CASE REPORTS

Refer to: Herbert DA, Fessel WJ: Idiopathic hypertrophic osteoarthropathy (pachydermoperiostosis). West J Med 134: 354-357, Apr 1981

Idiopathic Hypertrophic Osteoarthropathy (Pachydermoperiostosis)

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IDIOPATHIC HYPERTROPHIC OSTEOARTHROPATHY is a syndrome characterized by periosteal new bone formation, clubbing, swelling of joints and skin changes; patients may have one or more of these manifestations.[1,2] The condition is also known as pachydermoperiostosis, acropachyderma with pachyperiostitis, and the Touraine-Solente-Golé syndrome. We recently saw a patient with the characteristic findings of this rare disease; the case illustrates the usually benign course.

Report of a Case

A 72-year-old Filipino man was evaluated for clubbing of the fingers and toes. According to the patient's elder sister, he had had large, unusually shaped extremities since birth. These became progressively thickened through adolescence, but he had noted no further thickening until he was given an unknown lotion 25 years ago for his thick, somewhat pruritic skin. When he applied the lotion to his lower legs, the skin thickened further and became deeply hyperpigmented. Although he had worked as a laborer, he had only occasional arthralgias after strenuous work. He had not noted excessive sweating. To the best of his knowledge, none of his family had been similarly affected.

At physical examination, the patient had normal facial features without thickening of the skin of the forehead. His clavicles were obviously thickened, especially at their medial ends. He had severe clubbing of all fingers and toes (Figure 1). The skin on his lower legs was notably thickened, scaly and blackened. The circumferences of his distal tibiae were visibly increased (Figure 1). He had no bone tenderness; all joints had a full range of motion and were without effusions.

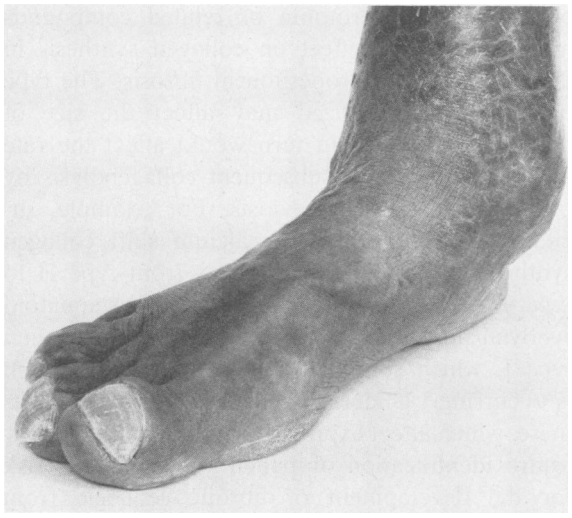

Figure 1.—Clubbing of toes in a 72-year-old man with idiopathic hypertrophic osteoarthropathy (pachydermoperiostosis).

Radiographic examination showed an extreme degree of periostitis in many bones, particularly the distal ends of the tibiae and fibulae (Figure 2), the proximal femora (Figure 3), and the distal radii (Figure 4). There was acro-osteolysis of the distal phalanges of the fingers (Figure 4) and toes (Figure 5). The radiographs showed that the periosteal new bone formation affected the midportion of the diaphyses as well as the epiphyses.

Discussion

Most patients with idiopathic hypertrophic osteoarthropathy develop normally until adolescence, when skin thickening and bone and joint deformities begin to occur. These changes progress for a number of years, then usually stabilize.[1-5] However, some patients, as did ours, may experience these changes at a much earlier age.[6]

The bony changes consist of symmetric, irregu-

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lar periosteal hypertrophy with new bone formation[3,4]; acro-osteolysis has also been reported.[7] These changes are most severe in the extremities[7] and can involve any bone, although the skull and vertebral column are rarely affected.[1,8] The articular surfaces are spared,[2,9] but intermittent swelling of joints and effusions are common; these are often moderately painful but may be asymptomatic.[1,10]

A considerable degree of painless clubbing usually results from proliferation of the soft tissues of the distal phalanges. This manifestation gives the digits a bulbous appearance, with the nails curving over the hypertrophied nail beds.[1]

Changes of the skin include generalized thickening (pachyderma), hyperhidrosis and seborrheic overactivity.[4] Eczema is also common.[11] The skin thickening and redundancy may become so excessive on the scalp and forehead as to cause a characteristic furrowing, termed cutis verticis gyrata.[1,2] The combination of thickened skin and bony enlargement can result in great thickening of the extremities, which is often the most striking physical finding.

On histological examination, involved bones

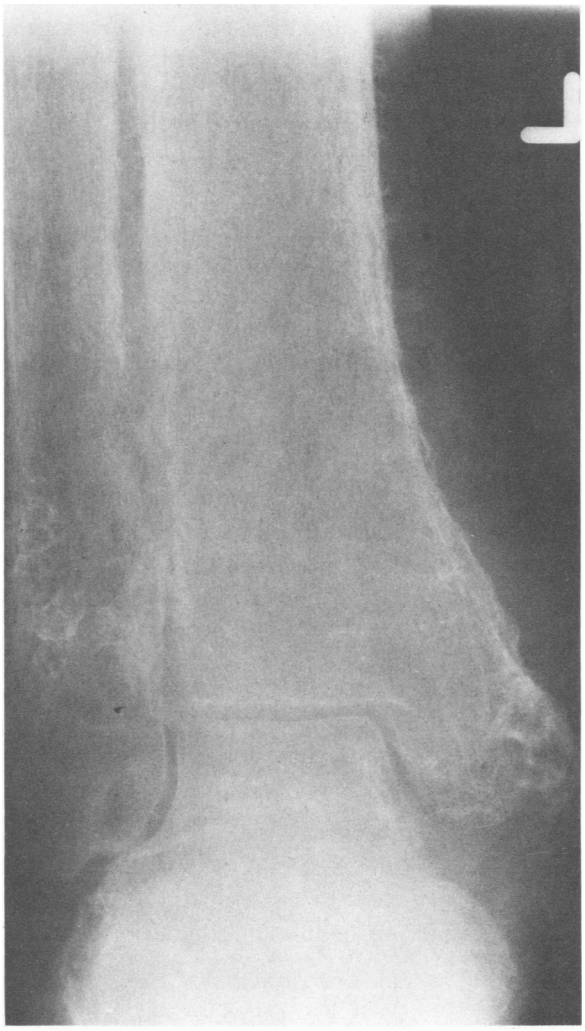

Figure 2.—Radiograph showing periostitis in distal ends of tibiae and fibulae.

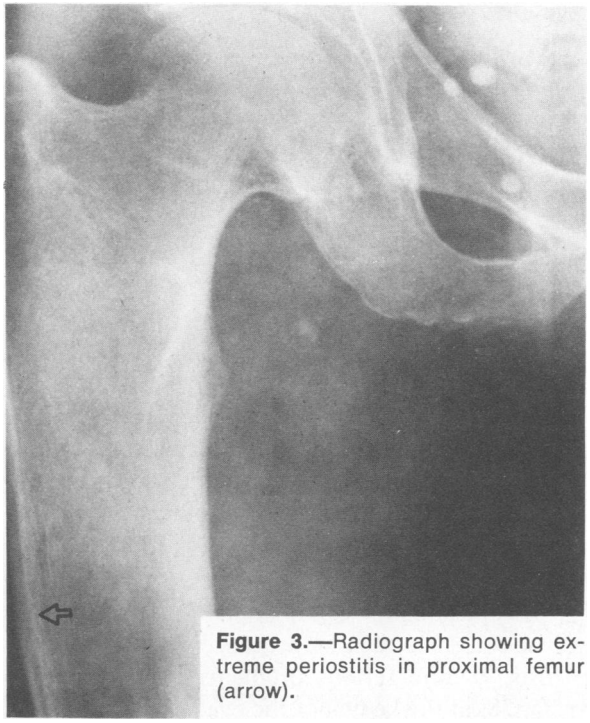

Figure 3.—Radiograph showing extreme periostitis in proximal femur (arrow).

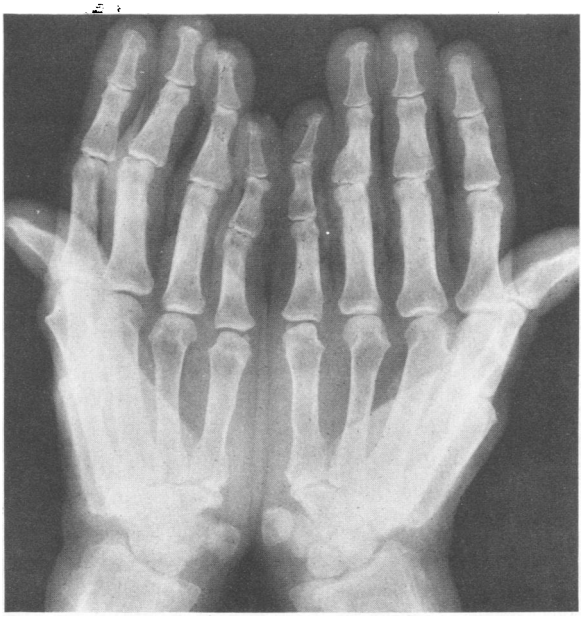

Figure 4.—Radiograph showing acro-osteolysis of fingers affecting the middiaphysis and epiphyses.

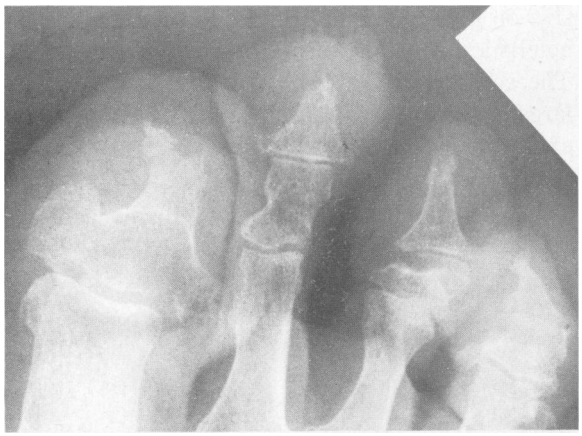

Figure 5.—Radiograph showing acro-osteolysis of toes.

show periosteal proliferation with round cell infiltration and new bone formation between the periosteum and the original cortex. This new bone later fuses with the old.[4] Hypertrophy of the nutrient arteries occurs.[1,4] Gradually the skin shows thickening and hyalinization of collagen and elastic fibers with interposed foci of necrosis and inflammation. The sebaceous and sweat glands become hypertrophied, as do the hair follicles.[4]

Most patients have only moderate discomfort from this disease and are able to lead normal lives, as did our patient. Although their main complaints often relate to their appearance and to hyperhidrosis,[2,4] more serious problems occasionally develop. Skin thickening of the eyelids may be severe enough to cause ptosis, which requires surgical correction.[1,8] Thickening of the skull has been considered responsible for a facial nerve palsy.[1] Finally, there are rare reports of bone marrow failure and extramedullary hematopoiesis attributed to destruction of the marrow by idiopathic hypertrophic osteoarthropathy.[12,13]

In 40 percent to 50 percent[2,4] of reported cases there is a familial history of idiopathic hypertrophic osteoarthropathy; family members, predominantly male, often had incomplete forms of the disease. Analysis of findings on studies of kindreds suggests that the condition may be inherited as an autosomal dominant trait with variable penetrance, which is most severe in males.[2]

Our patient's condition reflected many of these aspects of idiopathic hypertrophic osteoarthropathy. He had a great degree of periosteal new bone formation and clubbing, with notable thickening of the skin of his lower legs, although there was no hyperhidrosis or cutis verticis gyrata. The absence of progression of the disease in adulthood is typical.

Idiopathic hypertrophic osteoarthropathy should be compared with secondary hypertrophic osteoarthropathy, also known as pulmonary osteoarthropathy. The latter occurs as a complication of many diseases, especially thoracic infections and malignant conditions, but also of cardiac and gastrointestinal disorders and extrathoracic neoplasms as well.[10,11] The presenting features of pulmonary osteoarthropathy are similar to those of idiopathic hypertrophic osteoarthropathy, although the anatomic changes are usually less pronounced and the resulting discomfort more severe than in the idiopathic syndrome.[4] The two are usually differentiated by a history of early onset of the idiopathic hypertrophic osteoarthropathy, whereas pulmonary osteoarthropathy usually develops in association with a primary process. The osteoarthropathy, however, may precede clinical evidence of the underlying disease by several months.[11] A history of similarly affected family members may suggest idiopathic hypertrophic osteoarthropathy.

In contrast to the secondary form of the disease, which often improves with therapy of the underlying process,[11] there is no available therapy for the idiopathic hypertrophic osteoarthropathy. Fortunately, most patients with the latter condition have no more than moderate disability from their disease and have normal life spans.[1,5]

Summary

Idiopathic hypertrophic osteoarthropathy usually is a benign syndrome characterized by periosteal new bone formation, clubbing, swelling of joints and thickened, furrowed skin. Evaluation of a patient with this rare condition showed the clubbing and skin thickening, as well as radiographic changes in the distal ends of the tibiae and fibulae, proximal femora and distal radii, with acro-osteolysis of the distal phalanges of the fingers and toes. Clinicians can differentiate between idiopathic hypertrophic osteoarthropathy and secondary osteoarthropathy mainly by a history of early onset of the former, often with a positive family history, and the coexistence of pulmonary, gastrointestinal or neoplastic disease in the latter.

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Retroperitoneal Fibrosis as a Cause of Fever of Undetermined Origin

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A CONSULTING RHEUMATOLOGIST is sometimes asked to offer his expertise in seeking the cause of a multisystem illness that includes fever, arthralgias, myalgias, anorexia, weight loss, anemia and elevated sedimentation rate. Invariably, consultants in hematology, oncology, endocrinology, gastroenterology and infectious diseases have eliminated illnesses that would fall primarily within their domain. Almost as an afterthought, a rheumatologist is called in to "rule out vasculitis" or "exclude an evolving connective tissue disease." The present case report and discussion emphasize another entity whose prodrome can mimic a multitude of systemic illnesses. Though well described in the surgical literature, it merits more critical attention by internists and rheumatologists in their efforts to offer a more comprehensive differential diagnosis in the workup of a chronically ill patient.

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Report of a Case

A 21-year-old Marine was in excellent health until late October 1979 when a sensation of fullness in the right ear developed, followed by a temperature rising to 38.9°C (102°F), nonshaking chills, anorexia and fatigue. Studies were done at the medical clinic and a diagnosis of right-sided otitis media was made. Ampicillin and erythromycin were prescribed sequentially without improvement. His fever persisted accompanied by arthralgias of the elbows, wrists, knees and ankles as well as a retrobulbar headache without photophobia. Because the symptoms persisted, the patient was admitted to hospital at the Naval Regional Medical Center, Camp Lejeune, North Carolina, on November 5, 1979. On admission, a physical examination documented a temperature of 38°C (100.4°F). There were multiple, small, nontender posterior, cervical and supraclavicular nodes. Liver span was 12 cm and the spleen was soft, nontender and palpable 2 cm below the left costal margin. Humeroulnar joints and wrists were tender to palpation without obvious synovial thickening. Initial laboratory test results included a hematocrit of 30 percent, with a leukocyte count of 10,200 per cu mm and a normal differential. Platelet count was 178,000 per cu mm. Westergren sedimentation rate was 130 mm per hour. A peripheral blood smear was hypochromic. The following studies gave normal or negative findings: SMA-18, hepatitis-associated antigen, mononucleosis spot test, cold agglutinins, VDRL, Coombs direct and indirect tests, latex fixation, fluorescent antinuclear antibody (FANA) and analysis of the urine. Multiple blood cultures were negative. Purified protein derivative-standard (PPDS) and streptokinase-streptodornase (SKSD) skin tests were nonreactive. Additional studies included a bone marrow aspirate, which showed myeloid and megakaryocytic hyperplasia. A left posterior cervical node biopsy showed lymphoid hyperplasia. Adult Still disease was considered a reasonable possibility at this time, and high-dose salicylate therapy was initiated. The fever abated somewhat but the hematocrit decreased to 23 percent, with a hypo-