Pachydermoperiostosis – the complete form of the syndrome

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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. 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 osteoarthopathy, which differed from the most common form, secondary hypertrophic osteoarthopathy, which is always associated with a primary disease, principally with pulmonary or cardiac disease.

Classically, it affects adolescent males and its presentation is variable. 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.

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. 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.

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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1,3 Joints, with arthralgia, arthritis, hydrarthrosis and hemarthrosis occurring symmetrically in the major joints and in the small joints of the hands and feet. 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. 1,4 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-
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. 5 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.

REFERENCES

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