CASE REPORT

Primary hypertrophic osteoarthropathy

Sushil Ghanshyam Kachewar

Radio-diagnosis Department, Rural Medical College, Pravara Institute of Medical Sciences, Loni, Maharashtra, India

Correspondence: Sushil Ghanshyam Kachewar. Address: Rural Medical College, Pravara Institute of Medical Sciences, At-LONI Ta-Rahata, Dt-Ahmednagar, Maharashtra, India. Telephone: 91-242-2271-307. Fax: 91-242-2271-529. E-mail: sushilkachewar@hotmail.com

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Abstract

A rare case of 25 years male who had intermittent swelling over the ankles as well as knees, for the past 7 years is presented here. This case report not only enables diagnosing primary hypertrophic arthropathy, by systematically excluding other differentials but also brings out essential differences between primary and secondary forms of this entity.

Key words

Hypertrophic arthropathy, Imaging findings, Primary or secondary forms

Introduction

Primary hypertrophic osteoarthropathy (PHOA) is a rare familial disorder clinically manifesting as elephant like thick skin (pachydermia) and radiologically as symmetric periostitis of limbs. Proper diagnosis is a must to decide whether and what treatment would be effective. A rare case of 25 years male who had intermittent swelling over the ankles as well as knees, for the past 7 years is presented here. This case report not only enables diagnosing primary hypertrophic arthropathy, by systematically excluding other differentials but also brings out the essential differences between primary

and secondary forms of this entity. This differentiation is a must as sometimes the secondary form may be the only visible manifestation of an underlying grave disease.

Case report

A 25 years male presented with intermittent swelling over the ankles as well as knees, for the past 7 years. It was insidious in onset and gradually progressive. Clinical examination revealed clubbing of digits of both extremities. It was told to be present since childhood and was increasing in severity (Figure 1). His father also had clubbing of digits of both extremities. No other local or systemic abnormalities were found. Rheumatoid antigen, C reactive protein and anti streptolysin O were negative and serum uric acid was within normal limits. There was absence of thickening of the facial skin and scalp. The patient had no respiratory, cardiovascular or gastrointestinal symptoms.

Sonography of the abdomen and pelvis revealed no abnormality. Radiographs of both extremities showed shaggy irregular periosteal reaction and cortical thickening involving diaphysis and metaphysis of tibia and fibula (Figure 2). Radiographs of chest, skull and hands were unremarkable. Plain CT of the legs showed diffuse periosteal reaction, subperiosteal new bone formation and cortical thickening (Figure 3).

Clinic-radiological findings, made us to put forth the diagnosis of the incomplete form of Primary Hypertrophic Osteoarthropathy, after excluding the secondary form of hypertrophic osteoarthropathy and other differentials.

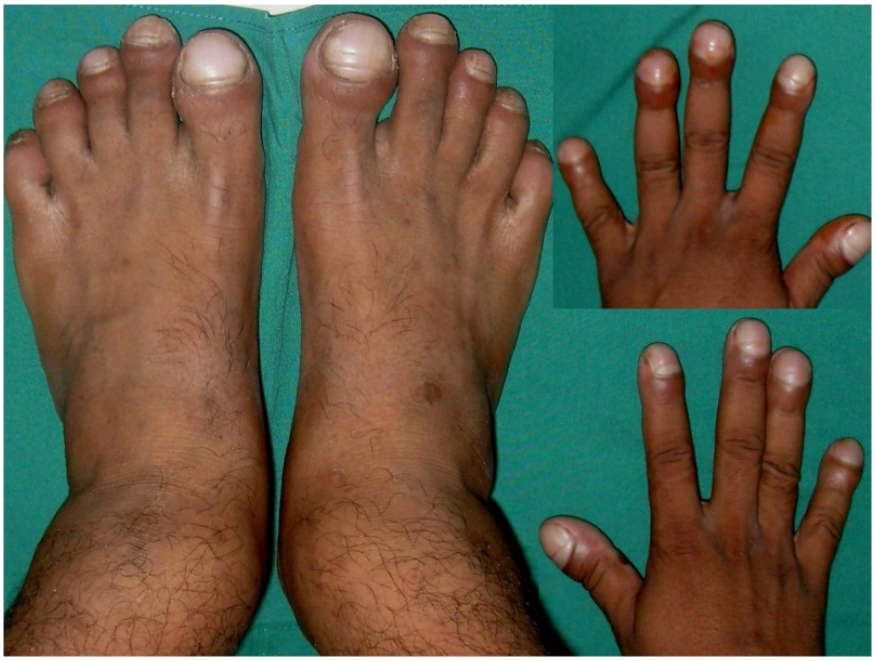

Figure 1. Clubbing of digits of both extremities in a 25 years male

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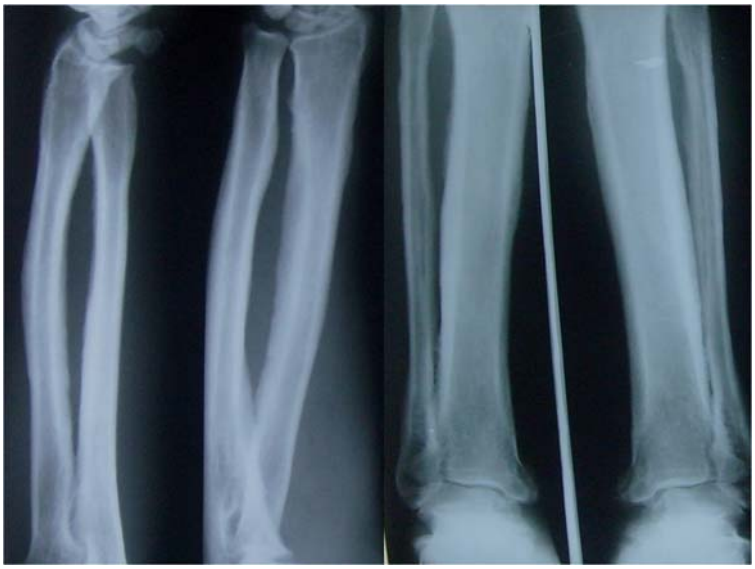

Figure 2. Radiographs of radius-ulna and tibia-fibula show periosteal reaction and cortical thickening involving diaphysis

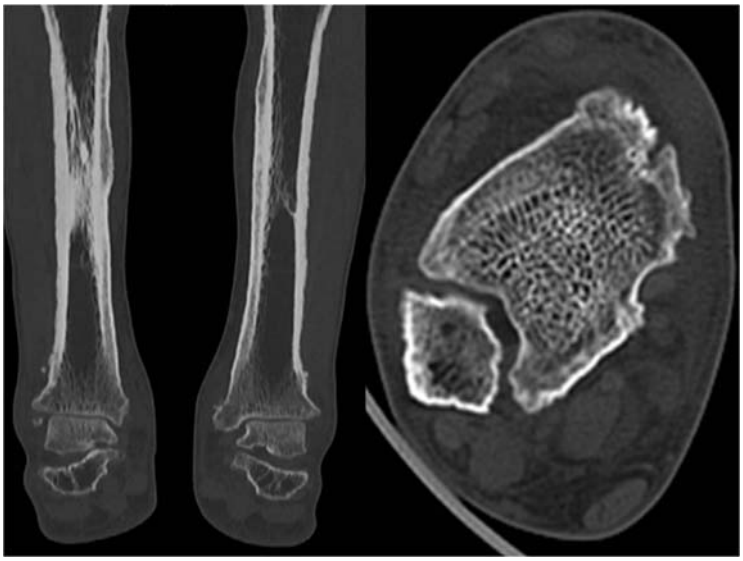

Figure 3. Plain CT scan coronal reformatted images and axial image of the legs shows diffuse periosteal reaction, subperiosteal new bone formation and cortical thickening

Discussion

Primary hypertrophic osteoarthropathy (PHOA) or Pachydermoperiostosis is a rare hereditary disorder that was first described in 1868. It is characterized by clubbing of digits, pachydermia and periostosis. Pachydermia refers to abnormal skin thickening, like that of a pachyderm (an elephant, rhinoceros or hippopotamus).

PHOA has 3 forms: (A) complete form with pachydermia and periostitis, (B) incomplete form with evidence of bone abnormalities but lacking pachydermia, and (C) forme fruste with prominent pachydermia and minimal to none skeletal changes [1].

PHOA is a familial disorder, and the precise incidence is unknown. The male to female case ratio is approximately 7:1. The condition is believed to be inherited in an autosomal dominant pattern with variable penetrance. Father of the patient described in this report also had similar history.

The predominant radiographic feature of PHOA is periostitis, which is depicted as symmetric osseous thickening. Periostitis mostly affects the tubular bones of the limbs, especially the radius, ulna, tibia and fibula. Periosteal proliferation is usually shaggy and is associated with irregular bony excrescences and diphyseal expansion, which is seen in this case [2]. Acro-osteolysis has been reported to be associated, in some patients with this syndrome [3]. However these findings were absent in this case. Also as there was no pachydermia, incomplete form of PHOA was kept as the diagnosis.

The secondary form is most commonly seen secondary to benign as well as malignant cardiopulmonary and gastrointestinal diseases [2, 4, 5]. Although it is mostly seen in thoracic malignancies like non-small cell lung cancers and Phylloides tumors, several non-malignant conditions of lungs, gastrointestinal system and cardiovascular system can also result in it. The essential differences between the primary as well as secondary forms are summarized in Table 1.

Table 1. Important differences between the primary as well as secondary forms

Sr No.	Criteria	Primary Hypertrophic Osteoarthropathy	Secondary Hypertrophic Osteoarthropathy
1	FamilyHistory	Positive	Negative
2	Genetic Link	Chromosome 4q33–q34	None
3	Presents in	Children or young adults (early)	At any age (usually later)
4	Associated with	No other pathologies	Neoplasms or infectious diseases
5	Onset	Insidious	Sudden if associated with neoplasms Insidious if associated with infections
6	Progression	Gradual	Rapid if associated with neoplasms Slow if associated with infections
7	Joint Pains	Usually none / less common	More common and severe
8	Changes in epiphyses	More common	Less common
9	Bony outgrowths in affected bones	Poorly defined	Well defined
10	Treatment	Symptomatic	Removal of underlying cause

Table 2. Diagnosis of incomplete form of Pachydermoperiostosis

Reference No.	Family history	Age in years at presentation	Duration of Illness in years	Most distressing clinical feature
6	None	45	30	Polyarthralgia, mainly in the wrists, knees and ankles
7	None	28	10	Pain in both knees
8	None	31	5	Swelling and pain in the knees, ankles and small joints of the hands
9	None	40	Not mentioned	Swelling of the gums and at the wrist, ankle, and knee joints; and thickening and folding of the facial skin.
10	None	25	1	Joint swelling and pain Purulent Scalp lesions with multiple tufts of hairs emerged from single openings
Current Case	Present	25	7	Swellings over knees and ankles

In this case there were no other associated systemic diseases. Although similar osseous changes may occur in both primary and secondary hypertrophic osteoarthropathy, bone changes in PHOA more commonly extend into the epiphysis. Involvement of the epiphyseal region distinguishes it from the secondary form, in which the epiphyses are usually spared [2].

Other differentials to be considered are diffuse idiopathic skeletal hyperostosis (DISH), thyroid acropachy and Hypervitaminosis A. Patients with DISH occasionally have diphyseal periostitis, particularly in the femur and humerus. Occasionally, the metacarpals are involved; this finding mimics that of PHOA. Periostitis in thyroid acropachy appears fluffy and speculated. It mostly affects the periosteum of the bones in the hands and feet and is rarely seen elsewhere. Hypervitaminosis A may cause periosteal proliferation, but the clinical and radiographic features enable one to distinguish this disease from PHOA. Periostitis is usually diphyseal, with undulating contour; raised intracranial tension, soft tissue nodules, painful ostitis which usually affects lower limb epiphyses and ultimately leads to premature epiphyseal closure so that there is decreased height of the affected individual.

Thus in this case as discussed, the diagnosis of incomplete form of Pachydermoperiostosis is the diagnosis by exclusion as no confirmatory gold standard test is available. As shown in table 2, the case reported here is unique in the form that there was swelling only over the joints of lower extremities. Moreover, a similar history was present in his father for a long duration.

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