

A Rare Case in Rheumatology Clinical Practice: Pachydermodactyly

Romatoloji Pratiğinde Nadir Bir Olgu: Pakidermodaktili

İD Ayşe Ünal Enginar¹, İD Ali Nail Demir¹, İD İlhan Sezer¹, İD Can Çevikol²

¹Akdeniz University Medicine School, Department of Physical Therapy and Rehabilitation, Rheumatology, Antalya, Turkey

²Akdeniz University Medicine School, Department of Radiology, Antalya, Turkey

ABSTRACT

Pachydermodactyly (PDD) is a benign acquired digital fibromatosis characterized by asymptomatic, progressive swelling of periarticular soft tissue, which is usually seen in young males. The etiology of PDD is not fully understood, but is thought to be probably a result of recurrent mechanical stimulation. PDD is usually diagnosed by clinical evaluation. There are thick collagen bundles and collection of dermal mucin in the histopathology. Treatment is not indicated because of benign prognosis. Rheumatological diseases, particularly rheumatoid arthritis affecting the joints of the hands should be considered in the differential diagnosis due to the similarity of the joint involvement.

Keywords: Pachydermodactyly, proximal interphalangeal joint, painless swelling

ÖZ

Pakidermodaktili (PDD), genellikle genç erkeklerde görülen, periartiküler yumuşak dokunun asemptomatik, ilerleyici şişliği ile karakterize benign edinsel dijital fibromatozdur. PDD'nin etyolojisi tam olarak bilinmemekle beraber muhtemelen tekrarlayan mekanik stimülasyonun bir sonucudur. Tanı genellikle klinik olarak konur. Histopatolojide kalın kollajen demetleri ve dermal musin birikimi vardır. Benign seyir gösterdiğinden tedavi genellikle endike değildir. Başta romatoid artrit olmak üzere el eklemlerini etkileyen romatizmal hastalıklar, eklem tutulumunun benzerliğinden dolayı, ayırıcı tanıda düşünülmelidir.

Anahtar Kelimeler: Pakidermodaktili, proksimal interfalangeal eklem, ağrısız şişlik

Introduction

Pachydermodactyly (PDD) is a benign skin fibromatosis which is characterized by asymptomatic painless periarticular swelling in the proximal interphalangeal (PIP) and sometimes metacarpophalangeal (MCP) joints, which usually affects young adult males. The etiology is not fully understood, but mechanical trauma is thought to be responsible. There is an accumulation of increased dermal collagen histopathologically (1). Rheumatological diseases, particularly rheumatoid arthritis affecting the joints of the hands should be considered in the differential diagnosis due to the similarity of the joint involvement (2).

We would like to draw attention to this rare situation which should be considered in rheumatology clinical practice as it causes painless joint swelling.

Case Report

A 22-year old male patient was referred to our outpatient clinic with the complaint of swelling of the 3rd PIP joint of the right hand for the last 2.5 years. Symmetrical swelling was observed around the PIP joints of bilateral digits 3-4 (Figure 1).

The patient had previously undergone methotrexate treatment in another medical center with the diagnosis of rheumatoid arthritis, but the patient discontinued the treatment on his own request as no benefit was obtained. There was no pain in the finger joints of the hands, no morning stiffness, and no night pain. The results of the systemic examinations were unremarkable. There was no family history of psoriasis or rheumatological disease. In the physical examination, there were joint swellings in the 4th and more evidently in the 3rd PIP joints of the right hand and the 3rd and 4th PIP joints of the left hand. There was no tenderness with palpation, and range of motion was complete. In the laboratory tests, the results of the complete blood count, liver function tests, kidney function tests, erythrocyte sedimentation rate, C-reactive protein, thyroid stimulating hormone test, and urinalysis were normal. Hepatitis markers, antinuclear antibody, rheumatoid factor, anti-cyclic and citrullinated peptide antibody were negative. Periarticular soft tissue swelling was observed on the direct radiograph but there was no sclerosis, joint space narrowing, erosion or periosteal reaction (Figure 2).

There was no evidence of inflammation in the PIP joint on ultrasonographic evaluation. Thickening of the subcutaneous tissue was

Address for Correspondence/Yazışma Adresi: Ayşe Ünal Enginar, Akdeniz University Medicine School, Department of Physical Therapy and Rehabilitation, Rheumatology, Antalya, Turkey

Phone: +90 532 723 99 88 **E-mail:** ftrdrayseenginar@gmail.com **ORCID ID:** orcid.org/0000-0003-0273-6268

©Copyright 2019 by the İstanbul Training and Research Hospital/İstanbul Medical Journal published by Galenos Publishing House.

©Telif Hakkı 2019 İstanbul Eğitim ve Araştırma Hastanesi/İstanbul Tıp Dergisi, Galenos Yayınevi tarafından basılmıştır.

Received/Geliş Tarihi: 22.09.2018

Accepted/Kabul Tarihi: 11.01.2019

detected on MRI of the hand, but no evidence could be associated with synovitis (Figure 3).



Figure 1. Photographic image of bilateral hands. Symmetrical swelling around PIP joints of bilateral digits 3-4



Figure 2. Hand radiograph showing soft tissue swelling around the proximal interphalangeal joints without joint narrowing or bony erosions



Figure 3. Magnetic resonance imaging reveals soft tissue swelling of the third to fourth proximal interphalangeal joints of the hand without articular or bony abnormality

A biopsy was taken from the lateral of the 3rd PIP joint of the right hand with the preliminary diagnosis of pachydermodactyly, and hyperkeratosis and thickened dermis with collagen deposition were detected (Figure 4).

The patient was diagnosed with pachydermodactyly after evaluation of the clinical, laboratory, radiology and biopsy findings. The patient was given information that PDD is a non-inflammatory disease with a benign course.

Written informed consent was obtained from the patient for details to be published in this study

Discussion

PDD is a rare digital fibromatosis, first reported by Bazex et al. in 1973 (3), then named PDD by Verbov in 1975 (4). To date, approximately 150 cases of PDD have been reported in literature (5). It is characterized by soft tissue swelling especially in the lateral part of the PIP joints of the 2nd-4th fingers of adolescent males. It is seen approximately 4 times more commonly in males (2). There are also familial cases as case reports in literature (6). Although the etiology is not fully understood, mechanical trauma is a precipitating factor in 44% of patients (2). Continuous pulling and docking of fingers can be seen in some occupations (athletes, musicians, computer-users, farm-workers) and also in subjects with Asperger's syndrome and obsessive compulsive disorder. Therefore, a psychiatric consultation is essential (1,7). Bardazzi et al. defined PDD in 5 classes. Classical (which affects multiple joints and is due to repeated microtrauma, localized (which affects only one joint), transgradient (which affects the dorsum of the hand and metacarpophalangeal areas), familial and the form associated with tuberous sclerosis (8).

There is no specific laboratory finding for the diagnosis. The acute phase proteins are normal and there are no specific auto-antibodies for the disease. No periarticular osteoporosis, periosteal reaction, erosion, osteophyte or cysts are observed on radiographs, only soft tissue swelling. No pathological findings such as synovitis, capsulitis, tendinitis or hypervascularization apart from soft tissue swelling are

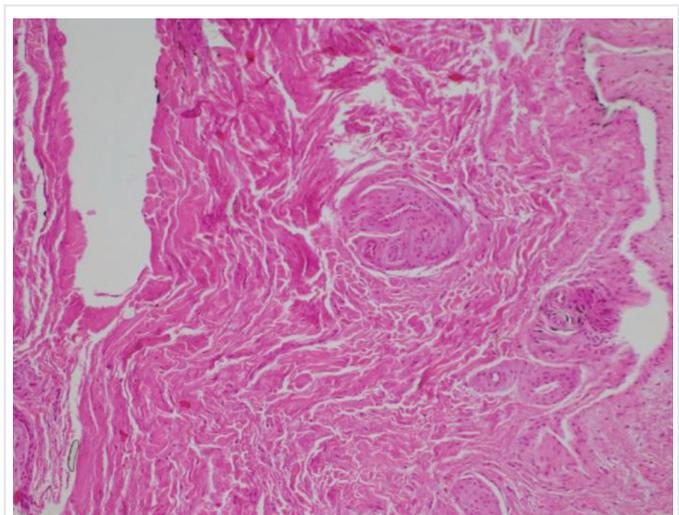


Figure 4. Skin biopsy specimen exhibits marked hyperkeratosis and thickened dermis with collagen deposition (H&E, ×100)

found on MRI screening (9). In the histopathological examination, PDD is characterized by hyperkeratosis and acanthosis of the epidermis, and thick collagen bundles and increased fibroblasts in the dermis. The cytological appearance of the fibroblasts are benign. There may also be mucin accumulation in the interstitium in varying degrees and the number of elastic fibres may be decreased (10). When collagen analysis is performed, the amount of collagen type III and V is higher. Collagen fibres are seen to be less uniform and of smaller diameter in electronmicroscopy (1,2).

Chen et al. proposed the diagnostic criteria of no symptom of the patient, no morning stiffness, no pain with movement and tenderness with palpation, finger swelling especially on the ulnar or radial side, but not peripheral, normal laboratory test results, and only soft tissue swelling on X-ray (1).

In the differential diagnosis, there should be consideration of rheumatological diseases such as juvenile idiopathic arthritis and rheumatoid arthritis, bone diseases such as spina ventosa, and pachydermatoperiostosis (11), skin diseases such as knuckle pads (12) and foreign body granuloma, and genetic causes such as Thiemann's disease (2).

There is no universally accepted treatment option. It may be useful to avoid mechanical stimulation. Some cases have been improved with oral tranilast, intralesional triamcinolone injection and surgical excision of fibrotic tissues. Tranilast is an anti-allergic drug which inhibits collagen synthesis (13,14). No treatment was applied to the current patient, but as he had a habit of pulling his fingers, he was advised to discontinue this habit. The patient was referred to the psychiatry clinic for consultation.

In conclusion, PDD is a benign, uncommon disease. As it causes swelling in the PIP joints, differential diagnoses must consider rheumatological diseases, primarily rheumatoid arthritis. Loss of function, restricted range of motion, no morning stiffness and no pathology determined with laboratory and imaging methods, are significant characteristics. Correct recognition of PDD prevents unnecessary referrals and tests and, most importantly, inappropriate treatment.

References

1. Chen CK, Shyur SD, Chu SH, Huang LH, Kao YH, Liu LC. Pachydermodactyly: three new cases in Taiwan. *J Microbiol Immunol Infect*. 2015;48(3):340-4
2. T. Dallos, B. Oppl, L. Kovács, and J. Zwerina. Pachydermodactyly: a review. *Current Rheumatology Reports*, 2014;16:9-442,
3. Basex A, Dupre A, Teillard J. Pachydermie digitale des premieres phalanges par hyperplasie conjonctive dermique et aplasie hypodermique. *Bull Soc Fr Dermatol Syphiligr* 1973; 80: 455-456.
4. Verbov J. Letter: Pachydermodactyly: a variant of the true knuckle pad. *Arch Dermatol* 1975; 111: 524.
5. Zuber Z, Dyduch G, Jaworek A, Dorota Turowska-Heydel, Małgorzata Sobczyk, Marta Banach-Górnicka et al. Pachydermodactyly - A report of two cases. *Reumatologia*, 2016; 54: 136-140.
6. F. Russo, A. Rodriguez-Pichardo, F. Camacho. Familial pachydermodactyly *Acta Derm Venereol*, 1994;74: 386-387
7. S.L. Woodrow, N.P. Burrows Pachydermodactyly in association with Asperger syndrome *Clin Exp Dermatol*, 28 (2003), pp. 674-675
8. Bardazzi F, Neri I, Raone B, Patrizi A. Pachydermodactyly: seven new cases. *Ann Dermatol Venereol*. 1998;125:247-50.
9. Paravina M, Stanojevic M, Jovanovic D, Ljubisavljevic D. Pachydermo-dactyly: a case report and literature review. *Serbian Journal of Dermatology and Venereology*. 2014;6(4):174-85.
10. W. P. James and A. H. Hosler. Pachydermodactyly, in *Weedons Skin Pathology*, Churchill Livingstone Elsevier, Edinburgh, Scotland, 4th edition, 2016.
11. Rai A, Zaphropoulos GC: An unusual case of peri-articular soft tissue finger swelling in an adolescent male: pachydermodactyly or pachydermoperiostosis? *Br J Rheumatol* 1994; 33: 677-9
12. Chamberlain AJ, Venning VA, Wojnarowska F: Pachydermodactyly: a form fruste of knuckle pads? *Australas J Dermatol* 2003; 44: 140-3
13. A. Plana Pla, J. Bassas Vila, M.A. Toro Montecinos, C. Ferrandiz Foraster. Pachydermodactyly successfully treated with triamcinolone injections. *Actas Dermosifiliogr*. 2014;105:319-21.
14. Higuchi C, Tomita T, Yoshikawa H. Pachydermodactyly treated with tranilast in a young girl. *Case Rep Orthop* 2014;ID 132854, 4