# Arthritis or maybe not? The great mimicker of juvenile idiopathic arthritis

Bento da Silva A[1]*, Lourenço MH[1], Gonçalves MJ[1], Branco JC[2], Costa M[1], Mourão AF[1]

[1] Department of Rheumatology, Centro Hospitalar de Lisboa Ocidental EPE

*ORCID: https://orcid.org/0000-0002-8590-8119

[2] Comprehensive Health Research Center (CHRC), NOVA Medical School



**Correspondence to**

Ana Bento Silva

E-mail: adbentosilva@gmail.com

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**Abstract**

Arthritis in the paediatric population is the hallmark of many rheumatic inflammatory diseases, as well as other cutaneous, infectious, or neoplastic conditions. It can be quite devastating, whereby prompt recognition and treatment of these disorders are essential. However, arthritis can sometimes be mistaken for other cutaneous or genetic conditions leading to misdiagnosis and overtreatment. Pachydermodactyly is a rare and benign form of digital fibromatosis, usually manifested by swelling of the proximal interphalangeal joints of both hands, mimicking arthritis. The authors report a case of a 12-year-old boy with a one-year history of painless swelling of the proximal interphalangeal joints of both hands that was referred to the Paediatric Rheumatology department due to the suspicion of juvenile idiopathic arthritis. The diagnostic work-up was unremarkable, and the patient remained asymptomatic over an 18-month follow-up period. A diagnosis of pachydermodactyly was assumed and no treatment was introduced, given the benign nature of the disorder and absence of symptoms. Therefore, it was possible to safely discharge the patient from the Paediatric Rheumatology clinic.

**Keywords:** Ultrasonography; Juvenile idiopathic arthritis; Soft tissue rheumatism; Adolescent rheumatology; Hand; Paediatric/Juvenile rheumatology.

## Introduction

Arthritis in children and adolescents can have a great diversity of differential diagnoses, including a wide number of rheumatic diseases, orthopaedic conditions, infections, and malignancies. Juvenile idiopathic arthritis (JIA) is the most common rheumatic disease in the paediatric population, mainly characterized by the presence of arthritis for more than 6 weeks[1], but even this can have a wide range of mimickers. Usually, a child is brought to the Rheumatologist following complaints of tender and swollen joints, frequently identified in the physical exam. However, the physical exam has its limitations, and not every apparent swollen joint has, in fact, an effusion. Intumescence of periarticular structures can be easily mistaken with joint swelling, leading to an erroneous diagnosis[2].

Pachydermodactyly (PDD) is a rare and benign form of digital fibromatosis, first described by Bazex in 1973 and later named by Julian Verbov in 1975[2,3,4]. It has an unknown incidence, but recent data reports around 150 cases worldwide, with a rising number in the last years, likely due to an increased awareness of this condition[2,5]. In Portugal, the first two cases were reported in 1995, both in otherwise healthy young adolescents (13 and 14 years old)[6].

Due to its rarity and possible lack of recognition of this clinical entity, few information is available regarding its etiology and pathophysiology, and the evidence is mostly based on clinical reports. PDD is characterized by the insidious and progressive thickening of periarticular tissues, mainly affecting the proximal interphalangeal (PIP) joints of both hands, giving the fingers a fusiform appearance[2]. It is an asymptomatic disorder with a benign course without impairment of function[7]. It is more common in males (male to female ratio of almost 4:1) and begins more frequently in the teenage years, although cases have been reported in patients from 5 to 74 years old[2,5,8]. Because of its clinical presentation, PDD may mimic many joint diseases, like JIA, and recognition of this still-unknown entity is necessary to avoid misdiagnosis and overtreatment.

## Case Report

The authors present a case of a 12-year-old Caucasian boy, who was referred to the Paediatric Rheumatology department of a tertiary hospital in Portugal with a one-year history of an insidious and slowly progressive swelling of PIP joints of both hands in a symmetric distribution. He denied fever, pain, impaired mobility of fingers, or the involvement of any other joint. In fact, he was a very active child, practicing paddle and soccer without any restrictions.

After a detailed clinical history, the patient denied personal or family history of psoriasis or other skin manifestations, gastrointestinal complaints, Raynaud's phenomenon, respiratory or cardiovascular symptoms, or previous trauma. He also discarded any other possible repetitive mechanical stimulation habits of the fingers. He had a personal history of atopic eczema and a family history of Celiac disease, diagnosed in his mother. The physical exam revealed an apparent swelling of the second to fifth PIP joints of both hands, more pronounced on the right side, giving the fingers a fusiform appearance (Figure 1). However, there was no tenderness on examination of the involved joints or limitation of the range of motion. A thorough and detailed physical exam was performed with no other relevant findings. The boy had previously been seen by his family doctor that prescribed a course of nonsteroidal anti-inflammatory drugs (ibuprofen 10mg/Kg/day for 7 days) without any benefit. Regardless of the absence of pain and impairment of function, the suspicion of rheumatic disease, mainly JIA, was raised, and further exams were performed.

His blood analysis did not show any changes in the complete blood count, C-reactive protein, erythrocyte sedimentation rate, renal and liver function, thyroid function, glucose, and electrolyte levels, including calcium; the urine sediment was also normal. Immunology revealed positive antinuclear antibodies (ANAs) with a titer of 1/160 and positive HLA-B27; anti-endomysial and anti-transglutaminase antibodies were also positive, but a duodenal biopsy excluded celiac disease. The remaining study was negative, including rheumatoid factor, anti-citrullinated peptide antibody (ACPA), anti-double stranded DNA antibody (anti-dsDNA), anti-soluble extractable nuclear antigens (anti-ENA), anti-soluble liver antigen, anti-liver cytosolic antigen type 1, anti-liver kidney microsomal type 1, anti-liver cytosol type 1, anti-mitochondrial, anti-Sp100, anti-PML, anti-glycoprotein 210, anti-*Saccharomyces cerevisiae* and antineutrophil cytoplasmatic antibodies. Serologies were also negative for toxoplasma, *SARS-CoV2* virus, human immunodeficiency virus, and hepatitis B and C viruses; rubella, cytomegalovirus, and Epstein-Barr virus were immunoglobulin (Ig) G positive but IgM negative.

An X-ray of both hands (Figure 2) was performed and revealed signs of periarticular soft tissue edema around the second to fifth PIP joints bilaterally, without joint or bone involvement; X-ray of the sacroiliac joints (requested because of positivity for HLA-B27 despite the absence of low back pain) was also normal. Hand ultrasound (Figure 3) was innocent for changes suggestive of articular or periarticular inflammatory activity, mainly synovitis or Doppler sign, and did not show joint erosions.

Despite the analytical findings, especially positive ANAs and HLA-B27, this male adolescent remained asymptomatic throughout the following appointments and did not reveal any signs or symptoms suggestive of a rheumatic inflammatory disease. Thus, a diagnosis of PDD

was assumed. Due to its benign course and absence of symptoms, pharmacological therapy was not prescribed. After 18 months of follow-up, this now 14-year-old boy remains asymptomatic, which allowed for his discharge from the Paediatric Rheumatologic clinic.

**Discussion**

The diagnosis of PDD is quite challenging due to both its extensive differential diagnosis and lack of awareness of this disorder. Fernández *et al.*[5] performed a systematic literature review of 97 studies, published in 2021, including a total of 139 patients with this disorder. Cases were identified around the globe and in people of every age group (5 to 74 years old). Male patients were affected four times more frequently than women, and the mean age at onset was 15 years old, but highest in female subjects (19 years versus 15 years in male patients)[2,5].

The most striking features of PDD are the absence of both pain and impairment of function or mobility, with only one case reported in the literature of a 39-year-old man with a deforming non-erosive involvement of PIP and DIP joints of both hands that was considered an atypical presentation[9;10]. The most frequently affected joints were the PIP joints of both hands (typically the second, third and fourth), usually with a symmetric distribution (87%), although affection of distal interphalangeal (DIP), metacarpophalangeal (MCP) and metatarsophalangeal (MTF) joints and unilateral involvement was also reported[5,11].

The etiology and pathophysiology of PDD are still unknown. Repetitive mechanical stimulation, such as that seen in musical activities like playing guitar, some sports, and manual workers, has been perceived as the most probable causative factor, present in 43% of patients. Some authors consider PDD a variant of knuckle pads, as both have a similar distribution and histopathological features and are associated with repetitive skin stimulation, while others believe they are distinct conditions of a wider spectrum of mechanical-induced disorders[2,5]. Associations with neuropsychiatric pathologies, like obsessive-compulsive disorders, hormonal factors, and hypermobility syndromes, have also been called into the equation[2,5,12]. Despite the fact that it has been diagnosed in otherwise healthy subjects, Zuber *et al.* described a case of a 14-year-old boy with undifferentiated connective tissue disease with Raynaud's phenomenon[11]. Family history is absent in the majority of cases (91%), but a familiar form of the disease with a different pathophysiology has been described, more frequently in female patients[5].

Complementary exams are usually unremarkable, highlighting only the soft tissue swelling observed in imaging of affected structures[2]. Ultrasound reveals only a thickening of dermal soft tissue, without signs of inflammatory activity, and without joint, bone or tendon involvement[13]. Similarly, Magnetic Resonance Imaging (MRI) does not have a major role in the diagnosis of PDD,

usually demonstrating non-enhancing soft tissue swelling without hypervascularization and absence of inflammation of other musculoskeletal structures[2]. Biopsy can reveal an increased deposition of abnormally structured collagen in the dermis and hypodermis and epidermal hyperplasia and hyperkeratosis[2,14]. To help with the diagnosis, Chen C. *et al.* proposed some diagnostic criteria based on the following: absence of symptoms and morning stiffness, radial and/or ulnar finger swelling, normal blood work, and presence of soft tissue swelling on radiographs without any other abnormality; the presence of these features makes biopsy rarely necessary[5,14].

If identified, the discontinuation of the causative factor is recommended, as well as psychological evaluation for those with neuropsychiatric pathology, but other interventions are usually not indicated. Intralesional steroid injections and surgery could be tried with some benefit in improving the swelling, but the risks of such invasive methods and the potential recurrence of clinical manifestations need to be weighed against the good prognosis and absence of symptoms or functional impairment reported in this disorder[2,5].

Because of its clinical manifestations mimicking joint swelling, and thus arthritis, PDD can be easily misdiagnosed as a wide range of osteoarticular disorders, especially JIA, as highlighted by this case report, making its diagnosis a challenge in clinical practice. Other diseases to have in mind in the differential diagnosis include other rheumatic inflammatory disorders, acromegaly, primary and secondary pachydermoperiostosis, Thiemann's disease, infantile or juvenile digital fibromatosis and progressive nodular fibrosis of the skin, just to name a few[2,5]. In this particular case, given the family history of celiac disease, positive anti-endomysial and anti-transglutaminase antibodies, and clinical manifestations resembling arthritis of PIP joints, one could think of celiac disease as the final diagnosis. Celiac disease can, in fact, be diagnosed in asymptomatic patients or in those with arthritis without any gastrointestinal complaints. According to previous data, joint abnormalities such as effusion or synovitis were reported in 30% of children with this disease[15]. However, this patient performed a duodenal biopsy that excluded this disorder.

The extensive differential diagnosis together with the low recognition of this disease can easily lead to erroneous diagnosis. In fact, 8 cases misdiagnosed as JIA have been reported in the literature, and some even started methotrexate without any benefit[5]. This is extremely important as overtreatment, especially immunosuppression, could lead to serious adverse events without any benefit for the patient and is also a source of considerable unnecessary costs.

This case report aims to raise awareness for this still underrecognized disorder and thus avoid unnecessary exams and overtreatment. Quality evidence on this disease is still lacking and

is mainly based on case reports; therefore, more studies are needed to improve diagnosis and patient management.

**Figures**

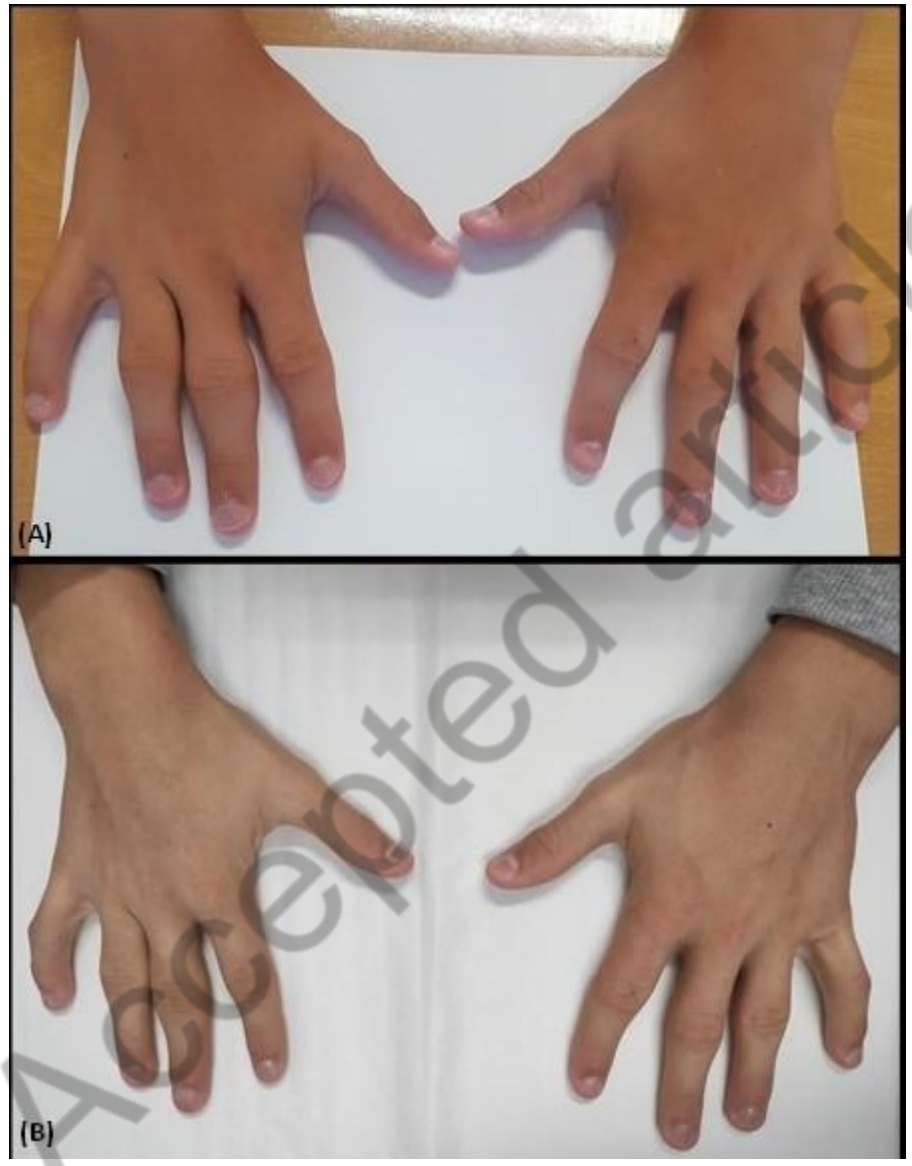


**Figure 1** - Swelling of the second to fifth proximal interphalangeal joints of both hands, more pronounced in the right side, in a 12-years-old boy at first Rheumatology appointment (A) and after 18 months (B).

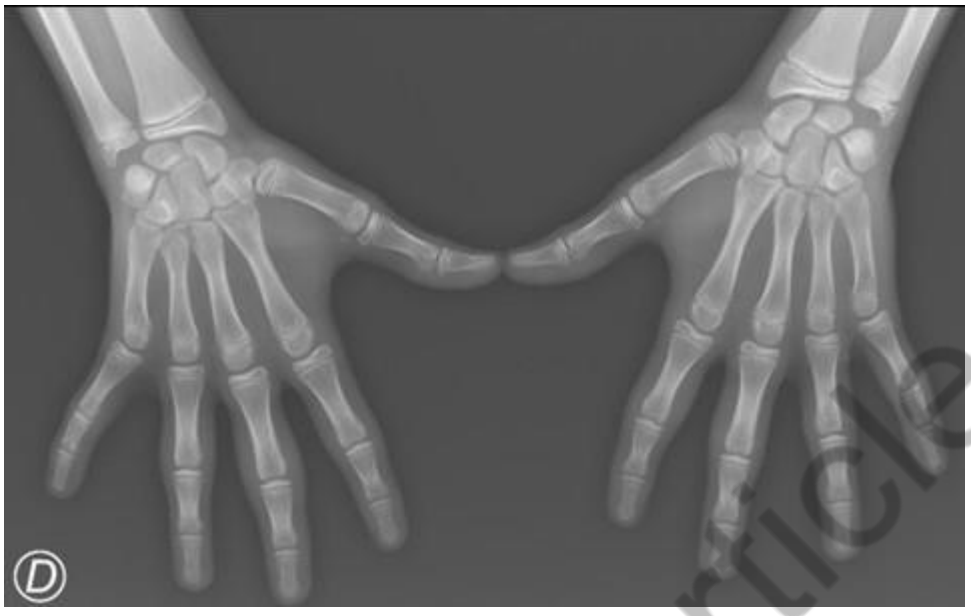

**Figure 2 -** Hand X-ray of the patient with signs of soft tissue swelling involving the second to fifth proximal interphalangeal joints of both hands. No evidence of bone or joint involvement.

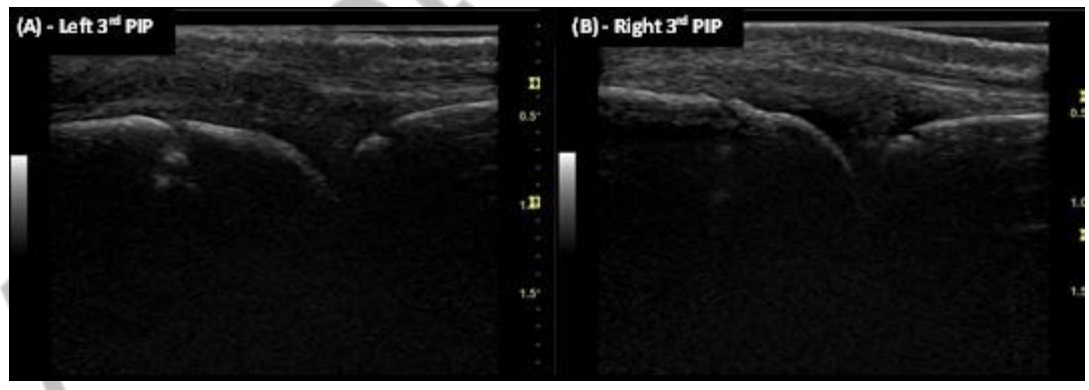


**Figure 3 -** Hand ultrasound of the patient showing the third left (A) and right (B) proximal interphalangeal joints without evidence of synovitis or tenosynovitis.

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