Pachydermoperiostosis – the complete form of the syndrome *

Paquidermoperiostose - forma completa da síndrome

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Abstract: Pachydermoperiostosis is a rare genodermatosis with various clinical presentations that include pachydermia (thickening of the skin), finger clubbing and periostitis. Its pathogenesis is uncertain and the condition affects mainly men. This report describes the case of a patient with typical, exuberant skin manifestations and classic radiological findings of this syndrome in its complete form.
Keywords: Hyperostosis; Osteoarthropathy, primary hypertrophic; Skin diseases

Resumo: A paquidermoperiostose é uma genodermatose rara, com apresentações clínicas variadas, que se apresenta com espessamento cutâneo, baqueteamento digital e periostose. Apresenta patogênese ainda incerta e acomete, principalmente, homens. Descreve-se caso de paciente com manifestações clínicas típicas e exuberantes e alterações radiológicas clássicas desta síndrome, em sua forma completa.
Palavras-chave: Dermatopatias; Hiperostose; Osteoartropatia hipertrófica primária

INTRODUCTION

Pachydermoperiostosis or primary hypertrophic osteoarthropathy is a rare, genetic syndrome characterized by progressive joint effusion associated with pachydermia, periostosis and finger clubbing. [1,2] This condition was first described in 1868 by Friedreich, who reported the condition in two male siblings and referred to it as "hyperostosis of the entire skeleton". In 1935, Touraine, Solente and Gole described pachydermoperiostosis as a primary form of hypertrophic osteoarthropathy, which differed from the most common form, secondary hypertrophic osteoarthropathy, which is always associated with a primary disease, principally with pulmonary or cardiac disease. [1,3] Classically, it affects adolescent males and its presentation is variable. [3,4] Although an autosomal dominant model of inheritance with incomplete penetrance and variable expression has been confirmed, both the autosomal-recessive inheritance and x-linked inheritance forms have been suggested. [1] The pathogenesis of this condition also remains to be clarified; however, it is believed to be triggered by abnormal amounts of some substance in the plasma of affected individuals. Many authors have reported an increase in fibroblast proliferation in bone marrow and skin biopsies, associated with diffuse hyperplasia of dermal endothelial cells, partial occlusion of vessels, thickening of collagen fibers and pericapillary lymphohistiocytic infiltrate. [1] The skin alterations would be secondary to the dysfunction of these fibroblasts due to increased activity in these cells and a consequent increase in collagen synthesis. [3]

CASE REPORT

A black, male patient of 45 years of age with a history of thickening of the skin, an increase in the volume of his hands and feet and finger clubbing since adolescence. Five years previously, he developed arthralgia associated with swollen joints in his

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ankles and knees. The patient is being followed up at the Rheumatology Department of this institute and is in use of prednisone 20 mg/day and analgesics, which have resulted in a partial improvement of his arthralgia. According to the patient, there are no similar cases in his family.

Dermatological examination revealed thickening of the skin, furrowing of the face, with cutis verticis gyrata mainly on his forehead, although the scalp was not affected (Figure 1), digital clubbing on hands and feet, with convex nails associated with hypertrophy of his hands and feet (Figures 2 and 3), swollen joints in his knees and ankles, and hyperhidrosis of his hands and feet.

Radiography of the patient's hands, wrists and feet showed irregular bony excrescences associated with lobulated periosteal reaction, cortical bone elongation and a diffuse swelling of the soft tissues (Figure 4). Knee x-rays also showed irregular bony excrescences with cortical bone elongation and signs of bilateral joint effusion with calcification at the insertion of the quadriceps tendon at the upper edge of the left patella (Figure 5).

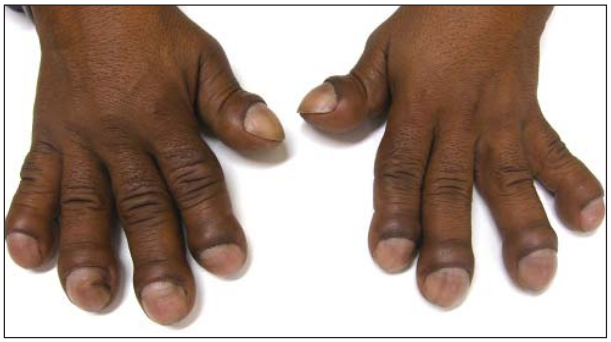

Figure 2: Presence of finger clubbing and swollen fingers

DISCUSSION

Pachydermoperiostosis is a rare genodermatosis, the onset of which normally occurs at puberty, with progressive thickening of the skin, furrowing of the face and scalp (cutis verticis gyrata), digital clubbing, edema of the periarticular tissues and periostosis of the long bones. [1-3,5] These alterations develop slowly over 5-20 years and consequently stabilize. [1] Other skin manifestations include seborrhea, palpebral ptosis, acne, hyperhidrosis, erythematous lesions on joints and a sensation of heat or burning in hands and feet. [1] The principal complications concern the joints, with arthralgia, arthritis, hydrarthrosis and hemarthrosis occurring symmetrically in the major joints and in the small joints of the hands and feet. [1,3] X-rays of the hands and feet show joint space narrowing, swelling in the soft tissues and acroosteolysis of the distal phalanges. There is also symmetrical periostosis that is more prominent in the distal lower limbs. [1,3] Histopathology shows cutaneous sclerosis and hialynosis, with perivascular infiltration of lymphoid cells. [1,3] Three distinct clinical forms of this syndrome have been proposed in accordance with the intensity of symptoms: 1) the complete form: prominent furrowing of the face, cutis verticis gyrata, digital clubbing and primary hypertrophic osteoarthropathy; 2) the incomplete form: absence of thickening of the skin and cutis verticis gyrata; 3) the fruste form: one or more skin changes, with minimal or no skeletal involvement. [3,4]

The case presented here corresponds to the complete form of the syndrome, with the presence of most of the characteristic radiological and clinical findings. The patient had significant joint involve-

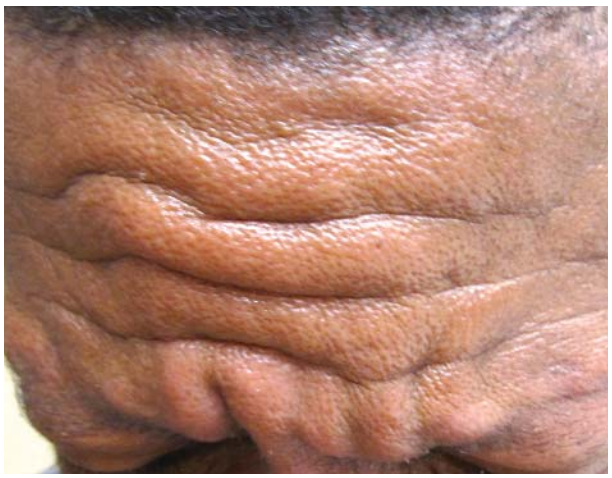

Figure 1: Cutis verticis gyrata on the patient's forehead

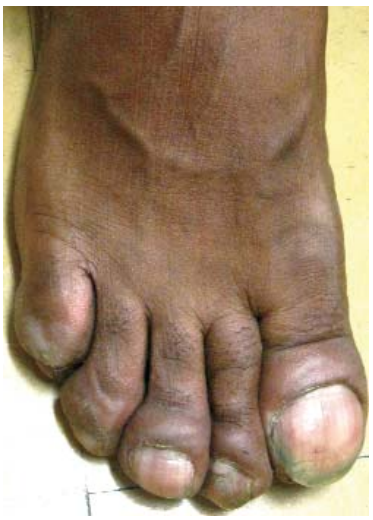

Figure 3: Increase in volume of the distal extremities of toes

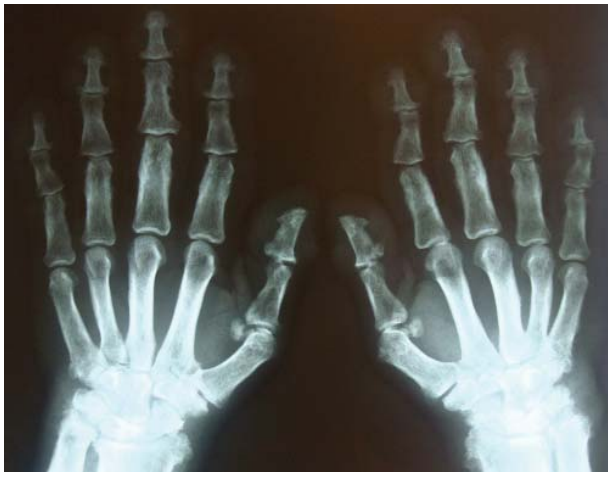

Figure 4: X-ray of the hands, showing irregular bony excrescences in phalanges, metacarpal and carpal bones, distal radius and ulna, associated with lobulated periosteal reaction, with cortical elongation of phalanges, metacarpi, radius and ulna and swelling of soft tissues

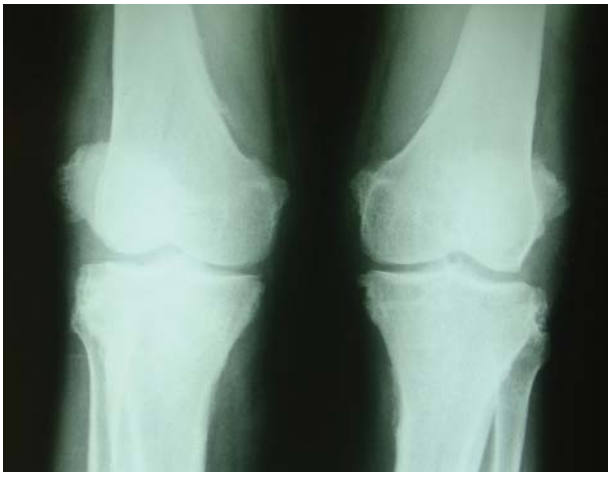

Figure 5: X-rays of knees, showing irregular bony excrescences in epiphyses, metaphysis and diaphyses of the bones of both knees and the patellae, irregular appearance, with cortical elongation of the distal femur, proximal tibia and fibula, and signs of bilateral joint effusion with calcification at the insertion of the quadriceps tendon in the upper margin of the left patella

ment, which is present in 20-40% of cases, [3] joint pain being his principal clinical complaint since it limited his routine daily activities. In addition, he had severe digital clubbing and the presence of bony excrescences was found, as shown in the x-rays of his hands and feet. Curiously in this case, cutis verticis gyrata affected only his forehead, not the scalp, which is the area most commonly affected. [1] Furthermore, although this syndrome has a strong association with heredity, [3] in this case there was no report of relatives with similar characteristics.

There is no specific treatment for this disease. The therapeutic options for the control of symptoms consist of salicylates, non-steroidal anti-inflammatory drugs (NSAID), systemic corticosteroids and colchicine. [3] A report has also been published on the use of infliximab in a patient with refractory arthritis. [6]

Since this is a disease associated with stigmatization and a consequent reduction in the patient's quality of life, diagnosis of its various clinical forms and regular follow-up by a team that includes a rheumatologist are factors of the utmost importance. This report shows that, although rare, pachydermoperiostosis should be considered by dermatologists in differential diagnoses. ❑

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