Pachydermoperiostosis

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Introduction

Pachydermoperiostosis or primary hypertrophic osteoarthropathy (HOA) is a rare hereditary disorder, characterized by digital clubbing, pachydermia and periostosis. This case report is of incomplete form of HOA where patient presented with digital clubbing and periostosis. Other radiological differentials for periostosis are also discussed in this report.

Case Report

A 25 year old male presented with intermittent pain and swelling of ankle and knee joints of both sides for last seven years. Swelling was gradually increasing around the joints. Clinically, patient had clubbing of nails of both extremities. Clubbing was present since childhood and was progressively increasing in severity (Fig. 1). 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.

Ultrasound examination of abdomen and pelvis were normal. Radiographs of both extremities showed shaggy irregular periosteal reaction and cortical thickening involving diaphysis, metaphysis and epiphysis of tibia and fibula. At the epiphysis of tibia, shaggy ill-defined bone excrescences were also seen (Fig. 2a, 2b). Radiographs of chest, skull and hands were unremarkable. Plain computed tomography (CT) of the legs showed diffuse periosteal reaction, subperiosteal new bone formation with cortical thickening (Fig. 3a, 3b, 3c).

Based on clinical and radiological findings, diagnosis of the incomplete form of pachydermoperiostosis was given. This diagnosis was made after excluding the secondary form of hypertrophic osteoarthropathy and other differentials.

Discussion

Pachydermoperiostosis or primary HOA is a rare hereditary disorder that was first described in 1868. It is characterized by digital clubbing, pachydermia and periostosis. Pachydermoperiostosis has 3 forms: the

complete form with pachydermia and periostitis, an incomplete form with evidence of bone abnormalities but lacking pachydermia and 'forme fruste' with prominent pachydermia and minimal-to-absent skeletal changes [1].

Pachydermoperiostosis 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.

The predominant radiographic feature of pachydermoperiostosis 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 diaphyseal expansion, which is seen in this case [1]. The pelvis, carpus, tarsus, metacarpals, metatarsals and phalanges may also be involved. Rarely, thickening of the calvarium and skull base is seen. Acro-osteolysis has been associated in some patients with this syndrome [2]. However these findings were absent in our case. Also, as there was no pachydermia, diagnosis of incomplete form of pachydermoperiostosis was put forth.

The secondary form results from cardiopulmonary, hepatic, gastrointestinal diseases and certain malignancies [1,3,4]. In this case there were no other associated systemic diseases. Although similar osseous changes may occur in both primary and secondary hypertrophic osteoarthropathy, periostitis in HOA more commonly extends into the epiphysis. Involvement of the epiphyseal region distinguishes it from the secondary form, in which the epiphyses are usually spared [1].

Other differentials to be considered are diffuse idiopathic skeletal hyperostosis (DISH), thyroid acropachy and hypervitaminosis A. Patients with DISH occasionally have diaphyseal periostitis, particularly in

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Fig. 1 : Photograph of patient showing severe clubbing of the nails of both extremities with swelling around the ankle joint.

Fig. 2 : Anteroposterior and lateral radiographs of both legs (a) and both forearms (b) show shaggy irregular periosteal reaction involving epi-meta-diaphysis with expansion of tibia in diaphyseal region.

Fig. 3 : Axial (a) and reformatted coronal (b, c) CT images of both legs showing diffuse periosteal reaction with subperiosteal new bone formation and cortical thickening.

the femur and humerus. Occasionally, the metacarpals are involved; this finding mimics that of HOA. Periostitis in thyroid acropachy appears fluffy and spiculated. It mostly affects the periosteal bone 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 HOA. Periostitis is usually diaphyseal, with undulating contour; it is often associated with epiphyseal abnormalities and soft tissue nodules [5].

Thus in this case, the diagnosis of incomplete form of pachydermoperiostosis was a diagnosis by exclusion as no confirmatory gold standard test is available.

Conflicts of Interest

None identified

References

1. Resnick D. Enostosis, Hyperostosis and periostitis: In: Resnick D, Kransdorf MJ, editors. Bone and Joint imaging. 3rd ed. Philadelphia: Elsevier Saunders; 2005; 1433-5.
2. Joseph B, Chacko V. Acro-osteolysis with hypertrophic pulmonary osteoarthropathy and pachydermoperiostosis: Radiology 1985; 154: 343-4.
3. Kowalewski M, Urban M, Gorska A. Familial occurrence of primary hypertrophic osteoarthropathy: A case report. Med Sci Monit 1997; 3: 229-34.
4. Bhaskaranand K, Shetty RR, Bhat AK. Pachydermoperiostosis: Three case reports. J Orthop Surg 2001; 9: 61-6.
5. Khan NA, Mohammed JA, Dare MS, Ian T, Macdonald S. Hypertrophic osteoarthropathy. Updated on 2008. Available from: <http://www.emedicine.com>.