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Pachydermodactyly: A Rare Type of Macrodactyly as a Dermatological Sign of Compulsive Behavior and Repetitive Minor Trauma; a Case Report and Review of the Literature

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Abstract

Introduction: Pachydermodactyly is a very rare type of macrodactyly. It clinically resembles juvenile rheumatoid arthritis, but it has a non-inflammatory etiology without bone, articular, or synovial involvement.
Case Presentation: In this study, we report the case of a 15-year-old boy with repetitive behavior diagnosed with a pachydermodactyly after evaluation of bilateral swelling of the proximal interphalangeal joints. Histologic examination revealed epidermal hyperorthokeratosis, thickening of the dermis with increased dermal collagen and absence of inflammatory cells. Pachydermodactyly is a very rare condition associated with compulsive behavior and results from repetitive minor trauma that causes localized proliferation of fibrous tissue.
Conclusions: Pachydermodactyly is a dermatological sign of compulsive behavior and repetitive minor trauma, and it should be distinguished from other forms of joint enlargement to avoid unnecessary medicamentous treatment. Control examinations did not reveal clinically relevant worsening even without any treatment applied.

Keywords: Behavior, Compulsive, Fibromatosis, Finger, Joint, Local, Macrodactyly, Pachydermodactyly, Repetitive, Trauma

1. Introduction

Pachy-dermo-dactyly (PDD), Greek for thick-skin-finger, is a rare and benign condition where less than 125 cases have been described. PDD represents an asymptomatic bilaterally symmetric enlargement of the proximal interphalangeal (PIP) joints of mainly the second, third, and fourth fingers, due to localized fibromatosis (1). It clinically resembles juvenile rheumatoid arthritis, but PDD has a non-inflammatory etiology without bone, articular, or synovial involvement (2). We, hereby, present the case of a young male with bilateral swelling of the proximal interphalangeal joints which was diagnosed as a very rare type of macrodactyly-pachydermodactyly.

2. Case Presentation

A 15-year-old boy with a two-year history of fusiform dorsolateral (ulnar-radial) soft-tissue enlargement of the proximal interphalangeal joints II-IV of both the hands came to the hospital (Figure 1A). He had mild mental retardation and a habit of repetitively manipulating his hands when he felt emotional distress and anxiety. The patient denied pain on motion or tenderness to palpation. Skin color was unchanged with normal skin temperature; joints had a normal function; and there was no morning stiffness, fever, and rash. Local lymph nodes were not enlarged, and there were no additional abnormalities of hands and feet. Prenatal and family history were unremarkable. His karyotype was 46, XY and congenital anomalies were absent.

The boy was diagnosed with pachydermodactyly (PDD) after clinical evaluation, laboratory analyses, ultrasonography, radiological study, and histopathologic examination. Laboratory findings were unremarkable. Sedimentation rate, C-reactive protein, fibrinogen, complete blood count, liver, and renal function tests, TSH, T3, T4, IgA, IgG, IgM, C3, C4, were within the normal ranges. Urine analysis was also normal. Rheumatoid factor and antinuclear antibody, aCLA (anticardiolipin antibody), anti-CCP (cyclic citrullinated peptide), ANCA (anti-neutrophil cytoplasmatic antibody), beta2glycoprotein, ASTO (Anti Streptolysin O), and human leukocyte antigen B27 (HLA B27) were negative. Abdominal ultrasound was normal. Ultrasonography findings showed dermal swelling without bone, articular or synovial involvement, or joint effusions. Plain radiography of the hands revealed soft tissue swelling without the following: bones and joints involvement, erosion of bones, joint space narrowing, or any other structural abnormalities (Figure 1B). MRI showed dermal edema without synovial inflammation and no erosion of the phalangeal bone or any other changes in bone and joints (Figure 1C). Histopathologic analysis revealed epidermal hyperorthokeratosis, thickening of the dermis with increased dermal collagen, and no inflammatory cells (Figure 2A - 2C).

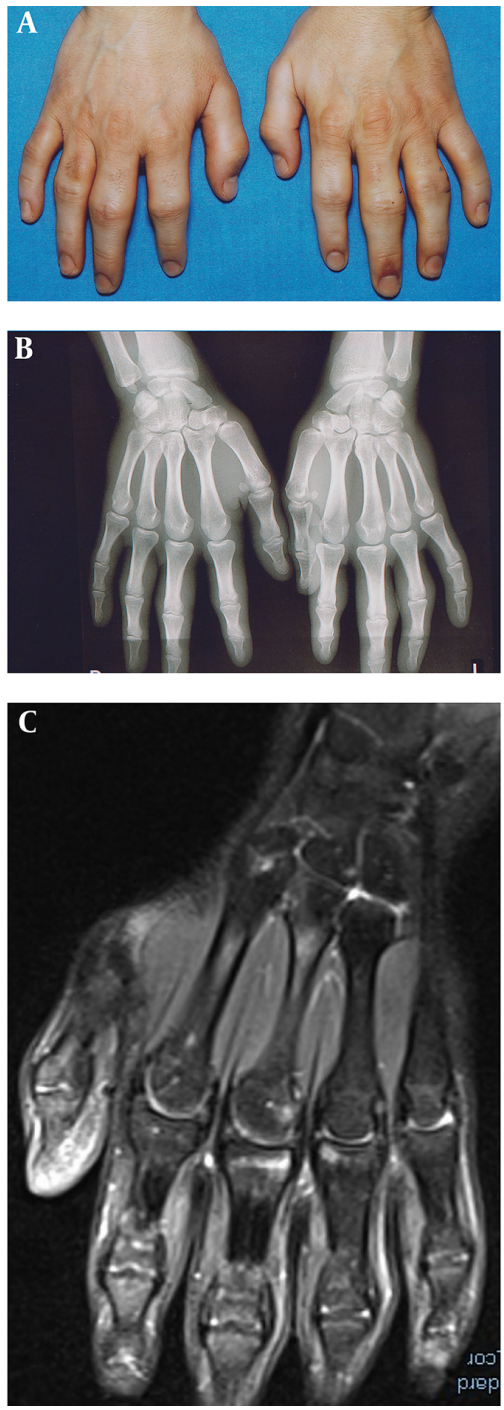

Figure 1. Enlargement of the proximal interphalangeal joints II-IV of the both hand. B, RTG showed soft tissue swelling without bones and joints involved. C, MRI showed dermal edema without synovial inflammation and erosion of the phalangeal bones.

3. Discussion

PDD is usually presented on both hands, but it could be unilateral, asymmetric, and present on feet. Young males are predominantly affected with a male to female ratio of 3.9:1. Periarticular thickening usually starts in puberty, progresses for several years, and stabilizes in adolescence. The etiology of PDD remains uncertain, and it has been suggested that it results from repeated, persistent minor trauma of the joints. This could be due to repetitive behavior and phalangeal manipulation during periods of emotional distress, anxiety, or obsessive-compulsive repetitive activity associated with using a computer and video games (1). Bardazzi et al. made a classification that recognizes five types of PPD: classic bilateral PDD (related to repeated microtrauma on more than one joint), localized PDD (one joint is affected), transgrediens PDD (extends to the dorsum of the hand and spreads to metacarpophalangeal region), familial (affects more than one person in family), and PDD associated with tuberous sclerosis (2). The case described here belongs to the classic form. The clinical significance of this classification has not yet been evaluated (3). PDD associated with tuberous sclerosis differs from four other forms of PDD. Tuberous sclerosis is a particular entity, and besides dermal and subcutaneous tissue,

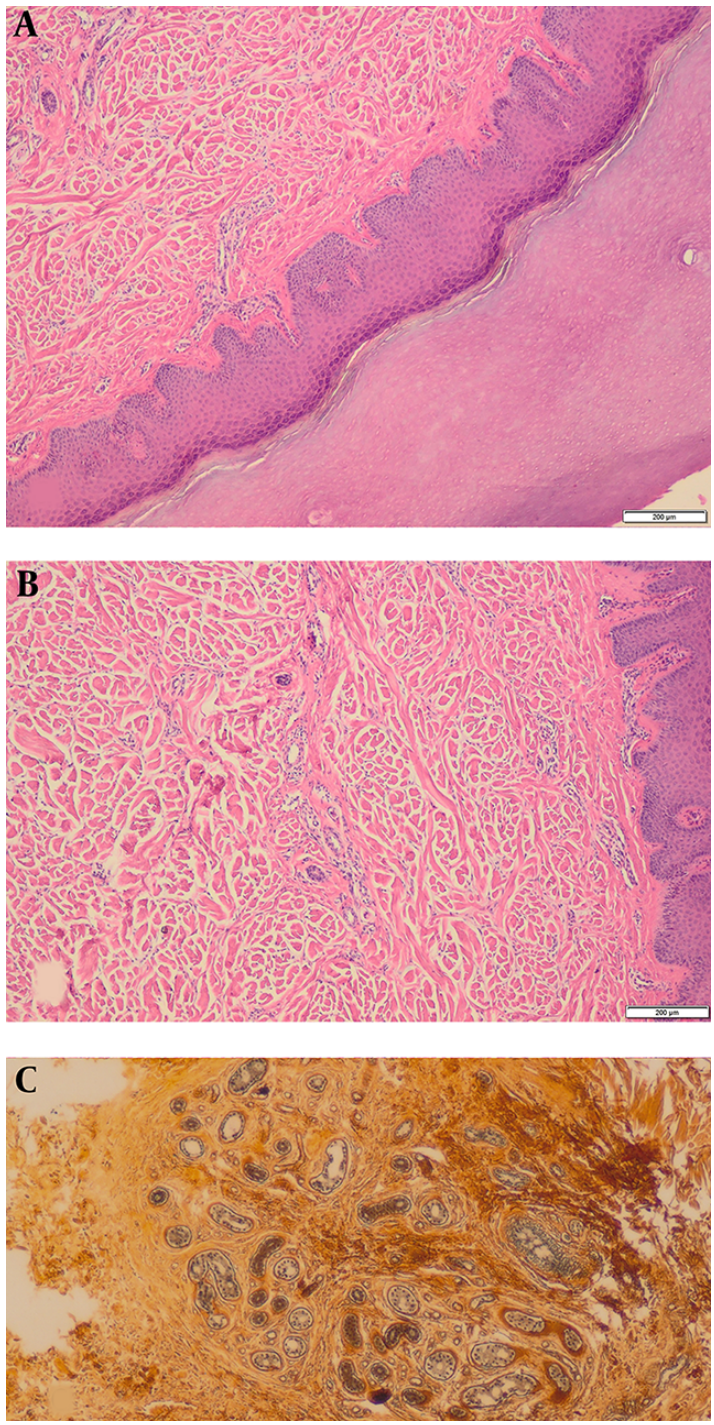

Figure 2. Epidermal hyperorthokeratosis. B, thickening of the dermis with increased dermal collagen, and no inflammatory cells. C, sweat glands surrounded with mild concentrically thickened collagen fibers.

metacarpal bones and phalanges can also be involved with irregular periosteal thickening, an increase in width, limited flexion, and painful response. It is not truly PDD but macrodactyly. PDD is very often mistaken for an inflamma-

tory disease and treated with immunosuppressive therapy (4). PDD must be differentiated from rheumatoid arthritis, primarily juvenile idiopathic arthritis, secondary pachydermoperiostosis, knuckle pads, pseudo knuckle pads, and progressive nodular skin fibrosis. The differential diagnosis should also include certain hereditary disorders (Ehlers-Danlos syndrome, tuberous sclerosis, infantile systemic hyalinosis, and neurofibromatosis), metabolic disorders (Mucopolysaccharidoses, and Farber disease), and overgrowth syndromes (Klippel-TrEnaunay, macro dystrophy lipomatosis, Proteus syndrome, multiple exostoses syndrome, progressive macrodactyly, digital gigantism, and hyperostotic macrodactyly).

An overview of pachydermodactyly case reports regarding locations and presentations is presented in Table 1.

An appropriate clinical and histologic evaluation is essential to differentiate this condition from other disorders and avoid inappropriate treatment. Psychological support and suspension of repetitive behavior, repetitive minor trauma, and irritation of the joints are among the suggested therapies. Suspension of repetitive trauma should result in spontaneous regression of periarticular swelling. Also, intralesional corticosteroid injection with triamcinolone acetonide can reduce periarticular swelling (14). Due to difficulties in social interactions with the boy, repeated movements could not be avoided. Also, parents rejected any psychological support or medicamentous therapy for the boy, but all control examinations during the next five years did not reveal clinically relevant worsening of symptoms even without any treatment.

3.1. Conclusion

In cases of macrodactyly, it is necessary to exclude other possible causes of the presenting joint enlargement, especially since differential diagnosis includes beside juvenile rheumatoid arthritis, rare metabolic and potentially treatable diseases such as Mucopolysaccharidoses and Farber disease. Early and correct diagnosis of PDD will allow many patients affected by this disease and their parents to reduce anxiety and concern for other possible causes of macrodactyly.

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Table 1. Location, Age, Gender, Causes and Presentations of Pachydermodactyly Case Reports

Author	Location	Age	Gender	Cause	Presentation
Nicolay et al. (Germany/2015) (4)	Unilateral pachydermodactyly transgrediens (left hand)	14-year-old	Male	Specific localized mechanical manipulation of the hands and rubbing behavior; worked as a farmer	Indolent indurate swelling of the third to fifth fingers and the lateral and hypothenar areas of the left hand
Schneider et al. (United States of America/2017) (5)	Bilateral pachydermodactyly on upper and lower extremities	15-year-old	Male	Chronic mechanical stimulation	Bilateral swelling and thickened of the joints of hands and feet
Rasi et al. (Iran/2017) (6)	Unilateral (left hand)	27-year-old	Male	Computer workaholic	Fusiform swellings, limited to the medial and lateral sides of the proximal interphalangeal joints of the second through to the fifth fingers of the left hand, extended to the metacarpophalangeal joints of the second and third fingers of the same hand
Saka et al. (Turkey/2005) (7)	Bilateral	Two patients: 18-year-old and her 14-year-old sister	Females	Familial pachydermodactyly	Bilateral swellings of the distal interphalangeal joints of the right index and little fingers, and left middle and little fingers, with similar swellings around the proximal interphalangeal joints of both little fingers. Her sister had swellings around the distal interphalangeal joints of her left index and middle fingers, and her right index finger
Lo and Wong (China/1993) (8)	Localized pachydermodactyly associated with tuberous sclerosis	5-year-old	Male	Tuberous sclerosis	Localized congenital form of pachydermodactyly since birth may represent an additional cutaneous sign of tuberous sclerosis
Vale et al. (Brazil/2009) (9)	Transgrediens pachydermodactyly	25-year-old	Male	There was no repetitive stimuli of the fingers, even though the patient had intellectual deficit	Nodules of fibroelastic consistency located on the lateral aspect of the right second metacarpophalangeal joint, medial aspect of left first metacarpophalangeal joint, back of bilateral first toe phalanges and calcaneus, with a thickened skin around
Tolis et al. (Greece/2016) (10)	Bilateral	19-year-old	Male	Intense stress and “over-rubbing” of hands	Combined proximal and distal interphalangeal joint involvement

Rachowska et al. (Poland/2010) (11)	Bilateral	21-year-old	Male	Tai chi exercises	Symmetrical diffuse swelling around the digits of the hands
Small et al. (United Kingdom/2011) (12)	Unilateral (left hand)	12-year-old	Male	A lot of time playing on games consoles and typing on keyboards	Diffuse thickening of the soft tissues around the proximal interphalangeal joints of the left middle and ring finger. Swelling of the proximal interphalangeal joints of only the left hand in a right-handed young male
Seo and Sung (Korea/2011) (13)	Bilateral	14-year-old	Male	Manipulated his hand repeatedly	Bilateral symmetric soft-tissue swelling on the proximal interphalangeal joint of the second through fourth fingers with thickening of the skin of both hands since the age of 4 years
Present case, Kavecan et al. (Serbia/2018)	Bilateral	15-year-old	Male	Mild mental retardation and a habit of repetitively manipulating his hands when he felt emotional distress and anxiety	Bilateral swelling of the proximal interphalangeal joints of both hands