



Pachyonychia Congenita Project

15 March 2005

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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.

Dermatology Online Journal

Volume 9 Number 4

DOJ

Pachyonychia congenita, type II

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Dermatology Online Journal 9(4): 12

Contents

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Abstract

A 5-year-old girl presented with extensor hyperkeratotic papules and subungual hyperkeratosis with nail-plate discoloration affecting all twenty nails. The mother reported that her daughter had natal teeth. By report, the father has a similar history and constellation of clinical findings. The patient's clinical presentation and history was consistent with pachyonychia congenita, which is a genodermatosis linked to mutations in the genes encoding keratins 6, 16, and 17.

Clinical summary

History.—A 5-year-old girl presented with a problem of the nails, elbows and knees, present since birth. The patient was born with natal teeth. During the first year of life, numerous hyperkeratotic papules appeared over the knees and elbows. Also, the distal nail plates of all twenty nails became thick and hyperkeratotic with an associated yellow-brown discoloration. There is no history of either cutaneous cysts or oral lesions. Topical keratolytics were minimally beneficial for both the skin and nail lesions. Her father has similar cutaneous and nail findings since birth.

Physical examination.—Multiple hyperkeratotic, perifollicular papules were present over the elbows and knees. Yellow-brown discoloration and subungual hyperkeratosis with distal elevation of the nail plates affected all fingernails and toenails. There were no oral lesions.

Laboratory data.—None

Histopathology.—None



Diagnosis.—Pachyonychia congenita, type II.



Comment

Figure 1

Pachyonychia congenita is a familial anomaly that affects the skin, nails, and oral mucosa. In most cases, the disease displays an autosomal dominant pattern of inheritance. Clinical findings that appear during the first year of life are thick hyperkeratotic fingernails and toenails with onycholysis, and frequent paronychia infections. The elbows and knees show numerous follicular keratoses, and there is palmoplantar hyperkeratosis. Furthermore, there may be palmar and plantar hyperhidrosis. Friction blisters may appear on both the palms and soles. Leukokeratosis of the tongue and oral mucosa may be present, which sometimes leads to hoarseness if there is laryngeal involvement. These mucosal changes are not premalignant [1, 3].

Pachyonychia congenita results from mutations in the genes encoding keratins. Specific mutations in keratins 6 or 16 lead to the type I (Jadassohn-Lewandowsky) syndrome as described above. The type II (Jackson-Lawler) syndrome involves mutations in keratin 17 and clinically resembles the type I syndrome with the additional findings of both natal teeth and steatocystoma multiplex. Additionally, the type II syndrome displays less severe palmoplantar keratoderma with milder or absent oral lesions. Some authors describe a type III (Schafer-Branauer) syndrome that features all the findings of the type I disease associated with leukokeratosis of the corneas [1, 3]. Finally, a type IV syndrome, or pachyonychia congenita tarda, applies to a late-onset of the disease during the second or third decade of life [2, 4].

Treatment for the nail changes is difficult, with avulsion of the nails providing only temporary benefit. Destruction of the nail matrix may be ineffective. The most effective therapy is the application with occlusion of a keratolytic ointment to the nail plate, followed by the vigorous paring. Oral retinoids clear the keratotic papules and the leukokeratosis but may not relieve the palmoplantar keratoderma, which is best treated with topical keratolytics [1, 3].

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