CASE REPORT

Primary hypertrophic osteoarthropathy

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Abstract

We describe two cases of familial primary hypertrophic osteoarthropathy. In this family, brother and a sister were affected with history of similar features in father.

Keywords: Primary Hypertrophic Osteoarthropathy (PHOA), Clubbing of the digits.

Introduction

Primary Hypertrophic Osteoarthropathy (PHOA) is a clinical syndrome characterised by clubbing of the digits, enlargement of the extremities and synovial joints effusions associated with pain and swelling. This rare condition occurs in the absence of any other disease involving other systems of the body. It is usually familial, accounts for 3 to 5% of all cases of HOA and has a painless, chronic course.[1] This disease is inherited as an autosomal dominant trait with variable expression. It usually begins insidiously at puberty and is nine times more common in males as compared to females. Although a familial condition there are only few families reported with this condition. Here we report a family affected with this rare condition.

Keywords: Primary hypertrophic osteoarthropathy, Clubbing, Synovial joints effusions.

Case Reports

Case-1

A thirty-eight years old Kashmiri man was seen with twelve years history of pain and swelling of multiple joints. His problems began at the age of twenty five years when he complained of occasional arthralgia in multiple joints especially after prolonged walking, playing football, strenuous work or exercise. He gave a history of profuse sweating of palms and soles and noticed progressive enlargement and widening of fingers, hands, wrists, feet, toes and ankles joints. He was concerned about the abnormal appearance of his hands and feet and also noticed thickening of skin on face and scalp and coarse features.

His parents were first-degree cousins and consanguineous marriages are common in the family. He gave a family history of identical condition affecting his father and elder sister who had similar appearance of hands, fingers and toes.

Physical examination showed advanced clubbing of all fingers, more severe in thumbs, enlargement of hand and wrist joints (Figures-1 and 2). There was no pain or restriction of movements of small joints of the hand. There was clubbing of toes, widening of feet and ankles (elephant feet) and knee joints. There was thickening of forehead folds with evident pachydermia. The other notable features were palmo-plantar hyperhidrosis and cutis verticis gyrata. There was no effusion of knee or ankle joints.

He had stage 1 hypertension. Rest of the cardiovascular examination was normal. There were no other physical findings.

Laboratory investigations showed Haemoglobin of 14.6gm/dl, normal differential counts. ESR. Urea, Creatinine, LFTs and lipid profile were all normal.

X-rays showed exuberant periosteal reaction along the distal meta-diaphyses of ulna, radius, tibia and fibula. There were no other associated lesions or erosions. The joint spaces were within normal range. There was clubbing and tufting of terminal phalanges of both hands (Figure-3). X-ray of knee joints showed marked irregular shaggy periosteal reaction along distal femur and proximal tibia and fibula extending up to epiphyseal area. There were early degenerative changes of left knee joint, with considerable soft tissue swelling and evidence of small effusion in suprapatellar region. There were no bony erosions or destructive lesions and joint spaces were normal. These findings were suggestive of patchy-dermo periosteosis. Chest X-ray was normal.

Tc-99m bone scan showed increased patchy tracer uptake involving shafts and distal ends of radius and ulna bilaterally. There was increased tracer uptake in both ends of femorii, tibia and fibula extending into the adjacent part of the shafts. There was increased tracer accumulation at wrist joints and on radial aspect of distal phalanx of thumb and toes.

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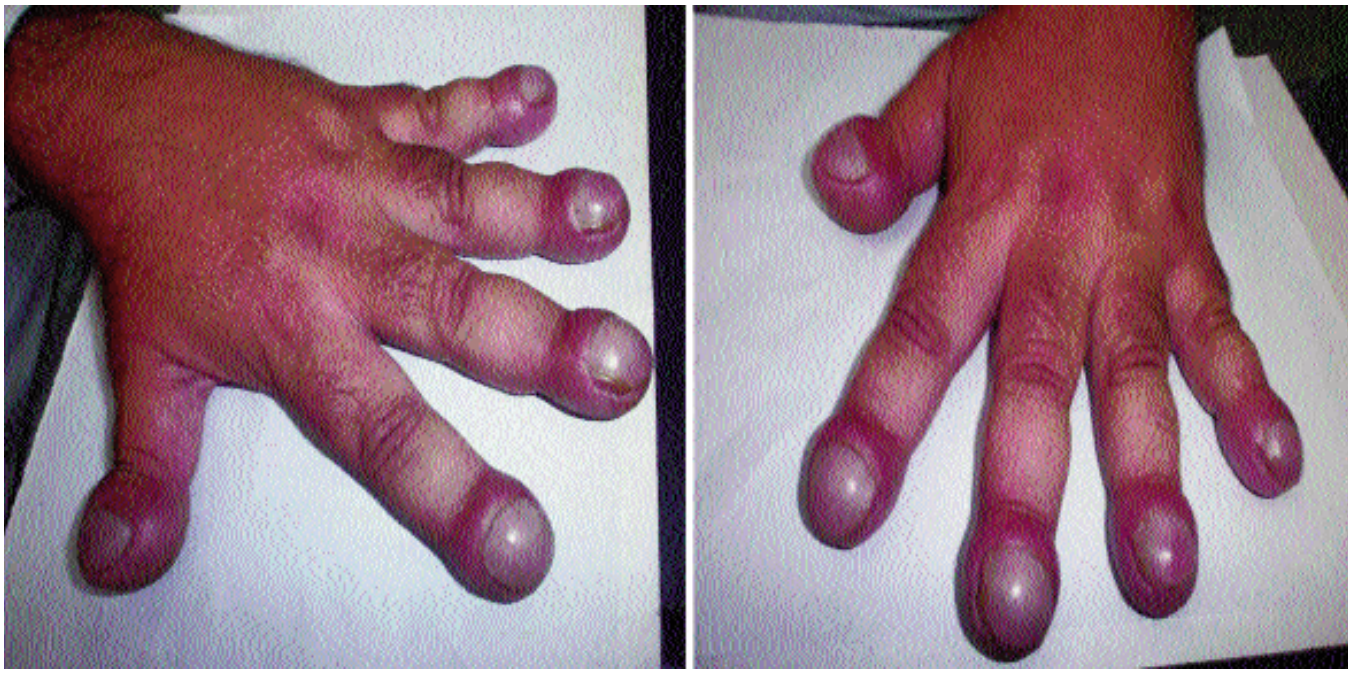

Figure-1 & 2: Clubbing of digits.

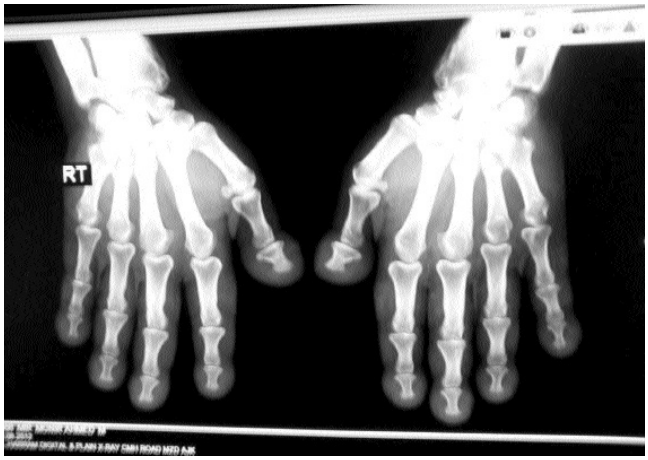

Figure-3: Radiograph of hands.

Case-2

A 44 years old, elder sister of our patient was called for examination. She was asymptomatic and showed little concern about her appearance. Her examination showed clubbing, enlarged distal ends of fingers and toes and wide hands. The rest of physical examination was normal. Her X-rays of hands revealed clubbing and tufting of terminal phalanges. The Blood C/P, Blood sugar, urea, creatinine, LFTs and lipid profiles were normal.

Discussion

Primary hypertrophic osteoarthropathy (PHOA) is a rare condition. It occurs in the absence of any disease involving other systems of the body. It is usually familial, accounts for 3 to 5% of all cases of HOA and has a painless, chronic course.[1] This disease is inherited as an autosomal dominant trait with variable expression. There are also few case reports of an X-linked transmission. It usually begins insidiously at puberty and is nine times more common in males as compared to females.[2]

Although a familial condition there are few families reported with this condition. Gómez Rodríguez N and colleagues described a family in which two brothers were affected with PHOA.[3] Diren HB reported a family with five members suffering from this disorder.[4] Our patient and his family lives in the area of low mountains in Himalayan region. These families live within their own relatively closed social setup. They have consanguineous marriages and other familial disorders are common in these families.

Our patient has a strong familial background. His father died in old age and according to him he had similar appearance of hands with severe clubbing. The elder sister is suffering from the same condition.

PHOA is a syndrome of three major features comprising of clubbing of the fingers and toes, periostitis of the long bones, and secondary arthritis.[5]

Clubbing of digits is the most prominent clinical feature of this condition. It is the increased vascularity and increased connective tissue growth in the nail bed that produces the characteristic features of clubbing.[6] New bone formation is the pathological hallmark of PHOA. It is due to neo-angiogenesis and osteoblast proliferation in the distal tubular bones that leads to sub-periosteal new bone formation. This new bone formation is more prominent along the distal diaphysis of tubular bones and extends proximally over time. As mineralization takes place, a cuff of new bone may be formed around the distal end of long bones.[7] The most common involvement of skin is in the form of skin thickening or pachyderma.

In PHOA the onset of the disease is insidious, gradually progressive while patient remains asymptomatic. These patients are usually diagnosed when they present with some other complaint.[8] The skeletal involvement may cause dull pain in hands and distal extremities. With the onset of secondary osteoarthritis, symptoms may become more severe as happened in our patient.[9] PHOA usually develops in adolescence and activity persists during the growth period. The progression of disease ceases after first decade and adults tend to become asymptomatic. However, dermo-skeletal changes persist for the rest of life.

The diagnosis of PHOA is based on typical clinical features of clubbing of digits and distal periostitis of long bones associated with pachydermia. The distinguishing radiological features are new bone formation leading to periosteal thickening along the distal diaphysis of tibia, radius, ulna, fibula and femur. Over time, it extends proximally to involve diaphysis and metaphysis of these bones. Radionuclide bone scan using technetium Tc 99m shows increased uptake of the tracer in the periosteum in the distal ends of long bones.

The medical treatment of HOAP is symptomatic. The main aim of treatment is to relieve pain with NSAIDs, corticosteroids, tamoxifen citrate, retinoids, risedronate and Colchicine. There are some reports of effectiveness of pamidronate and octreotide in the management of pain relief.[10]

This condition is associated with significant morbidity according to the severity of the disease however prognosis is excellent. All patients need reassurance about good prognosis.

Conclusion

Primary hypertrophic osteoarthropathy is a rare familial disorder. There are individual case reports in literature with no history of family involvement. We reported a family with living healthy siblings with this condition. Further evaluation and genetic testing can provide valuable insight in this rare heterogenic disorder.

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