

Chronic Foot Pain due to Pachyonychia Congenita in a Pediatric Patient: A Successful Management Strategy

Sarah Tariq, MD,* Michael L. Schmitz, MD,† and Megha Karkera Kanjia, MD‡

We report the case of an 11-year-old girl who presented to our multidisciplinary pain center with the chief complaint of chronic bilateral foot pain because of a rare congenital keratin disorder. This patient had been diagnosed with pachyonychia congenita, an extremely rare genetic disorder primarily affecting the skin and nails. The child had bilateral foot pain for years because of the characteristic blisters and calluses on the soles of her feet. Chronic pain was negatively impacting her quality of life; she was severely limited in her activities of daily living secondary to pain. Furthermore, she reported absenteeism from school, lack of social activities, and frequent nighttime awakenings. We discuss the successful management of her chronic foot pain using a multimodal, multidisciplinary approach. (A&A Case Reports. 2016;6:305–7.)

Pachyonychia congenita (PC) is an extremely rare genetic disorder afflicting only several thousand people worldwide.¹ Given the rarity of this disease, it is not possible to estimate its actual prevalence.² It is inherited in an autosomal-dominant pattern and affects the skin and nails. The following are the characteristic features^{3–6}:

- Hypertrophic nail dystrophy: fingernails and toenails are thickened and abnormally shaped;
- Palmoplantar keratoderma: very painful blisters and calluses on the soles of feet and, less commonly, on the palms of hands; and
- Palmoplantar hyperhidrosis: excessive sweating of the palms and soles.

In addition, affected individuals may have oral lesions, usually leukokeratotic plaques affecting the tongue and buccal surface. Other features of this disorder include the presence of pilosebaceous cysts and natal teeth.^{2–4}

The condition may present during early childhood; friction plays an important role, and once the child starts walking and weight-bearing, calluses and blisters occur on the soles of feet, making it very painful or even impossible to walk. Blistering is exacerbated by excessive sweating of the soles. Written consent was obtained from the patient's mother for publication of this report.

CASE DESCRIPTION

An 11-year-old girl presented to our multidisciplinary pain clinic with a longstanding history of bilateral foot pain. This pediatric patient was diagnosed with PC; she had blisters

and calluses on the soles of her feet that caused painful walking and weight bearing. She reported recalling experiencing foot pain her entire life. She described her pain as vacillating; it varied from being sharp and throbbing to shooting, accompanied by tingling as well as pins-and-needles sensations. On average, she rated her pain as 7 to 8/10 in intensity on a visual analog scale (VAS). Her pain was exacerbated by walking, weight-bearing, and wearing shoes. She could only wear open shoes or flip flops even in very cold weather because friction from wearing closed shoes significantly aggravated her pain. Her pain was somewhat relieved by pain medications and warm soaks. She had taken opioid pain medication for years. She initially had been prescribed 5 mg hydrocodone every 4 to 6 hours as needed for years, but for the previous year, she had been prescribed 5 mg oxycodone as needed. Her daily oxycodone requirement averaged 25 to 30 mg. She was also taking over-the-counter analgesics, including acetaminophen and ibuprofen; however, her pain was poorly controlled.

The child reported significant pain-related disability. She described limited ability to walk, inability to participate in age-typical activities such as sports, and absenteeism from school, all because of pain. She was socially withdrawn from her peers, and her school attendance had only been 60% that particular semester. She was an average student but perceived her pain to be a barrier to her academic success. Furthermore, she reported poor nocturnal sleep quality and nighttime awakenings 3 to 4 nights per week secondary to pain. She also reported multiple visits to the emergency department because of intractable foot pain.

A psychological evaluation revealed poor self-esteem and depressed mood. She had multiple sources of stress, including having changed schools because of teasing by peers. Her biologic parents were divorced, and she had a strained relationship with her biologic father.

On physical examination, she was a female of normal build; she was in the 60th percentile for height and 50th percentile for weight. Pertinent findings on examination included several plantar blisters and calluses (Fig. 1) and tenderness even to light touch. Her fingernails and toenails were thickened and abnormally shaped (Figs. 2 and 3). The findings of her neurologic examination were normal; she had full strength in all extremities, and there were no focal deficits. There was an absence of erythema, color changes, and temperature changes.

From the *Department of Anesthesiology and Pain Medicine, UT Houston, Texas Medical Center, Houston, Texas; †Arkansas Children's Hospital, Little Rock, Arkansas; and ‡Texas Children's Hospital, Houston, Texas.

Megha Karkera Kanjia, MD, is currently affiliated with the Cincinnati Children's Hospital, Cincinnati, Ohio.

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Address correspondence to Sarah Tariq, MD, Department of Anesthesiology, Arkansas Children's Hospital, 1 Children's Way, Little Rock, AR 72202. Address e-mail to stariq@uams.edu.

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Figure 1. The plantar aspect of our patient's foot showing calluses.



Figure 3. The patient's foot showing abnormal toenails.

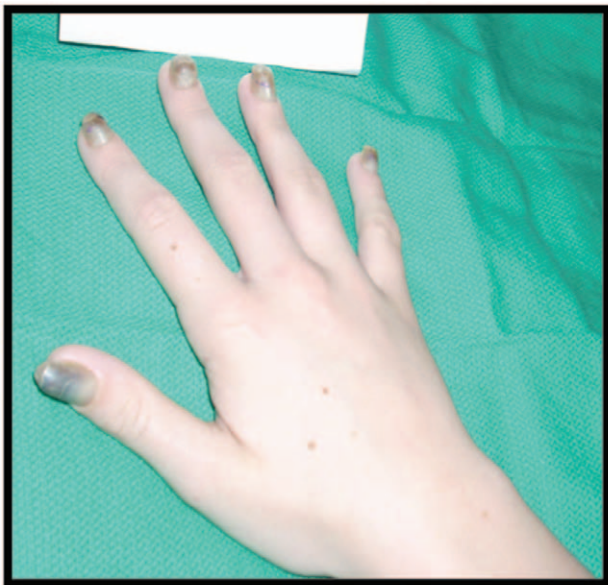


Figure 2. The patient's hand showing abnormal nails.

A multimodal, multidisciplinary approach was adopted in the management of this patient's pain. She was evaluated by the pain physician, psychologist, and physical therapist during the initial visit, and a comprehensive treatment plan was devised based on the input from all providers. The treatment plan was discussed with the patient and her parent. The goals of treatment were provision of analgesia and return to normal functional status. We will discuss the treatment modalities individually.

- **Pharmacotherapy:** We started gabapentin based on her description of pain with neuropathic characteristics. Gabapentin was started at a low dose, and the dose was escalated gradually. She continued receiving oxycodone on an as-needed basis, with the eventual goal of weaning from oxycodone.

- **Intervention:** After extensive research for treatment options for this condition, plantar injections of botulinum toxin were discussed with the patient and her mother. Cutaneous botulinum toxin injection inhibits sweating, thereby decreasing resultant blistering. Both the patient and the parent agreed to proceed with this treatment, and we performed these injections 4 weeks after her initial visit.
- **Physical therapy:** We enrolled the child in physical therapy to improve the range of motion for her feet.
- **Psychologic interventions:** We recommended counseling. The psychologist, patient, and parent discussed cognitive behavioral therapy with a focus on coping with pain mechanisms, deep breathing, and relaxation techniques.
- **Physical medicine and rehabilitation:** Orthotics, special shoes, and insoles were created to evenly distribute her weight and minimize friction. A wheelchair was provided to be used only for extremely painful episodes associated with severe plantar blistering so that healing could occur during a temporary non-weight-bearing interval. Previous medical reports indicate that bed rest or non-weight-bearing promotes healing.² It was also felt that having a wheelchair would improve her school attendance.
- **Four-week follow-up:** Our patient reported pain scores of 4 to 5/10 on VAS 2 weeks after the initiation of gabapentin. She also reported improved sleep quality and reduced opioid consumption (5–10 mg oxycodone/d compared with 25–30 mg/d previously). She received plantar injections of botulinum toxin type A to both feet. The procedure was performed under general anesthesia with midazolam, fentanyl, and propofol. One hundred units of botulinum toxin type A was injected intradermally, 50U per foot after identifying the areas of maximal sweating. Hyperhidrotic areas were detected by an iodine starch test.

- Six-month follow-up: The child reported minimal pain 1 to 2/10 on VAS and significantly improved functional status as indicated by her ability to walk and her school attendance. She had not required any opioid medication in the past 4 months and did not report any nighttime awakenings.

DISCUSSION

PC is an extremely rare genetic disorder that is inherited in an autosomal-dominant pattern.³⁻⁶ It is included in the list of rare diseases maintained by the Office of Rare Diseases of the National Institutes of Health.⁷ Affected individuals can experience incapacitating chronic pain, significant inability to function socially, and an overall poor quality of life.^{2,3} Because this disorder is so rare, there is a paucity of data regarding effective management strategies for affected individuals. A review of the current literature reveals that although management strategies aim to control manifestations of the disease, there is no curative treatment available.⁸ Chronic pain secondary to plantar blistering and hyperhidrosis is often the most debilitating aspect of the disease.^{2,3,5} Unfortunately, little clinical information is published about effective strategies to manage this debilitating pain. To our knowledge, this is the first case report that describes a successful pain management strategy for a pediatric patient with foot pain because of PC.

Given that our patient experienced symptoms associated with neuropathic pain (pins-and-needles sensations and shooting pains) and that she responded well to gabapentin therapy, we believe that there is a neuropathic component to the pain associated with PC. One possible explanation is that the presence of chronic pain results in central sensitization, evoking an exaggerated pain response. Also, mechanical insult because of chronic blister formation and keratoderma may cause trauma to nerve endings making them more susceptible to injury.

The only report of successful pain management of adult patients with PC was found in the *British Journal of Dermatology*.^{9,10} A study at a Swedish center used plantar

injections of botulinum toxin for patients with PC and epidermolysis bullosa on the premise that botulinum toxin can counteract the hyperhidrosis associated with these conditions, resulting in decreased blistering and pain.

Our patient responded very well to a combination of different modalities. In conclusion, gabapentin and plantar injections of botulinum toxin combined with psychological counseling, cognitive behavioral therapy, and physical therapy were all useful in alleviating chronic foot pain in our patient with a complex medical diagnosis. ■■

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