Pachyonychia Congenita Type 2

—A Case Report—

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Pachyonychia congenita type 2 (PC-2), also known as Jackson-Lawler type PC, is a rare autosomal dominant disorder characterized by hypertrophic nail dystrophy associated with focal palmoplantar keratoderma and multiple pilosebaceous cysts. Only one case of PC-1 has been previously reported in Taiwan. In this report, we describe a typical case of PC-2, the first such case to be documented in Taiwan. The keratoderma was treated with etretinate, which was quite effective in reducing the hyperkeratotic lesions on the patient's palms and soles. (Dermatol Sinica 19: 204-209, 2001)

Key words: Pachyonychia congenita type 2, PC-2, Palmoplantar keratoderma, Etretinate

INTRODUCTION

Pachyonychia congenita (PC) is a group of rare, inherited ectodermal dysplasias whose most prominent clinical feature is hypertrophic nail dystrophy. Two main clinical variants of PC are recognized, PC-1 and PC-2. In PC-1, the pachyonychia is accompanied by severe keratoderma which mainly involves the palms and soles. Features such as angular cheilosis, follicular keratosis, hoarseness and oral leukokeratosis are not fully penetrant and occur in both types. The PC-2 form is most readily distinguished by the presence of multiple steatocysts which appear at puberty. Only one

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Accepted for publication: February 5, 2001
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case of PC-1 has been previously reported in Taiwan. In this report, we describe a typical case of PC-2, the second PC case report and the first PC-2 case report in Taiwan.

CASE REPORT

A 16-year-old Taiwanese male presented with marked thickening of all 20 nails present since infancy and palmoplantar hyperkeratosis which had developed during childhood with painful bullae making walking and participation in sports difficult. It was not clear whether natal or neonatal teeth were present. The patient's intelligence was normal and his pedigree is shown in Fig. 1. The patient's father had similar abnormally thickened nails but declined clinical examination.

Physical examination revealed follicular hyperkeratosis (Fig. 2A) on the extensor aspects of the extremities and trunk, marked thickening of all finger and toe nails (Fig. 2B), callus-like focal plantar keratoderma on the palms and soles (Fig. 2C), hyperhidrosis, and leukokeratosis of the tongue (Fig. 2D). A potassium hydroxide (KOH) mount of an oral lesion was characteristic of candidiasis. This was treated with nystatin but the leukokeratosis persisted. There was no evidence of ocular, hair or dental lesions.

Based on the characteristic clinical findings, a diagnosis of pachyonychia congenita was made. The patient was started on oral etretinate 10 mg bid for the keratoderma, which after four months of treatment was much improved (Fig. 3). The subungual hyperkeratosis persisted nevertheless. The treatment was discontinued owing to the mucocutaneous side effects of the etretinate. The patient was then lost of follow-up from 1993 until recently because he had left the country. A recent follow-up examination of the now 26-year-old patient showed that the mucocutaneous and hyperkeratotic lesions had relapsed back to their pretreatment states. KOH mounts and fungal cultures of the finger and toenails showed negative result. In addition, there were numerous cysts and nodules of various sizes all over the patient's body (Fig. 4). The cysts had first appeared on his neck and trunk two years prior and had since increased in number. Excision biopsy of one cystic lesion from the chest showed steatocystoma. With this additional finding, his pachyonychia congenita was classified as type 2.

DISCUSSION

Pachyonychia congenita (PC) is an autosomal dominant genodermatosis which was first described by Muller. It is characterized by nail dystrophy and varying features of ectodermal dysplasia. A number of classifications of this disorder have been proposed, with the classification proposed by Schonfeld based on clinical features the most widely used. Jadassohn- Lewandowsky type PC (PC-1) is characterized by onychogryphosis, focal palmoplantar keratoderma, follicular hyperkeratosis and leukoplakia of the oral mucosa. It is associated with mutations in the keratin 16 gene, or its expression partner keratin 6A. Jackson-Lawler type PC (PC-2) is characterized by, in addition to the features seen in type I, bullae, palmoplantar hyperhidrosis, and cysts arising from the hair follicle infundibulum and sebaceous duct (eruptive vellus hair cysts and steatocystomas). Other
findings in PC-2 include natal teeth, hair abnormalities (pili torti), and hidradenitis suppurativa. An asn92-to-asp mutation in the helix initiation motif of the keratin 17 gene (K17) was previously identified in a family with 25 affected members. Type III (Schafer-Brunauer) is like type I but with leukokeratosis of the corneas.

Feinstein et al. incorporated the features of steatocystoma multiplex, cataracts, angular cheilosis, mental retardation and laryngeal lesions into another classification system which is detailed in Table I. The clinical features in the patient we have described were consistent with PC-2.

PC is a rare autosomal dominant genodermatosis. Since 1904, only 300 cases of this syndrome have been reported. In Taiwan, there has only been one previous case report describing PC-1. Our case is only the second reported case of PC in Taiwan and is the first report case of PC-2.

The differential diagnosis of PC includes various conditions that are manifested by focal

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Fig. 2A
Clinical manifestations of pachyonychia congenita (PC)
A. Follicular-based keratotic papules

Fig. 2B
Yellow-brown thickening of the nails with subungual hyperkeratosis resulting in elevation and exaggeration of curvature
palmoplantar keratoderma and nail thickening, which include dermatophytosis, pityriasis rubra pilaris, psoriasis, Unna-Host disease, Papillon-Lefèvre syndrome and congenital onychogryphosis. Generally, PC should be readily distinguished from most of these conditions based on the constellation of characteristic mucocutaneous lesions. Congenital onychogryphosis is an autosomal dominant nail disorder. The onychogryphosis manifests in adolescence and predominantly affects the thumb and large toenails, lacking the massive subungual hyperkeratosis and other associated findings seen in PC.

Various forms of treatment have been proposed for the skin and nail problems associated with PC, including high doses of vitamin A, vitamin E, x-ray therapy, protective footwear, and topical aluminium chloride. None of these measures have produced any permanent solution to the distressing morbidity primarily related to the painful hyperkeratosis and blistering of the soles.

Treatment with keratolytic agents and lubricants is indicated for areas of palmar and plantar hyperkeratosis. Oral retinoids have been reported as being effective in the treatment of pachyonychia congenita. However, Soyuer and Candan failed to demonstrate any beneficial effect from a 5-week course of etretinate therapy (1mg/kg/day). Given the palmoplantar hyperkeratosis with painful bullae, our patient was treated with etretinate 10 mg bid, which was quite effective in reducing the hyperkeratotic lesions of the palms and soles. Unfortunately, the nail dystrophy showed no significant improvement after four months of treatment. Onychogryphosis does not respond to systemic therapies of this nature and treatment typically requires surgical removal of the nail plate, bed and matrix.
Fig. 3
Plantar keratoderma.
Pre- (A) and post- (B) treatment with etretinate for four months.

Fig. 4
Numerous truncal papules and nodules.

Table I. Feinstein’s classification of pachyonychia congenita.

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REFERENCES
1. Schönfeld PHIR: The pachyonychia syndrome. 