Pachydermodactyly in an adolescent male

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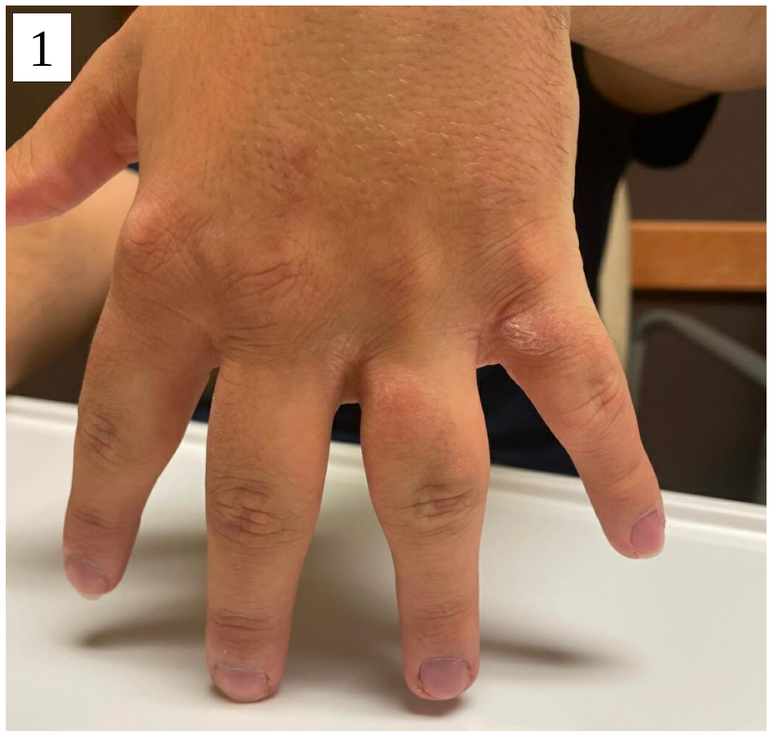
Introduction

Pachydermodactly is a rare, benign tissue swelling on the medial and/or lateral aspects of the proximal interphalangeal joints that predominantly affects adolescent males. We present the case of a 19-year-old male who presented with painless, non-pruritic, bilateral soft tissue swelling of the fingers. We discuss the clinical presentation, differential diagnosis, and management options for this rare condition.

Case

An 18-year-old male presented to outpatient orthopedics with asymptomatic, slowly progressive swelling of his fingers over the course of two weeks. Radiographs were taken and unremarkable, prompting a referral from orthopedic surgery to dermatology. Examination revealed marked fusiform swelling of the proximal phalanges, sparing the thumb and interphalangeal joints. There was no edema, and the overlying skin appeared normal. There was no fever or leukocytosis. Serum protein electrophoresis was negative for monoclonal gammopathy, and thyroid stimulating hormone levels were normal, which ruled down rheumatologic or thyroid disease. Punch biopsy showed mucin deposition and was otherwise normal. The patient's mother suspected he may have had an autism spectrum disorder and noted that he can be fidgety and often cracked his knuckles and joints. At age 12, the patient was evaluated for autism and did not meet the diagnostic criteria. His past medical history included anxiety and a mood disorder, treated with aripiprazole daily and hydroxyzine as needed. There was no improvement reported after the dermatology visit.



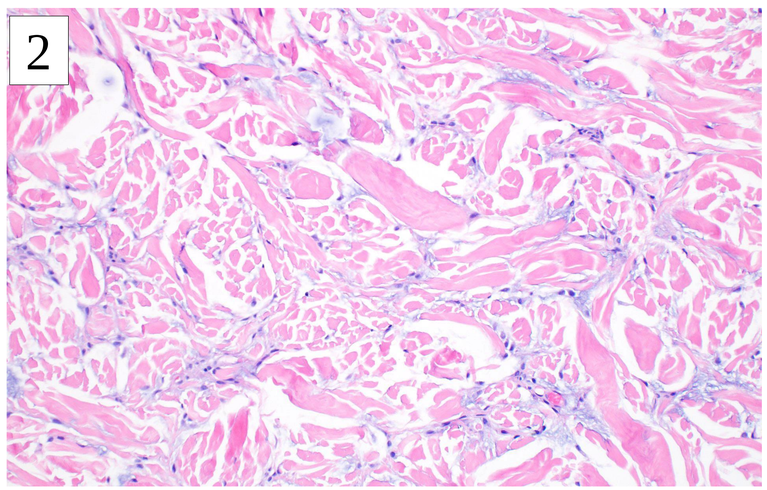


Figure 1: Patient's left hand with notable fusiform swelling of the proximal phalanges.
Figure 2: Punch biopsy showing mucin deposition. 20x magnification.

Discussion

While the etiology of pachydermodactyly is unknown, Dallos et al. found that 43% of cases reported were associated with repetitive mechanical behaviors of the digits such as repeated stretching, rubbing, or cracking of the interphalangeal joints (1). Previous case reports have also associated pachydermodactyly with autism spectrum disorders (3). In our case, the patient's mother reported that he had a history of repetitive cracking of his knuckles and joints, which may have contributed to the development of pachydermodactyly. The patient also had a history of anxiety and mood disorder, treated with aripiprazole daily and hydroxyzine as needed. A 2022 case report described an adolescent male with an anxious personality and "intense knuckle-cracking habit" who had a five-year history of painless periarticular swelling at the proximal interphalangeal joints that was diagnosed as pachydermodactyly (4). A 2010 case report detailed an adolescent female with pachydermodactyly who had a history of trichotillomania and attention deficit hyperactivity disorder (5). The differential diagnosis for pachydermodactyly includes other conditions that cause soft tissue swelling of the fingers, such as scleroderma, scleredema, and reactive mucinosis. In our case, there was no laboratory evidence of paraproteinemia or diabetes mellitus, and dermatopathology revealed dermal mucin deposition without fibrosis to establish the diagnosis of pachydermodactyly. Pachydermodactyly is not the only dermatologic disease associated with repetitive behaviors. Trichotillomania is an irresistible compulsion to pull out one's hair. This can result in hair loss, most commonly involving the scalp, eyebrows, or eyelashes (6). Excoriation disorder, also called dermatillomania, is the recurrent picking of skin, which leads to erosive skin lesions most commonly on the face followed by the hands, fingers, arms, and legs (7). Habitual picking of the cuticle is associated with habit tic deformity, a finding of median ridging of the fingernail, most often the thumb (8). Onychophagia is compulsive biting of the nails and can lead to infection and dental problems in addition to nail damage (9). Excoriation disorder can be treated with N-acetylcysteine, but pharmacological treatments for other body-focused repetitive behaviors have not been described. Management of pachydermodactyly is typically focused on symptom relief. In our case, the patient did not report any improvement after the dermatology visit. However, it is important to reassure patients that pachydermodactyly is a benign condition and does not pose any serious health risks. Patients should be advised to avoid repetitive mechanical trauma to the affected fingers and to seek medical attention if the swelling becomes painful or significantly affects their daily activities.

Conclusion

In conclusion, pachydermodactyly is a rare, benign condition that predominantly affects adolescent males. The clinical presentation is often nonspecific, and a skin biopsy is necessary to establish the diagnosis. The etiology of pachydermodactyly remains unclear, although repetitive mechanical trauma may play a role in its development. There is no specific treatment for pachydermodactyly, and management is typically focused on symptom relief. Further investigation may be warranted to explore potential underlying mechanisms and treatments, especially in patients with comorbid psychiatric disorders.

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