on the chest X-ray were normal or negative.

Case 2

A 43-year-old woman (the proband's maternal aunt) presented with a slowly progressive scarring

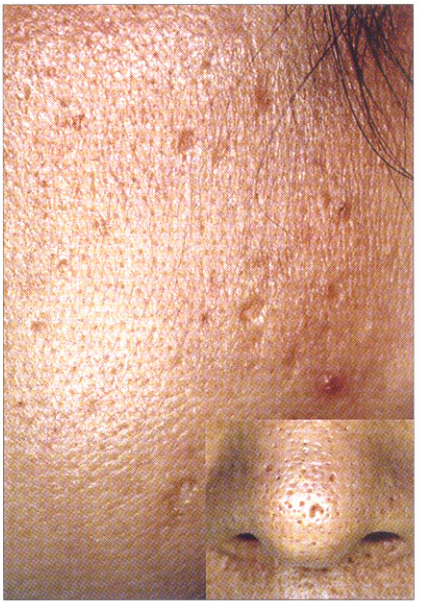

Fig. 3. Atrophic scars on the cheeks and nose (Case 2).

disorder of the face, which had been present since the age of 15. She denied preceeding inflammation. She had no history of hypohidrosis or basal cell carcinoma. Physical examination revealed multiple pits on the face (Fig. 3) and multiple subcutaneous nodules, 1-3 cm in diameter on the trunk, neck, axillae and groin. Two nodules were removed and histologically identified as epidermal cysts.

A family history revealed the proband (case 1), proband's mother, two maternal aunts (including case 2), maternal grandfather, maternal uncle and two older sisters also suffered from similar facial skin lesions and multiple subcutaneous nodules on the trunk and extremities. An autosomal dominant mode of transmission was indicated from this family tree. A pedigree is shown in Fig. 4. No other familial disorders were known.

DISCUSSION

AV is a rare skin condition, which is classified as one of the keratosis pilaris atrophicans group of disorders, the others being ulerythema oophyrogenes and keratosis pilaris decalvans. Most cases are sporadic, but autosomal dominant inheritance has been described[2,8]. Generally, AV begins in early childhood with erythematous follicular papules on the cheeks. These papules then go on to develop

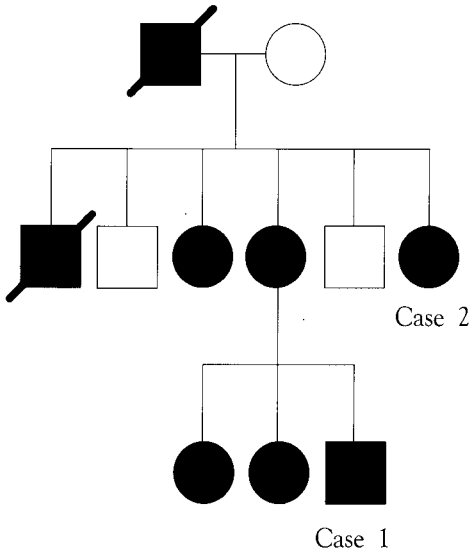

Fig. 4. Pedigree of a family with atrophoderma vermiculata.

pitted, atrophic and depressed scars in a reticulated or honeycomb pattern. These lesions can extend to the forehead, preauricular region, upper lip, ear lobes and the rarely the limbs[9]. Eyebrows, eyelashes and the scalp are notably spared. AV is typically symmetric, but unilateral occurrence has been reported [4,10,11]. The severity or morphology of lesions varies, and a few milia may be seen. The histologic examination reveals atrophy of the epidermis, fibrosis of the connective tissue, dilated follicles, dermal horn cysts and perivascular inflammation. The course is generally one of slow progressive worsening. A variety of therapeutic modalities have been used, including topical and intralesional steroids, topical tretinoin, systemic isotretinoin, dermabrasion and collagen implants[9,12]. However, there has been no consistent benefit from these treatments.

AV usually presents as a skin lesion without concomitant anomalies. Rare reports in the literature have described associations with epidermal cysts, folliculitis decalvans, a congenital heart block, atrial septal defect with Eisenmenger's complex, neurofibromatosis, Down's syndrome and Marfan syndrome[5,6,13]. In addition, various syndromes which are characterized by the presence of AV have been reported, including Rombo syndrome (milia, hypotrichosis and basal cell carcinomas), Bazex syndrome (hypohidrosis and basal cell carcinomas), Nicolau Balus syndrome (syringomas and milia) and Tuzun syndrome (scrotal tongue)[14,15].

We report on two patients, a proband and his maternal aunt, who presented with the characteristic changes of facial multiple pits. They also had multiple, 1-3 cm sized, subcutaneous nodules on the trunk, axillae, groin and extremities. Four nodules were removed and identified as epidermal cysts. In addition, seven members of the proband's family also had the characteristic facial changes of AV and multiple subcutaneous nodules. In previously reported cases of AV, although epidermal cysts or milia were rarely noted, almost all these cysts were small and limited to the facial lesions of AV.

The main interest of our cases, besides their rarity, is the observation of the association of AV with epidermal cysts. Whether the simultaneous occurrence of these diseases is coincidental or due to related pathogenic mechanisms remains to be seen. The etiology of AV is unknown, but the primary defects appear to be an abnormal keratinization of the pilosebaceous units[2]. It is widely assumed that epidermal cysts are related to the pilosebaceous unit. In addition, eight members of a family all had multiple subcutaneous nodules on the trunk and extremities, as well as the characteristic facial change associated with AV. Although only four of subcutaneous nodules were examined, it is believed that the remaining nodules may have also been epidermal cysts. These cases support a theory that AV and epidermal cysts may have a similar pathogenic mechanism.

In summary, we present two cases of AV associated with epidermal cysts in a nephew and a maternal aunt. Although these findings may be coincidental, we believe these cases highlight the association of AV with epidermal cysts. Further investigations are needed to demonstrate the pathogenesis.

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