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Pachyonychia Congenita in a Toddler

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Dear Sir:

We report management of a 3-year-old boy with genetically confirmed pachyonychia congenita (PC), a rare (prevalence, 1:5,000–10,000) autosomal dominant genetic disorder characterized by excessive keratinization of the glabrous skin of the hands and feet and nail thickening that can mimic onychomycosis.[1] Our patient was referred for treatment of painful, debilitating thickenings on the palms and selected nail beds of both hands and feet that has severely limited his ability to ambulate (Fig. 1). Prior temporizing measures, including sanding of the lesions, were ineffective in functional improvement.

PC is caused by autosomal dominant mutations in selected keratin genes, has 2 principle subtypes: PC type 1 (Jadassohn-Lewandowski syndrome, KRT6A, and KRT16 genes) and PC type 2 (Jackson-Lawler syndrome, KRT6B, and KRT17 genes).[2] Approximately 50% of PC cases are inherited. All subtypes of PC manifest with hypertrophic nail dystrophy, and focal, painful palmoplantar keratoderma, but other clinical symptoms can include palmoplantar blistering, oral leukokeratosis, follicular keratosis on the trunk and extremities, dystrophic toenails and fingernails, pilosebaceous cysts, palmoplantar hyperhidrosis, hoarseness, pili torti, and natal teeth.[2]

Treatment alternatives for painful hyperkeratoses have met with limited success due to short-term mitigation and high recurrence rates. Conservative measures include limiting friction and trauma to feet by minimizing walking or standing, though compliance in young children limits feasibility. Reducing hydration of the epidermis with wicking socks and ventilated footwear may help. Mechanical treatments such as filing, grinding, cutting, and clipping of affected nails are somewhat effective in reducing symptoms in mild cases.[1]

Reports of surgical management are sparse in the literature. Electrofulguration, deep curettage, and radical excision with autologous reconstruction have met with variable success due to recurrence and significant associated morbidity with more radical techniques.[3–5] The most common pharmacological management employs retinoid agents that may cause thinning of the epidermis with subsequent painful blistering and secondary infections.[1,2] Current medical research is examining the possible role of using mutation-specific small interfering RNA, rapamycin, anti-tumor necrosis factor biologics, and botulism toxins as treatment to reduce keratinization and patient symptoms.[1,2]

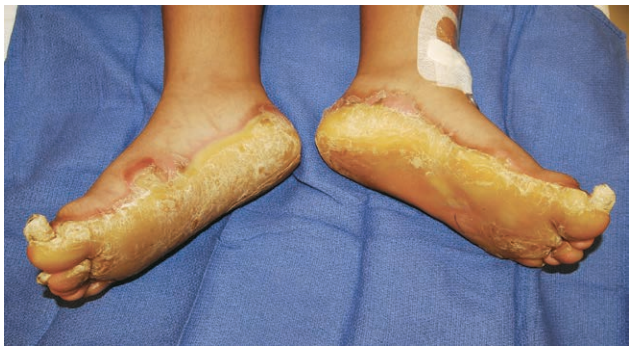

Fig. 1. PC is characterized by excessive keratinization of glabrous skin, often forming thick plaques on the plantar foot surfaces. These lesions can cause secondary functional disability with ambulation.

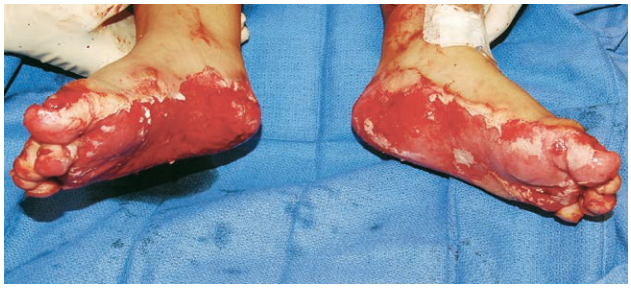

Fig. 2. Our approach in this case was to bluntly remove the plaques along a natural cleavage plane in the dermis, allowing significant symptomatic relief. This minimally invasive procedure may need to be repeated during the patient's lifetime according to regrowth of the lesions and symptomatic profile.

Our patient underwent blunt debridement of the affected skin and nails along a natural plane of dissection in the deep dermal layer (Fig. 2). This strategy was chosen by the parents, despite the risk of recurrence, to limit the morbidity of the procedure and shorten the recovery. The patient was discharged home with local wound care and healed uneventfully by 7 days. At 6-month follow-up, the toddler ambulates with minimal discomfort, and there is only minor thickening of the plantar and palmar skin.

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DISCLOSURE

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