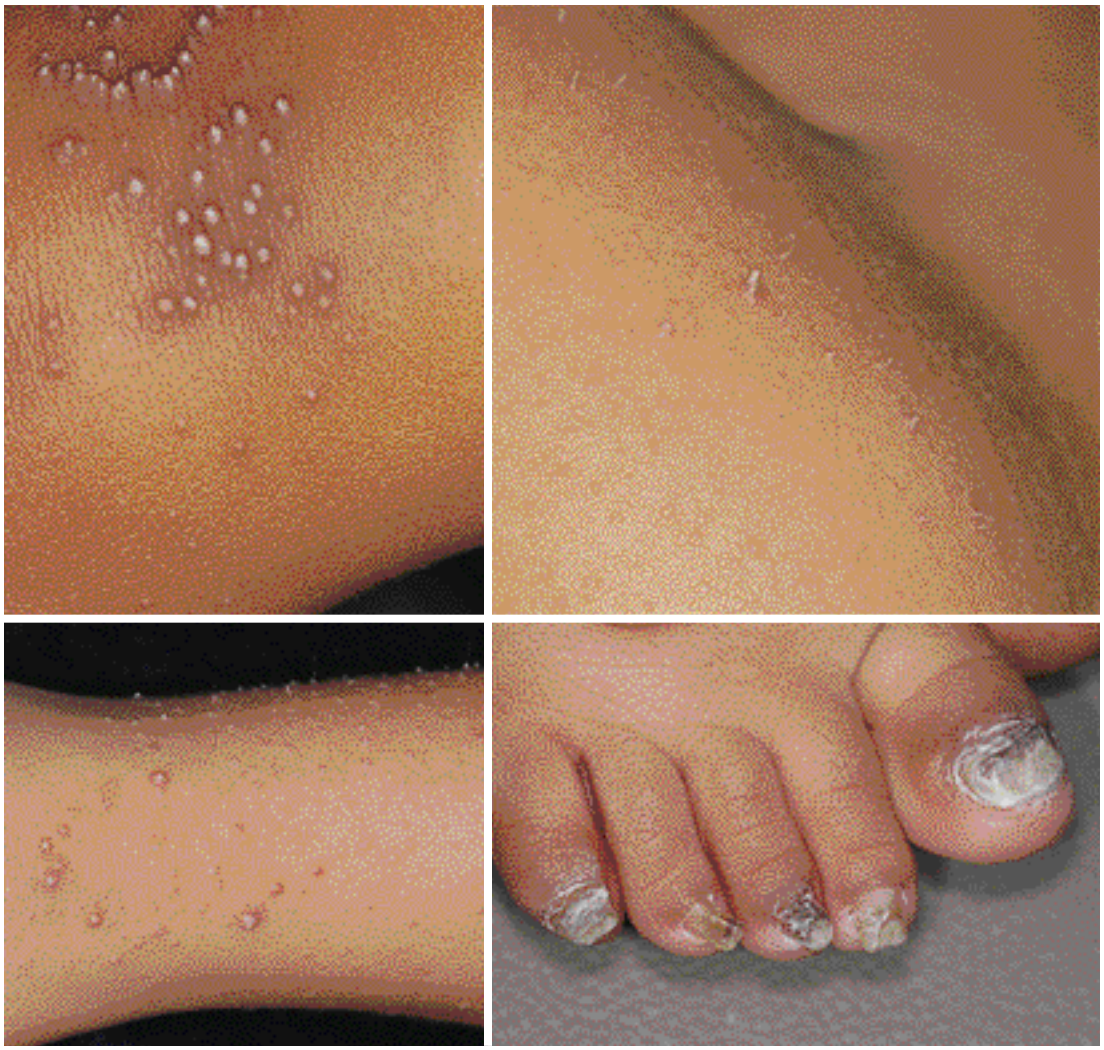
What Is Your Diagnosis?



A 9-year-old boy presented with dystrophic nails and follicular hyperkeratosis.

PLEASE TURN TO PAGE 143 FOR DISCUSSION

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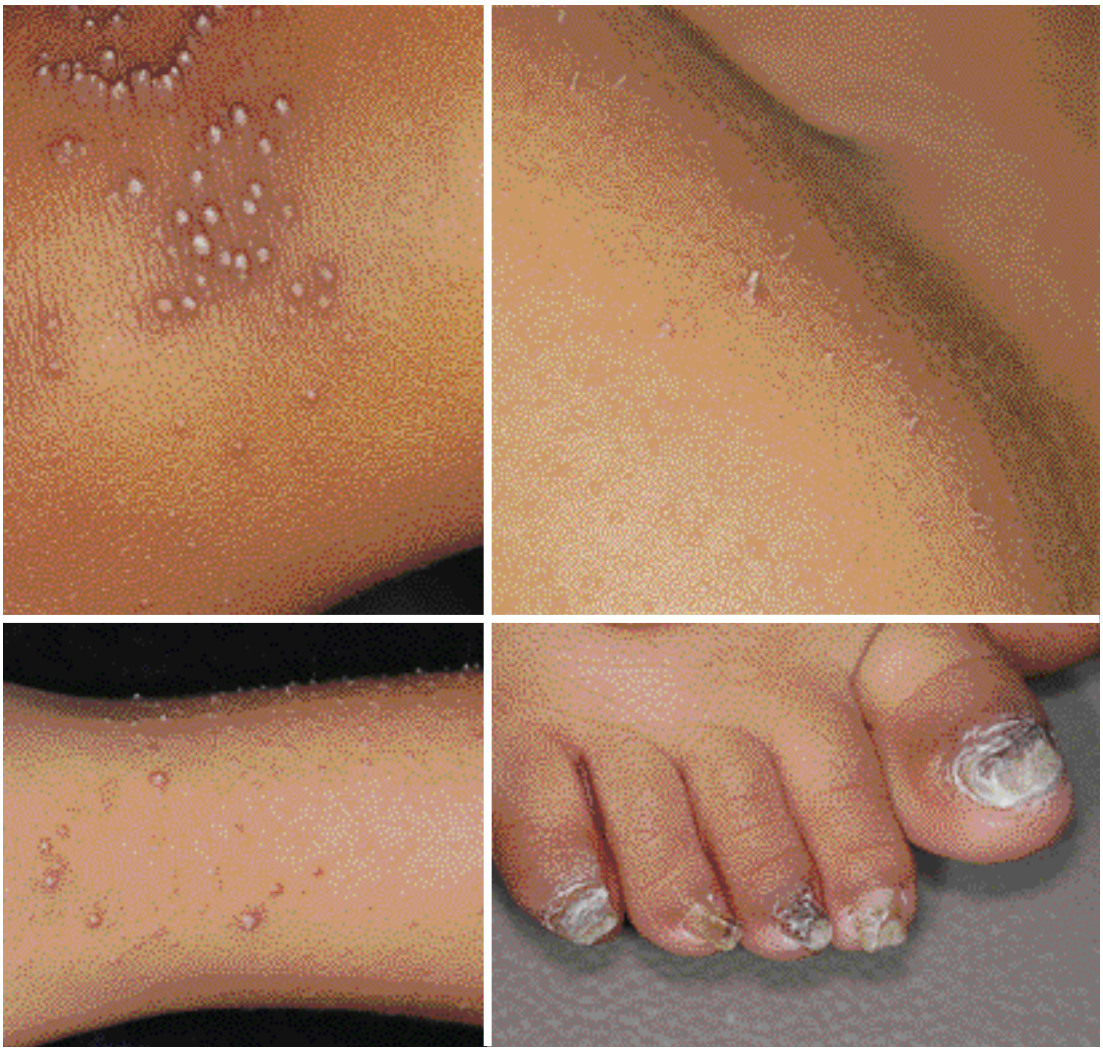
The Diagnosis: Pachyonychia Congenita



Pachyonychia congenita (PC) is an autosomal-dominant form of ectodermal dysplasia characterized by nail dystrophy, follicular keratotic spines, and focal palmoplantar keratoderma. Type I PC (PC1), also known as the Jadassohn-Lewandowski syndrome, is associated with mutations in the keratin 16 or keratin 6a genes.[1,2] Similar mutations also can cause focal palmoplantar keratoderma alone, without other manifestations of PC. PC1 is associated with oral lesions similar to white sponge nevus and, rarely, laryngeal leukokeratosis, which can lead to severe respiratory distress.[3] The oral lesions associated with PC do not undergo malignant degeneration, but squamous cell carcinoma has been reported in chronic plantar ulcerations of PC.[4] Patients with PC1 lack the epidermal cysts seen in type II PC (PC2).

PC2, also known as Jackson-Lawler syndrome, is associated with epidermal or pilosebaceous cysts, palmoplantar bullae, hyperhidrosis, natal teeth, twisted hair, and mutations in the keratin 17 gene. The cysts associated with PC2 predominantly affect the upper trunk and resemble steatocystoma multiplex or eruptive vellus hair cysts. Keratin 17 mutations also can manifest solely as steatocystoma multiplex with little or no nail dystrophy.[5]

PC2 also has been described with mutations in the keratin 6b gene.[6,7] All 3 PC keratins (6, 16, and 17) are abundantly expressed in the nail bed. Keratin 17 also is expressed in the nail matrix and in the hair follicle matrix of the eyebrows and other facial hair.[8]

Other forms of PC exist but are not as well characterized. Type III PC has been described as combining clinical findings of types I and II with angular cheilitis, corneal dyskeratosis, and cataracts. Type IV PC includes findings of types I through III plus laryngeal lesions, hoarseness, mental retardation, hair anomalies, and alopecia.[9] Some cases of PC involve the nails only. Rarely, signs of PC are delayed until adulthood—a phenomenon described as PC tarda.[10]

PC is difficult to treat. Keratolytics are of limited benefit, and nail matrix ablation is often of limited benefit because the disease principally affects the nail bed, not the matrix. However, Thomsen et al[11] have reported favorable results after nail matrix destruction. My own experience with nail ablation has been mixed; I have observed only partial responses, but patient satisfaction has been good. Acitretin has proved useful in the treatment of PC, including PC tarda.[12,13] Tendon calcification, demineralization, premature closure of the epiphyses, scoliosis, and limb length discrepancy are potentially devastating risks when children are treated with oral retinoids.[14]

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