Case Report

**A Case of Type 1 Pachyonychia Congenita with Response to Acitretin**

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**Abstract**

An 8-year-old male born of nonconsanguineous marriage presented with multiple raised lesions all over the body, over palms and soles, and thickening of all nails of fingers and toes for 5 years. On examination, there were hyperkeratotic plaques over palms and soles with subungual hyperkeratosis of all finger and toe nails. Oral cavity revealed white hyperkeratotic plaque over buccal mucosa. There were hyperkeratotic papules distributed over dorsum of hands, feet, elbows, knees, trunk, and thighs. Biopsy taken from hyperkeratotic lesion over palms revealed findings suggestive of palmoplantar keratoderma. Considering the clinical features and histopathological examination, a diagnosis of Type 1 pachyonychia congenita was made. After hematological investigations, the patient was started on oral acitretin at a dose of 25 mg thrice a week. There was a marked improvement in the form of reduced hyperkeratosis over palms and soles and subsidence of hyperkeratotic papules and oral lesions, 2 months after treatment with acitretin.

**Keywords:** Acitretin, nails, pachyonychia congenita, palmoplantar keratoderma, retinoids

**Introduction**

Pachyonychia congenita (PC) is characterized by focal palmoplantar keratosis and subungual hyperkeratosis of nails. It is usually transmitted as autosomal dominant trait, but autosomal recessive forms are also described. According to Feinstein et al., four types of PC have been described. Two types are commonly reported. There is mutation of gene encoding keratin K6a and 16 in Type 1 PC and 6b and 17 in Type 2 PC. Type 1 PC (Jadassohn–Lewandowsky syndrome) is characterized by focal palmoplantar keratoderma, oral leukokeratosis, and follicular hyperkeratosis. Emollients and keratolytics may be useful in mild palmoplantar keratoderma. Systemic retinoids may be effective but causes increased tenderness of lesions in some cases. Thickened nails can be treated surgically.

**Case Report**

An 8-year-old male child, born of nonconsanguineous marriage, presented with thickening of nails, raised painful lesions over palms and soles, and raised lesions over the body for 5 years. There was no history of mental retardation, hoarseness of voice, or palmoplantar hyperhidrosis. None of the family members were affected. Cutaneous examination revealed thick hyperkeratotic plaques with fissuring over palms and soles along with subungual hyperkeratosis and discoloration of finger and toe nails. There were multiple follicular, hyperpigmented, and keratotic papules present over elbows, knees, dorsum of hands, feet, trunk, and thighs. Oral examination revealed white hyperkeratotic plaque over buccal mucosa. Other mucosa, hair, and dentition were normal. KOH microscopy and culture of nail clipping did not reveal any fungal elements. Histopathological examination of plaque over palms revealed hyperkeratosis, orthokeratosis, and prominent irregular acanthosis. All routine investigations including lipid profile were normal. Genetic studies could not be done due to economic constraints. Considering the clinical presentation and histopathological findings, he was diagnosed as Type 1 PC. He was managed with acitretin at a dose of 25 mg thrice weekly and emollient containing urea and salicylic acid. After 2 months of therapy, there was a marked improvement as

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**How to cite this article:** Supekar BB, Rambhia KD, Mukhi JI, Singh RP. A case of type 1 pachyonychia congenita with response to acitretin. Indian J Drugs Dermatol 2017;3:90-2.
evidenced by a reduction in the thickness of hyperkeratotic lesions over palms and soles with substantial relief of pain and subsidence of keratotic lesions over the body [Figures 1 and 2]. There was a significant improvement in disability and disfigurement caused by the disease.

**Discussion**

PC is a rare genodermatosis caused by mutation in keratin gene and is characterized by dystrophic, thickened nails (pachyonychia), symmetric focal palmoplantar keratoderma, and oral leukokeratosis. The first documented observation was made by Muller in 1904[4] which was followed by Wilson[5] and Jadassohn and Lewandowsky in 1906.[6] It is characterized by focal palmoplantar keratosis and subungal hyperkeratosis of nails. In other clinical variants, involvement of the nails alone has been reported.[7] According to Feinstein et al., four types of PC have been described. Type 1 PC (Jadassohn–Lewandowsky syndrome) is characterized by palmoplantar keratoderma, nail hypertrophy, oral leukokeratosis, and follicular keratosis. Type 2 PC (Jackson–Lawler syndrome) has additional findings such as bullae, hyperhidrosis of palms and soles, natal teeth, and steatocystoma multiplex along with findings of Type 1 PC. Type 3 has additional findings such as angular cheilosis, corneal dyskeratosis, and cataract. Type 4 is rarely described and characterized by laryngeal lesions, mental retardation, and hair abnormalities along with thickening of nails and palmoplantar keratoderma.[5]

Treatment of PC is notoriously difficult. Basic measures in management include topical emollients, keratolytic agents, mechanical removal of excessive hyperkeratotic skin, and avoidance of physical activity. Oral retinoids can also be effectively used.[5,8,9] Acitretin acts at cytosolic proteins and intranuclear receptors, which are part of the steroid–thyroid hormone super family. The metabolites of acitretin bind to retinoic acid receptors (RARs), leading to alteration of gene transcription through response elements which is responsible for its antiproliferative and anti-inflammatory effects. In psoriasis and other disorders of keratinization, acitretin normalizes epidermal cell proliferation, differentiation, and cornification.[10] Low-dose acitretin is used in the treatment of PC to get balance between hyperkeratinization and local residual tenderness. Hence, acitretin in low dose (25–35 mg/day) may be effective to overcome side effects and for better efficacy and safety. Regular monitoring of liver enzymes, fasting serum cholesterol, and triglycerides every 2–4 weeks for the first 2 months of therapy and then every 3 months is advised. A prolonged course of retinoids may produce a degree of flattening of the nails. There may be other chronic complications such as periosteal hyperostosis, increased sensitivity and fragility of the underlying epidermis.[11] Therefore, the use of acitretin in PC is difficult.

We described a case of Type 1 PC which has been successfully treated with oral acitretin with improvement in palmoplantar keratoderma and hyperkeratotic plaques after 2 months of therapy.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

**Financial support and sponsorship**

Nil.

**Conflicts of interest**

There are no conflicts of interest.

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