An Unusual Presentation of Pachydermodactyly: A Case Presentation and Discussion

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Abstract

Pachydermodactyly (PDD) is an infrequently diagnosed, benign joint swelling of the proximal interphalangeal joints. It is most frequently found in males and is associated with a variety of disorders and syndromes, including psychiatric and congenital conditions. The majority of cases are associated with repetitive mechanical trauma to the hands. Many other disorders mimic PDD, leading to extensive testing. We report a case of proximal interphalangeal joint swelling in a young adult male with a history of congenital heart disease.

Introduction

Pachydermodactyly (PDD) is a benign condition typified by skin thickening in the joint spaces of the hands, particularly the proximal interphalangeal (PIP) joints. The term comes from the Greek for “thick-skin-finger,” PDD was first described by Bazex et al. in 1973 as “pachidermie digitale des premières phalanges,” and the term “pachydermodactyly” was coined by Verbow in 1975. It is frequently found in asymptomatic, healthy, adolescent males and has some associations with hereditary disorders and repetitive trauma. On histopathology, the findings include increased accumulation of irregular bundles of thickened collagen in the dermis, typically with no other distinguishing characteristics. Few cases have been reported in the literature. We present a case of PDD in an 18-year-old male with a history of congenital heart disease, as well as a review of this disease.

Case Report

An 18-year-old male presented with continuing complaints of progressive, painless swelling of the proximal interphalangeal joints of both hands. He was referred by a pediatric rheumatologist for evaluation of a possible connective-tissue disorder. His mother reported the swelling of his knuckles has worsened since childhood. The patient described his knuckles as slightly pink and swollen, with no nodularity. Symptoms did not improve or worsen with activity. He denied any pruritus, fatigue, fever, joint pain, easy bleeding or bruising. Furthermore, the patient reported no rashes, arthralgias, fever, or gastrointestinal symptoms. The patient denied any recent trauma or repetitive-motion activities. Medical history was significant for asthma, allergic rhinitis, hypertension of unknown etiology, and congenital heart disease. His surgical history included repair for coarctation of the aorta and bicuspid aortic valve, for which he was followed by a cardiologist. Birth history was significant for his being one of triplets. The remainder of the exam was benign.

Testing
Radiographs of the hands ordered by the rheumatologist showed no bony changes. Complete blood count, liver function tests, C-reactive protein, erythrocyte sedimentation rate, rheumatoid arthritis panel, antinuclear antibody testing, and serum ferritin were within normal limits. Labs out of normal range included angiotensin-converting enzyme, which was slightly elevated at 87 U/L (reference range 14-82 U/L).

A 4-mm punch biopsy of the proximal interphalangeal joint of the right ring finger was obtained. Histopathological sections demonstrated slight epidermal acanthosis with thickening of the dermis. Within the dermis there were thick collagen bundles ordered in a haphazard arrangement, consistent with pachydermodactyly (Figure 2).

Differential Diagnosis
Multiple etiologies for this patient’s condition were considered. The most benign cause considered was knuckle pads (KP), which can be primary (idiopathic) or secondary. The differential diagnosis also included juvenile idiopathic arthritis, psoriatic arthritis, rheumatoid arthritis, gouty tophus, juvenile hyaline fibromatosis, and subcutaneous granuloma annulare.

Diagnosis and Treatment
The histopathology and clinical history led to the diagnosis of pachydermodactyly. The patient was reassured that this is a benign disorder and was instructed to avoid excess manipulation of the hands. Because of his congenital medical conditions, genetic testing for possible syndromic presentation of a larger genetic condition was recommended. After referral to a geneticist, our patient elected to postpone further testing due to logistical constraints.

Discussion
Pachydermodactyly is a rare, benign disorder of the skin characterized by swelling of the soft tissue at the proximal interphalangeal joints, particularly in young males. Most commonly, the second, third and fourth digits are affected, sparing the thumb. Occasionally, the disease may involve other extremities or extension into the proximal hand, which is further described as pachydermodactyly transgressens. Originally, this disorder was described as a variant of knuckle pads; however, unlike knuckle pads, PDD is associated with ulnar and radial expansion of the soft tissue without dorsal growth. The typical manifestation presents most similarly to juvenile idiopathic arthritis, psoriatic arthritis, and rheumatoid arthritis. Because of this, many patients, such as the one in this case, undergo numerous lab and imaging studies to evaluate and treat possibly disfiguring disorders. The lack of pain, tenderness, or functional limitations despite swelling of the joints in PDD helps differentiate this diagnosis from more serious possibilities, such as juvenile idiopathic arthritis or rheumatoid arthritis.

PDD has been associated with various disorders in the past but is frequently idiopathic. Familial PDD has been reported, but it is uncommon. It has also been suggested as another cutaneous finding in tuberous sclerosis and Ehlers-Danlos. PDD is associated with repetitive mechanical friction, including repetitive-motion activities, along with manual labor and psychiatric disorders. Tic-like behaviors, obsessive-compulsive disorder, autism, and playing video games have all been linked to PDD. These associations with PDD are believed to be a result of trauma, such as knuckle-cracking, frequent interlocking of the fingers, or squeezing of the joints.

The diagnosis of PDD is based on clinical picture, laboratory findings, and imaging. Laboratory findings are consistent with a non-inflammatory or non-immunological response. Imaging reveals soft-tissue swelling with possible increased vascularity but without any injury to bone or joint spaces.
Histologically, PDD is defined by non-specific changes like epidermal hyperkeratosis and increased collagen deposition in a haphazard arrangement. There is typically an increased amount of type III and V collagen compared to normal skin.8,15

Because of its benign nature, treatment is symptomatic. For many patients, discontinuing repetitive-motion or trauma-inducing activities is first-line treatment. Controlling mechanical trauma may resolve or reverse the appearance of swelling.8 For those patients with tics or compulsive hand movements, treatment focuses on the underlying psychiatric disorder. For those wishing to reduce the cosmetic appearance of PDD, options include topical and intralesional steroids or surgical intervention.6,8

**Conclusion**

The case presented herein is an example of pachydermodactyly in a patient with a history of congenital heart disease. Our patient did not present with historical findings of repetitive mechanical trauma or any psychiatric disorders, which prompted further testing. This may indicate that PDD could be part of a larger genetic syndrome in some cases and could lead to a better understanding of associations between PDD and other disorders. Because of the presentation of this disorder with other multisystem concerns, this patient was encouraged to seek further testing. In the future, similar reported cases could potentially strengthen a syndromic association.

**References**


