Ind J Dermatol Venereol Leprol 1994; 60 : 369-71

PACHYDERMOPERIOSTOSIS : THE DISEASE, AND ITS DISFIGUREMENT

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We describe 2 patients of pachydermoperiostosis with the "complete" form of the disease. We report these cases to highlight the clinical features of this rare disorder; and discuss certain social problems related to its disfiguring consequences.

Key Words : Pachydermoperiostosis, Leonine face

Introduction

Pachydermoperiostosis or idiopathic hypertrophic osteoarthropathy is an uncommon disorder, with a presumably autosomal dominant inheritance.[1] The disorder is predominant in males and has it's onset in late adolescence.[2] Variations in the phenotypic expression results in clinical features ranging from mild to severe; and in a given case the skin, soft tissues, and bones may be involved independently or together. Touraine and Solente, who first characterised pachydermoperiostosis, distinguished its three forms (1) The "complete" form with pachyderma and pachyperiostosis, (2) the "incomplete" form with face, acral and bony involvement, (3) the "forms fruste" which may present with only clubbing and thickening of face and/or scalp, but no periosteal ossification.[2] Hyperhidrosis of palms, soles and intermittent swelling or pain over large joints are other features.[1-3] Skin and soft tissue hyperplasia is attributed to an increase in acid mucopolysaccharide of the dermal ground substance.[4]

We describe 2 patients with the "complete" form of pachydermoperiostosis.

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Case Reports

Case 1 : A 28-year-old patient was admitted to our ward for gradually progressive thickening of his scalp, face and extremities. The patient attributed the onset of this change, to an episode of head injury which he suffered 2 years back. The patient was born of a non-consanguinous marriage. There were no stigmata of a similar nature, either in his parents or other family members. Physical examination revealed classic "cutis verticis gyrata" of the scalp. A striking "leonine like" facies was evident due to the deeply forwarded forehead thrown into folds (Fig. 2a). He had "spade like" hands and feet and column like forearms and legs. There was moderate clubbing of the fingers and toes, but the hands had "claw like" appearance due to a persistent flexion of the interphalangeal joints (Fig. 3). Clinical examination of all other systems was normal.

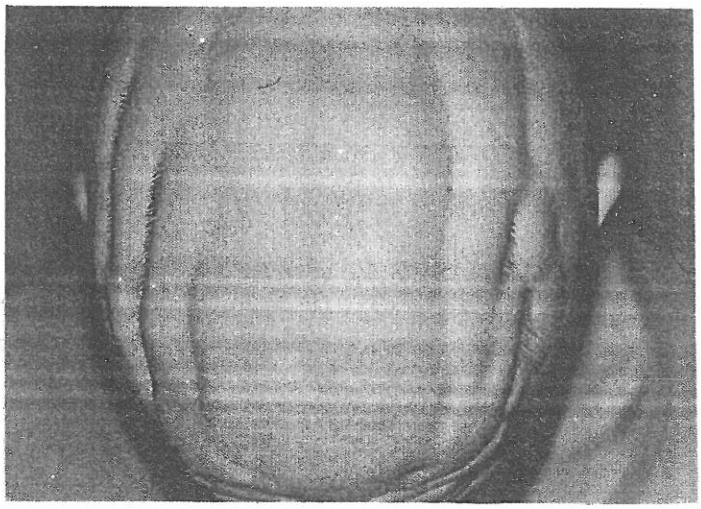

Fig. 1. Figure showing cutis verticis gyrata.

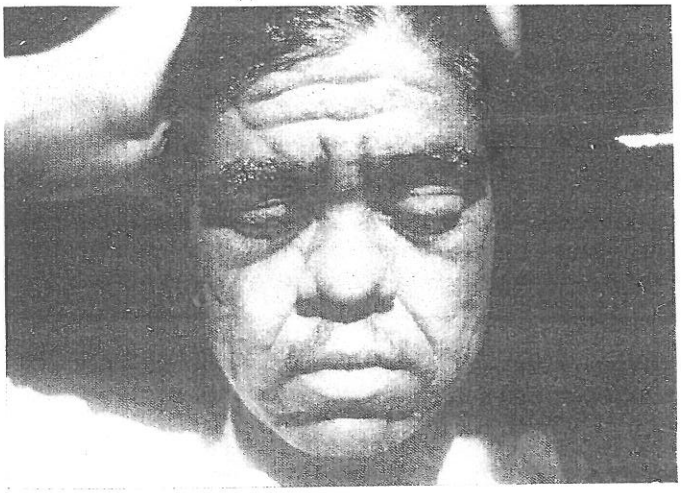

Fig. 2a A "leonine like" appearance of the face in both the cases mimicks lepromatous leprosy

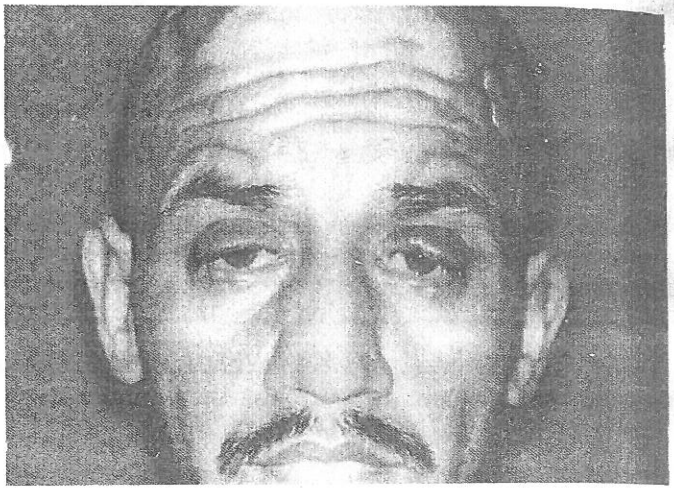

Fig. 2b A "leonine like" appearance of the face in both the cases mimicks lepromatous leprosy

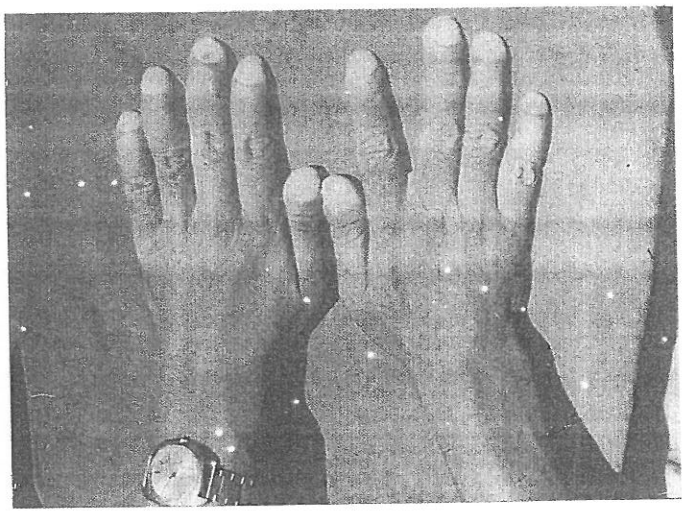

Fig. 3 Clubbing of fingers and a "Claw like" appearance of the hands.

Case 2 : A thirty year old factory worker had a 5 year history of progressive coarsening of his face and scalp and enlargement of his hands and legs.

The grotesque disfigurement of the face was the most striking feature on physical examination. The thickened forehead, thrown into transverse folds gave the face a "leonine" appearance. Cutis verticis gyrata, massive enlargement of the hands and feet and clubbing of the fingers and toes were also present. Clinical examination of other systems was normal.

X-ray of long bones, metatarsals, metacarpals and phalanges revealed periosteal ossification more pronounced at the distal ends of the bones. X-ray skull, lateral view showed a normal sella turcica, and hazy shadow of the hypertrophic scalp tissues. X-ray chest was normal.

Serum calcium and phosphate, alkaline phosphatase were normal. Serum sodium and urinary calcium were within normal range. Fasting and post-prandial blood sugar levels were normal. Radioimmunoassary of plasma growth hormone with L-dopa provocation test revealed normal growth hormone responses. Slit skin smears for acid fast bacilli were negative.

Histopathological examination of skin showed hyperkeratosis, acanthosis and hypertrophy of dermal collagen as significant findings. Alcian blue stain at pH 2.5 demonstrated an increase in acid mucopolysaccharide of dermis.

Discussion

Since historical times, sufferers of pachydermoperiostosis have been confused with acromegaly, leprosy, or syphilitic periostitis.[2] The facial appearance of

pachydermoperiostosis closely mimics the "leonine" facies of lepromatous leprosy. Unfortunately our first patient not only had a so called "leonine" face but also had a mild persistent flexion of his fingers which gave his hands a "claw-like" appearance. Our patient, with his facial disfigurement and his "claw hands" was readily stamped as a "leper" by his unkind fellowmen and was doomed to social ostracism. Our second patient too met with the same tragic fate.

Absence of supraciliary bossing, and prognathism with a normal sella turcica on X-ray skull and growth hormone assay within normal limits ruled out acromegaly. Differentiation of "idiopathic" type of hypertrophic osteoarthropathy which is concisely labelled as "pachydermoperiostosis" (as were our cases); has to be made from "secondary" hypertrophic osteoarthropathy. Here pachydermia and osteoarthropathy are secondary to a "primary" lesion like bronchogenic carcinoma, bronchiectasis or ulcerative colitis.[1,2] Normal chest X-rays and thorough clinical examination, which revealed no primary lesion exclude the possibility of the "secondary" type of disorder, in our patients. Yet, pain in the joints which is a characteristic feature of the secondary form was present in our first case.

In first case the onset of clinical manifestations started soon after the patient had a head injury. We wonder whether it would be too far-fetched to postulate that the head injury may have been the "primary lesion" which triggred off "neurogenic" reflexes in a genetically predisposed individual to result in the dermal and osseous hypertrophy of pachydermoperiostosis.

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