A Case Of Pseudoacromegaly

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Citation

A Biswas, P Sahana. *A Case Of Pseudoacromegaly*. The Internet Journal of Endocrinology. 2009 Volume 6 Number 1.

Abstract

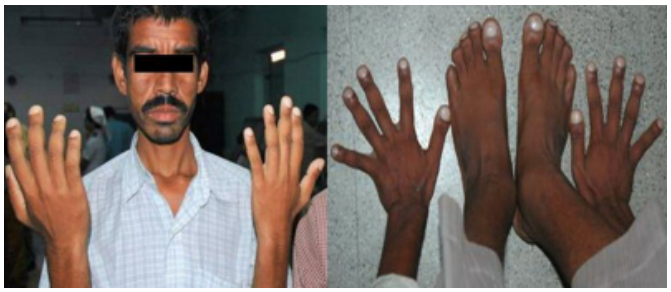
We report a single case primary pachydermoperiostosis in this report. The patient is a young male who presented with progressive enlargement of the hands & feet along with hyperhydrosis in absence of macrognathism and macroglossia.IGF1 level was found to be normal but x rays showed acroosteolysis with periosteal reaction indicating the diagnosis. This is a rare condition often mimicking some other well known conditions like acromegaly and the leonine facies seen in leprosy. This case is unusual because of the lack of a family history and the late age of presentation.

CASE REPORT

A 30-year-old male, non-smoker and non-alcoholic presented to us with complaints of progressively painful enlargement of hands and feet for last 4 years. It was accompanied by excessive sweating of the face and extremities with thickening and furrowing of the skin over the forehead for last 2 years. He also complained of pain in multiple joints (knees, ankles and the wrists). There were prominent vertical furrows over the forehead and ridges over the scalp (cutis vertices gyrata) and enlargement of hand, feet, fingers and toes (Figure 1).

Figure 1

Figure 1 shows enlargement of hands and feet with digital clubbing; thickening and coarse texture of skin with furrowing of facial skin is seen



Clubbing was present in all fingers and toes. Digital X-rays showed symmetric subperiosteal new bone formation with acroosteolysis of distal phalanges of both feet (Figure 2, 3).

Figure 2

Figure 2 shows symmetric periosteal reaction at the lower end of the tibia with cortical thickening.

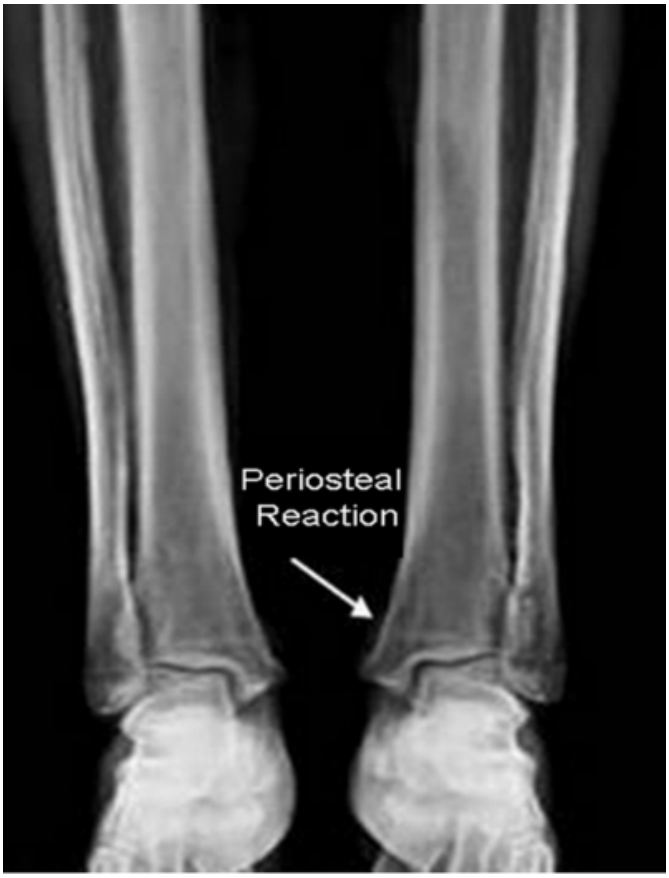
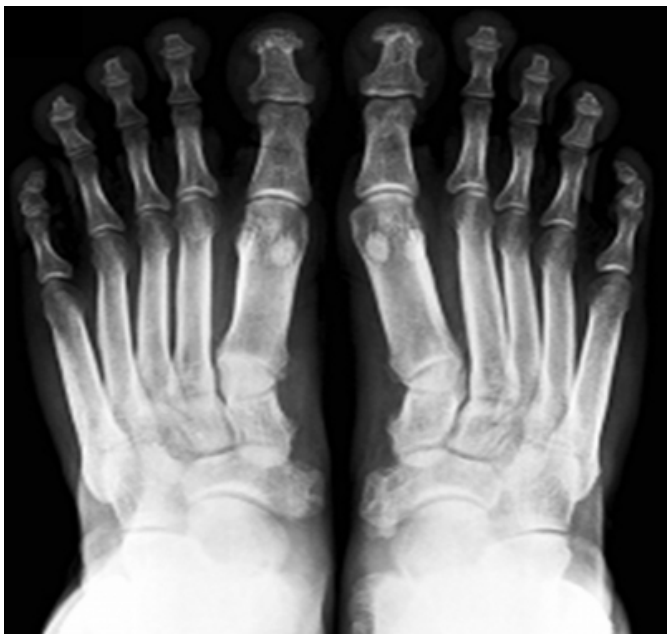


Figure 3

Figure 3 shows erosion of the distal phalanges of both feet (acroosteolysis)



Serum IGF1 (age and gender matched) was found to be within the normal range. CT scan of brain revealed a normal sella turcica. The chest X ray was also normal. Contrast enhanced CT imaging of the chest did not reveal features of malignancy, bronchiectasis or lung abscess. Rapid plasma regain was negative. Serum calcium, phosphate, sodium, potassium and alkaline phosphatase levels were found to be within normal range. Thyroid hormone levels, serum creatinine and urine studies were also normal. A family history of similar condition could not be elicited; neither was there any history of consanguinity in the family.

DISCUSSION

Primary pachydermoperiostosis (Syn. Touraine- Solente-Gole syndrome) is a rare developmental defect that occurs predominantly in males. The first case was described by Friedrich in 1868[1].The disease is characterized by thickening of skin of the forehead, scalp and thickening of the phalanges and bones of the limbs producing spade like hands and feet and clubbing of the fingers and toes[2,3]. Our patient had cutis vertices gyrate which is so called due to its similarity between the markedly thickened and furrowed facial skin on the forehead seen in this condition and the surface of the brain. The association of cutis verticis gyrata and pachydermoperiostosis is sometimes referred to as Tourane-Solente-Gole syndrome[4].The differential diagnoses considered for such a presentation include:

Secondary causes of HOA like malignancies, Rosenfeld-Kloepfer syndrome which is a variant of pachydermoperiostosis and is characterized by enlargement of the mandible and/or the maxilla; large hands, feet, nose, lips, and tongue; prominence of the upper part of the forehead; cutis verticis gyrata; and corneal leukoma.

Currarino idiopathic osteoarthropathy is a juvenile incomplete form of pachydermoperiostosis characterized by eczema and wide cranial sutures

Thyroid acropachy, which is usually painless is another differential diagnosis for this condition. Syphilitic periostitis,endosteal hyperostosis (van Buchem disease), macrodystrophia lipomatosa complete the list. Radiology usually shows significant periosteal thickening in the metacarpals and the long bones[5,6]. Acroosteolysis has also been reported in the literature[3] as seen our case particularly in the third and fourth digits of both feet. Proliferative periostitis of leg bones especially the diaphyses of the tibia, fibula, radius and ulna leads to increasing circumference of the affected bones without increase in their length.

Skin biopsy reveals hyperplasia with thickening of subcutaneous collagen and deposition of PAS positive material. Alteration in fibroblast activity with deposition of collagen is thought to be the underlying cause[7]. Studies have also shown dysregulated production of matrix molecules with deposition of decorin protein molecules[8]. Platelet growth factors may play a role in stimulating the fibroblasts and growth factors like epidermal growth factor, platelet derived growth factors are found in increased levels in the body[9].

Skin and bones changes become progressively severe for 5-10 yrs and then remain unchanged throughout life[2,5]. Pamidronate and tamoxifen have been described as being effective in treating painful osteopathy along with vagotomy in severe cases[10,11,12].Few others have used colchicine with improvement in articular symptoms, folliculitis and regression of thickened skin[13] . Isotretinoin has also been suggested as a treatment for skin the manifestations seen with this condition[14].Approximately 40% of cases have a family history of similar disease and transmission is usually autosomal dominant with variable expressivity[15]. Other more recent studies have also supported an autosomal recessive and X linked mode of transmission[16].This patient probably represents a new mutation or an autosomal recessive form of disease.

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