Journal of Microbiology, Immunology and Infection (2015) 48, 340—344

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CASE REPORT

Pachydermodactyly: Three new cases in Taiwan

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Received 15 November 2011; received in revised form 1 September 2012; accepted 13 September 2012
Available online 3 December 2012

KEYWORDS
Collagen;
Fibroblast;
Fibromatosis;
Pachydermodactyly;
Proximal interphalangeal joint

Pachydermodactyly (PDD), Greek for thick-skin-finger, is an infrequently recognized benign disorder characterized by painless fusiform swelling of the soft tissues around the proximal interphalangeal joints of the hands. Histopathologic features include increased dermal accumulation of collagen fibers. Young males are predominantly affected. PDD is quite rare with approximately 90 cumulative cases reported worldwide. We report three new cases of PDD in Taiwan, including two female patients. Except for patient 1 having the habit of cracking the knuckles, and patient 2 having a history of patent ductus ateriosus post catheterization, the histories of all three patients were unremarkable. X-ray of bilateral hands revealed no abnormal finding except for soft tissue swelling around proximal interphalangeal joints. Laboratory examinations all showed negative results. No local or systemic treatment was given to these patients to treat PDD, with the exception of non-steroidal anti-inflammatory drugs prescribed to one patient for a short period. Skin care with local irritation avoidance was explained to all three patients. The long-term outcome of PDD was benign.

Introduction

Pachydermodactyly (PDD) is a rare disease with non-inflammatory bulbous swelling involving the lateral aspects of the fingers at the level of proximal interphalangeal (PIP) joints. It affects almost exclusively young

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1684-1182/$36
http://dx.doi.org/10.1016/j.jmii.2012.09.002

males. Histopathologic features include increased dermal accumulation of collagen. PDD was first described by Bazex et al in 1973. Two years later, Verbov coined the word pachydermodactyly.[1] To date, more than 90 cases have been reported worldwide.[2] We present three additional cases from Taiwan, and a brief review of this disease is presented.

Case reports

Patient 1

A 13-year-old boy visited our clinic, presenting with bilateral second to fifth PIP joint and thumb swelling which had developed over a period of 1 year. The swelling of the joints was more noticeable on the lateral aspects of the fingers, and range of movement was not impaired (Fig. 1A and B). No pain was noted. Furthermore, there was no morning stiffness, fever, rashes, mouth ulcer, uveitis, or gastrointestinal symptoms. He denied any trauma history, but he did exhibit the habit of cracking his knuckles frequently. There were no other articular changes. He had generally been in good health with the exception of asthma and allergic rhinitis history. Complete blood count with differential count (CBC/DC) was normal. Antinuclear antibody (ANA) was negative. Rheumatoid factor (RF) was $<$20 IU/mL. Erythrocyte sedimentation rate (ESR) was 2 mm/h. X-ray showed swelling of the soft tissue surrounding PIP joints of bilateral second to fifth fingers, and interphalangeal joint of bilateral thumb fingers was also seen (Fig. 2). However, no definite bony pathology of both hands was observed. His parents' hands were also examined, but no similar findings were found. Naproxen was initially prescribed to the patient because he was suspected of having polyarticular juvenile idiopathic arthritis (JIA). We stopped Naproxen afterwards due to there being no effect on the related lesions. The patient was taught proper skin care techniques and was encouraged to refrain from knuckle cracking. He received follow-up at our clinic for more than 1 year, and related swelling was shown to be reduced at his most recent examination.

Patient 2

A 13-year-old girl visited our clinic, presenting with bilateral second to fifth PIP joint swelling that had progressed for 3–4 months. The swelling of the joints was more noticeable on the lateral aspects of the fingers. Right-hand findings were more dominant than left-hand findings (Fig. 1C). No pain was noted related to the swollen joints, and range of motion was normal. No morning stiffness, fever, rashes, mouth ulcer, uveitis, or other global symptoms were noted. She denied any trauma or family history. There were no other articular changes. She had been healthy before developing this condition, except for atopy and patent ductus ateriosus (PDA) post catheterization. PDA was identified at the age of 12 via school health screening with the discovery of a grade 1 to grade 2 systolic murmur. No shortness of breath, cyanosis, or syncope was mentioned. Echocardiogram showed one PDA of 0.208 cm, and catheterization was suggested. During the catheterization (December 31, 2010), no coil embolization was performed because the PDA caliber was quite small (0.13 cm). After the procedure, she recovered, and echocardiogram follow-ups performed at 4 and 8 months after catheterization all showed very small PDA with trivial residual flow. Then she was referred by a cardiologist due to issues with her fingers. Her laboratory examination results, such as CBC/DC, were normal. ANA was negative, and anti-double strand DNA (anti-dsDNA) was also negative. RF was $<$20 IU/mL and ESR was 7 mm/h. X-ray showed soft tissue swelling involving the second to fifth PIP joints. The bony structure of both hands appeared to be normal. She received follow-up at our clinic for several months without taking medication. After the patient received training with regard to skin care and avoidance of local trauma, related finger swelling lesions showed improvement, without any pain or progression.

Patient 3

A 12-year-old girl visited our clinic presenting with bilateral second to fourth PIP joint swelling that had developed over 2.5 years. No pain was noted, and range of movement was not impaired. Right-hand findings were more dominant than the left-hand findings. There was no morning stiffness, fever, rashes, mouth ulcer, uveitis, or other systemic symptoms. She denied any trauma history or family history. There were no other articular changes. She had been in good health before diagnosis, with the exception of asthma and allergic rhinitis history. CBC/DC was normal, while ANA was negative. RF was $<$20 IU/mL and ESR was 12 mm/h. X-ray showed soft tissue swelling involving second to fourth PIP joints. She accepted follow-up at our clinic for a half-year without receiving treatment. After being educated about proper skin care and the avoidance of local trauma, her finger swelling lesions showed improvement, without any sign of discomfort.

Discussion

Pachydermodactyly, from the Greek pachy (thick), dermo (skin), and dactylos (finger), was first described by Bazex et al in 1973 and named by Verbov in 1975. It is a rare and benign form of digital fibromatosis.[1] Similar changes on hands had already been described by Garrod in 1904.[3] To date, more than 90 cases have been reported worldwide.[2] In Asia, one case from Taiwan and four cases from South Korea have been reported.[4,5] PDD is most commonly seen in adolescent boys and has been reported among several ethnicities.[1,4,6] The mean age of PDD is 21.2 years with a male/female ratio of 3:2 to 5:1.[2,4] However, in our report, two of the three cases were young females, which is rare in comparison with previous reports. Often there is a history of repeated friction to the sides of the fingers, such as that caused by interlocking fingers or continual hand rubbing. This pattern may also be seen with occupational exposure and in individuals with Asperger syndrome or obsessive compulsive disorders.[7] Patient 1 in our report was an adolescent male who had a personal habit of cracking his knuckles for a long time, but he denied any history of psychiatric problem. Therefore, the PDD of the boy may be related to the habit of cracking or squeezing

the finger joints. The two girls in our report denied having a similar habit, and the cause of PDD in their cases remains unknown. PDD may occur as an additional cutaneous sign of tuberous sclerosis. Familial PDD has also been reported,[5,8] but none of our patients had a family history of PDD according our reports.

PDD presents as asymptomatic fusiform swelling, lacking pain, and early morning stiffness. The second through the fourth PIP joints are primarily affected, rarely the fifth, and the thumbs are most always spared.[4,6,7] Swelling is radial and ulnar in distribution, not circumferential, and there is no localized tenderness, warmth, or reduced range of motion. As in other articles presented, our three cases had typical manifestation of bilateral second to fourth PIP joints involved, and two (patients 2 and 3) had predominant swelling sign over their right hands. In addition, two cases (patients 1 and 2) exhibited fifth PIP joint involvement, and one case (patient 1) had thumb interphalangeal joint swelling. Typical changes are rarely seen over the distal interphalangeal joints. Fibromatous thickening of the dorsum of the hands, or firm nontender nodules with metacarpophalangeal involvement, has been previously described.[9] These were not observed in our patients.

Bardazzi et al[10] suggested classifying PDD into five main types: (1) classic pachydermodactyly, both idiopathic and associated with trauma, and primarily described in male patients with several affected fingers; (2) monopachydermodactyly or localized pachydermodactyly, which may also be idiopathic or induced from trauma; (3) pachydermodactyly transgrediens, in which the cutaneous changes may extend to the palms, particularly to the metacarpophalangeal joints; (4) familial pachydermodactyly, which can be classic or pachydermodactyly transgrediens; and (5) pachydermodactyly, which is associated with tuberous sclerosis and may sometimes be painful. Our three patients may be classified under type I based on their clinical presentations.

Investigations regarding PDD reveal normal inflammatory markers, and negative ANA and RF assays. The same negative results were found in our three PDD patients. Routine imaging studies show solely periarticular soft tissue swelling without osteopenia, joint space narrowing, or marginal erosions. The X-ray survey of the only male patient in our report (patient 1) showed swelling of soft tissue surrounding PIP joints of bilateral second to fifth fingers, and also interphalangeal joint swelling of bilateral thumb fingers, which has been rarely seen in past reports.[4] Patient 2 had the same image results but no abnormal thumb finding. When magnetic resonance imaging (MRI) is performed, fusiform soft tissue swelling is seen at the PIP joints with normal collateral ligaments, tendons, and bone.[11,12] When a technetium-labeled bone scan shows increased uptake, this pattern is suggestive of pachydermatoperiostosis rather than PDD.[13] Biopsy of the overlying skin exhibits hyperkeratosis, mild epidermal hyperplasia, and a thickened dermis with extension of collagenous fibers into the subcutaneous tissues. The cytology of the fibroblasts appears benign. Collagen

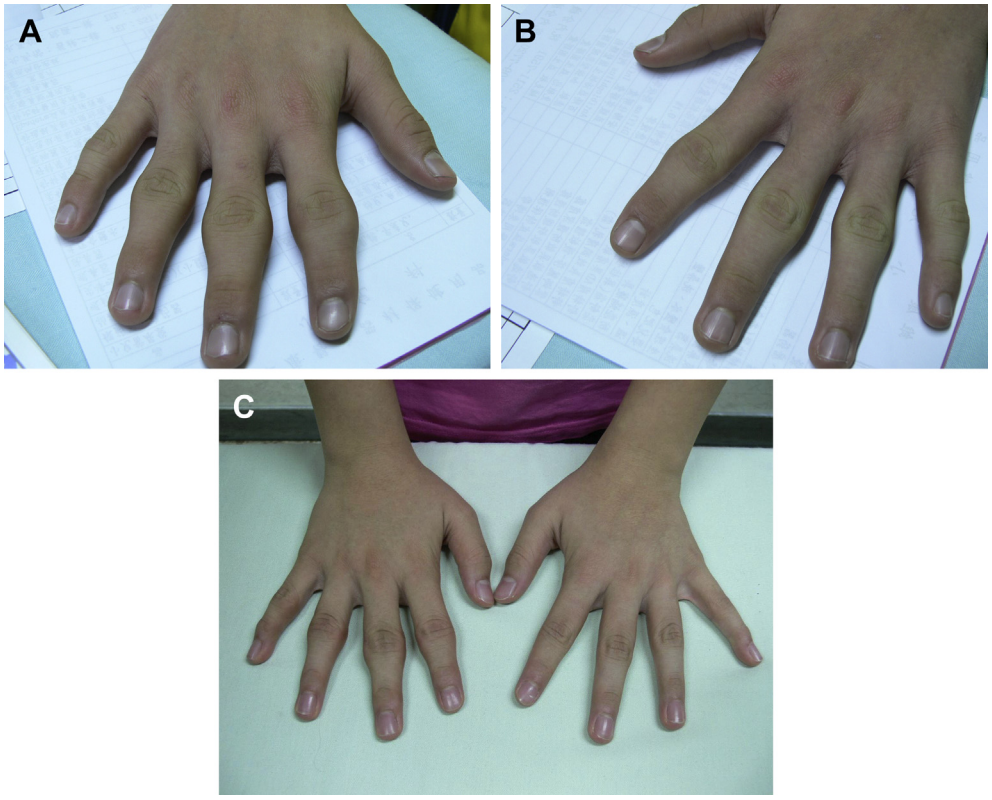

Figure 1. (A, B) Bilateral second to fifth proximal interphalangeal (PIP) joints and interphalangeal joints of bilateral thumbs swelling, especially on the lateral aspects of the fingers (patient 1). (C) Bilateral second to fifth PIP joints swelling, right-hand dominant. This patient had past history of patent ductus ateriosus post catheterization (patient 2).

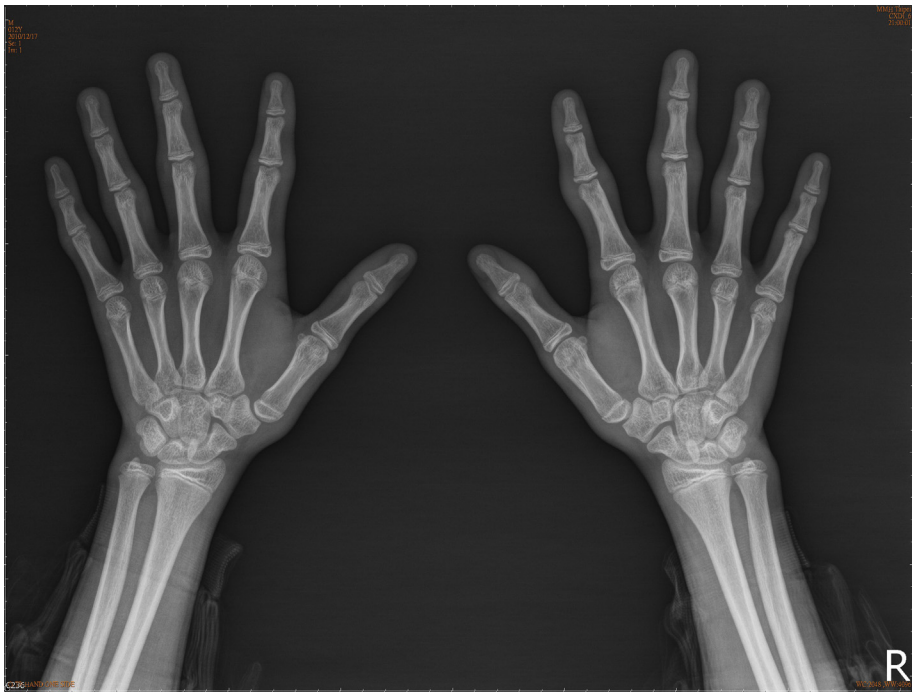

Figure 2. X-ray of hands (patient 1): swelling of soft tissue surrounding PIP joints of bilateral second to fifth fingers and interphalangeal joints of bilateral thumbs.

analysis sometimes demonstrates larger amounts of types III and V collagen in a pattern typical of fibromatosis. Electron microscopy discloses collagen fibers that are less uniform and of smaller diameter.[4,6,10,13] We did not perform MRI, bone scan, or skin biopsy survey at our clinic for these three cases. Moreover, patient 2 had a history of PDA post catheterization about 4 months before PIP joint swelling was observed. No coil embolization was performed because the PDA was very small. We considered that the PDD was not associated with the PDA in the patient because she had neither signs of respiratory nor heart failure.

The most important differential diagnosis with PDD was polyarticular JIA,[11,13,14] with the characteristics of erythematous change, swelling, warmth, and pain over multiple joints persistent for 6 weeks. It is also worth noting that we initially mistook patient 1 as having JIA, but corrected the diagnosis in favor of PDD after a thorough review of past PDD-related articles. Other diagnostic considerations include Thiemann's disease, Knuckle pads, pachydermatoperiostosis, and xanthomatous deposits. Another similar disease, Knuckle pads, which occur on the dorsal aspect of the PIP joints, is either true (primary) or false (secondary), the former being of unknown origin and the latter occurring as a result of biting or chewing. Thiemann's disease (hereditary necrosis of the epiphysis) has an initially similar clinical appearance but shows progression with painful deformity of the fingers and limitations in function, and X-rays are sometimes helpful for a differential diagnosis.[2] Pachydermoperiostosis, a very rare genetic disease associated with seborrhoea, causes digital enlargement from a combination of pachydermia, periostosis, and clubbing of the fingers, related to altered proteoglycan synthesis by fibroblasts.[2] Our cases can be differentiated easily from the above similar diseases according to the clinical features and image findings.

There is no effective medical treatment for PDD at this time. Intralesional corticosteroids may help reduce swelling; however, recurrences may occur.[15] Surgical excision may achieve cosmetic benefit.[16] Although no satisfactory treatment for PDD has been described, considerable improvement may occur with cessation of the source of mechanical trauma.[14] Our patients all received regular follow-up at our clinic for several months. No steroids or cytotoxic agents were used. Skin care education was provided to them, and local irritation or friction was discouraged. Only patient 1 was prescribed with a nonsteroidal anti-inflammatory drug (Naproxen) for a short period, but this treatment had no effect. We have tried our best to review previous articles about the long-term prognosis of PDD; however, to date, no articles have addressed this issue. At the last follow-up, the finger swelling lesions showed continued improvement in all of the patients. There was no sign of disease progression sign or complication about the PDD. A longer period is required to evaluate these three cases in terms of long-term outcome, especially in patient 1 after ceasing the habit of "cracking the knuckles."

Finally, we concluded that PDD diagnosis criteria are as follows: (1) the patient has no symptoms; (2) morning stiffness is absent; (3) pain on motion and tenderness to palpation is absent; (4) finger swelling is radial or ulnar in location, rather than circumferential; (5) laboratory test results are unremarkable; and (6) plain radiographs show only soft tissue swelling. With these typical findings, additional investigations, such as MRI or skin biopsy, are rarely needed to establish a diagnosis of PDD.

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