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A RARE CASE OF PRIMARY HYPERTROPHIC OSTEOARTHROPATHY : CASE REPORT

Paediatrics

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ABSTRACT

BACKGROUND: Hypertrophic osteoarthropathy (HOA) is characterized by abnormal growth of skin and osseous tissue at distal parts of the extremities, also known as orphan syndrome and Pierre Marrie-Bamberger syndrome. The peculiar bulbus deformity of the digital tips known as "clubbing", periosteal proliferation of long bones and painful effusion of synovium.[2] HOA can be primary(idiopathic) and secondary. Although HOA is rare, secondary HOA is more common in certain diseases like chronic pulmonary disease, congenital heart disease, gastrointestinal disease, and malignancies which may improve with treatment of the underlying disease.[5] On the other hand primary HOA is idiopathic and its pathophysiology remains unclear. When idiopathic HOA is associated with pachyderma (thickened skin) is known as pachydermoperiostosis. In extremely rare instance idiopathic HOA may occur without pachyderma.[4]

CLINICAL DESCRIPTION: We report a case of 4 years old female child came with complaints of pain and swelling in bilateral knee and ankle joints since 2 years, and developed pain and swelling in elbow and wrist joint since 1 year. On general examination she has grade 4 clubbing in fingers and toes. Suspecting HOA, relevant investigations sent to rule out secondary causes of HOA. After ruling out all causes of secondary HOA, we arrive at diagnosis of primary (idiopathic) HOA.

MANAGEMENT: As there is no definitive management for primary HOA, we managed our patient symptomatically with painkillers.

CONCLUSION: Reporting a rare case of primary HOA without pachyderma treated symptomatically with NSAIDS.

KEYWORDS

Hypertrophic osteoarthropathy, Pchyderma, Pachydermoperiostosis, clubbing, orphan syndrome.

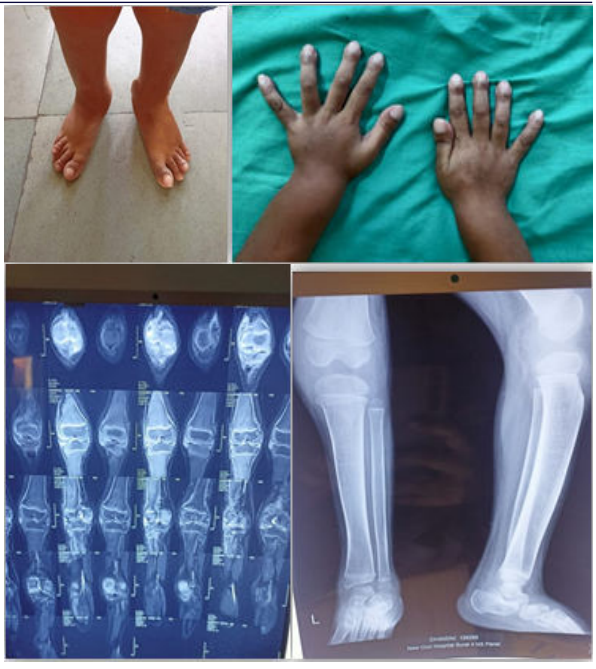
INTRODUCTION:

Pachydermoperiostosis or primary hypertrophic osteoarthropathy (HOA), was described by Friedreich in 1868, is a hereditary disease and a rheumatologic condition in which the patient presents with digital finger clubbing, enlarged extremities secondary to bone and periarticular tissue proliferation, joint pain and edema, bilateral eyelid ptosis, periostosis, leonine face, and skin thickening (pachyderma), coexisting with a variety of clinical manifestations including hyperhydrosis, arthritis, cutis verticis gyrata, joint pain, edema, and hypertrophic gastritis.[6] The ratio of the disease in male:female is 8:1. It has genetic aggregation in 25% to 38% of cases, being mainly autosomic dominant in nature. It is classified as either primary (pachydermoperiostosis) or secondary. The primary, or idiopathic, form is considered rare, appearing in 3-5% of all cases. Here, we describe the clinical and radiological manifestations of a patient with the primary form of hypertrophic osteoarthropathy, focusing on the therapeutic challenge of refractory joint pain.

Case Description:

A 4 year old female child presented with bilateral knee and ankle pain and swelling since 2 years and also develops pain and swelling in bilateral elbow and wrist since 1 year. When she was 2 years old her mother noticed peculiar shape of finger and toe nails, with the time her anomaly persisted and progressed. No associated skin changes noted. On physical examination she was well nourished and well developed with weight of 15kg and height of 93cm. examination of skull, chest and abdomen show no abnormality.

Grade 4 clubbing in all digits present with swelling of soft tissues of bilateral knee, ankle, elbow and wrist with bony anomalies in the form of bowing of knees (genu varus). All haematological parameters including serum calcium, vitamin D3, RA factor, ASO titre, CRP, peripheral smear for microfialria, ALP, PTH, and ABGA found to be normal. Electrocardiogram and 2d echo are also normal. On radiological evaluation lungs, heart and mediastinum are normal on CXR PA view. Xray bilateral wrist and knee joint suggestive of smooth solid periosteal thickening involving long bones of upper and lower limbs in diaphyseal region. Long bones appear tubular with loss of normal diaphyseal concavity in metacarpals. Soft tissue swelling noted involving bilateral knee and wrist joint.



However, no evidence of erosion noted. No evidence of any metaphyseal cupping, splaying or fraying noted. Overall changes suggestive of possibility of primary hypertrophic osteoarthropathy. MRI of both knee joints with distal femure and proximal leg suggestive of mild periosteal thickening with thin circumferential subperiosteal fluid and subtle bonemarrow edema surrounding visualized distal femure, proximal tibia and fibula bilaterally. Overall findings suggestive of primary HOA in absence of other underlying conditions. Patient is treated symptomatically with NSAIDS (tab neproxene) effective in alleviating pain. Patient is currently under follow up for disease process.

DISCUSSION:

Primary HOA is characterized by hereditary predisposition where one

third of patients having close relative with same illness. In our case, patient's elder sister has features involving clubbing of all digits. Primary cases of HOA prone to show more disseminated skin hypertrophy known as pachydermoperiostosis which is absent in our case and it is rare. Primary HOA without pachyderma may be familial or sporadic.[4] Primary HOA is diagnosed based on radiological features consisting symmetrical and pronounced periosteal new bone proliferation in long tubular bones, earliest noticed in distal third of both legs and forearms. The cortex is thickened with presence of marrow edema. The diaphysis of bone is the most common site. These findings are not seen in secondary HOA.[3] Demonstrable involvement of carpus, tarsus, vertebra and skull is rare. Laboratory studies have no positive aid in diagnosis of primary HOA, although help to rule out secondary causes. Aside from its unsightly appearance, clubbing is normally asymptomatic and requires no treatment. In most situations, NSAIDs are useful in relieving pain in people with painful osteoarthropathy.[3] A recent study found that after NSAID treatment, clubbing and pachyderma were reversed in the major HOA case series (Etoricoxib) treatment. Etoricoxib is a prostaglandin inhibitor with a high potency. Intravenous bisphosphonate infusions are beneficial in HOA patients with recurrent fractures.[3]

CONCLUSION:

We presented a rare case of primary HOA without pachyderma presented with joint pain and swelling and clubbing, was treated with NSAIDS, with patient responding well to the treatment.

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