PACHYDERMO PERIOSIS
(Primary or idiopathic hypertrophic osteo-arthropathy) Case report and brief review of literature.

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Summary

Literature regarding pachydermo periostosis is briefly reviewed, underlining the progressive and self-limited nature of the condition.

Clinical manifestations, morbid histological features of the soft tissues and roentgenological findings in an adult of pachydermo periostosis are described. No cause for the malady could be identified except history of consanguinity among the parents of the patient and history of bronchial asthma in the patient 3 years prior to the onset of the condition.

Pachydermo periostosis is a syndrome of insidious development during adolescence, of symmetrical bilateral thickening of the fingers, toes, clubbing of the terminal phalanges, 'paw like or spade like' enlargement of hands and feet and symmetrical and cylindrical thickening of the legs and forearms involving the bones and soft tissues. There is progressive coarsening of the skin of the face, of the forehead and scalp and often excessive sweating of feet and hands and gaping sebaceous glands. On the forehead the skin is gradually thrown into heavy transverse and vertical folds, separated by deep furrows which give the patient 'a worried and angry look'. The nasolabial folds become deep and sharp. The thickening of the scalp tends to produce corrugated surface described as 'bull dog scalp or cutis verticis gyrata'. There is thickening of eyelids with ptosis and narrowing of palpebral fissure. X-ray of the affected long bones — vertebrae, metatarsals, metacarpals, and phalanges reveal symmetrical irregular periosteal ossification and irregular thickening. Sella turcica is normal. It is a familial disorder and familial occurrence is striking. Consanguinity among parents is frequent. The condition is self-limited. Progression stops at the end of adolescence. Vague pains in the bones and joints during active phase of the disease is complained of. It is seen in both sexes, although more frequently in males before or after puberty. The disease may be mild or extremely severe involving a few or all the soft parts and skeleton. Blood and urine are normal. The condition is reported in Japanese, American and African Negroes, Hindus and South American Indians.

Vogl[1] and Goldfischer in 1962 while reporting two cases of pachydermo periostosis in a male and a female had given an excellent review of cases earlier reported. This is probably the best account of reported cases of pachydermo periostosis with illustrations of

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clinical features, inclusive of autopsy findings covering illustrations of the changes in long and small bones and vertebrae.

Freidriech[2] in 1868 reported pachydermo periostois in two brothers named Wilhelm Wagner and Karl Wagner. These brothers had two of the most severe cases ever observed. They were followed by Erb[3], Virchow[4], Sternberg[5] and Marie[6] in the mid 19th century. Wilhelm Hagner, the elder brother noted enlargement of feet at the age of 18 years and 2 years later his hands became enlarged and he could do no skilled work. He had no pain and had no change in general health. Karl Wagner, younger brother noted the changes at the age of 17 years. 20 years later in 1888 Erb and Virchow re-examined the patients and diagnosed them as acromegaly. Elder brother died in 1891 and an autopsy was done by Arnold under the title — Acromegaly — Pachyacria. Sternberg[5] expressed it as not acromegaly but a separate disease entity. Bone changes were extensive. Vertebrae, long bones, and small bones showed stalactite exostosis. Fraentzel[7] in 1888 reported a paper entitled "on Acromegaly". Father and daughter had pachydermo periostosis and Virchow performed autopsy. He found the pituitary normal, osteophyte formation at the phalanges and spine and fusion of a part of dorsal vertebrae. In 1889 Gourand[8] presented to the Societie Medicale des, Hospitaux de Paris, a 50 year old man who showed enlargement of feet, hands, clubbing, coarse wrinkled features, greasy facial skin with acne and wide opening of sebaceous glands. He perspired profusely with sweat drops visible at the finger tips. He was nick named "Grosse pattes". It caused no pain and no discomfort. Tournier described in a 28 year old man, massive thickening of scalp with parallel folds described by Jadassohn[9] in 1906 and named by Unna[10] as 'cutis verticis gyrata'.

Earlier case reports were mostly from France. In advanced cases of pachydermo periostosis, facial features of the patients bear a close resemblance one another.

Touraine[11] and Solente to whom we owe the first comprehensive description of the condition distinguished three forms.

(1) The complete form with pachydermia as well as pachy-periostosis.

(2) Incomplete form without involvement of scalp and

(3) A forme fruste in which clubbing and thickening of the skin of the face and/or of the scalp are present while periosteal changes are either minimal or absent at the time of examination. It can be monosymptomatic.

Vague reviewed 60 cases of pachydermo periostosis.

In 1970 Bourssing[12] reported in a mother and daughter pachydermo periostosis. Typical features of the syndrome were seen at the birth of the daughter. Both had symptoms of gonadal dysfunction. Mother began menstruation at 20 years of age and had scanty menses. The daughter aged 30 years had primary amenorrhoea. David Rimoin[13] grouped clinical manifestations of pachydermo periostosis under a 'syndrome' and indicated it as of autosomal dominant inheritance. Of the 70 reported cases of pachydermo periostosis familial occurrence had been recorded in 30 kindreds — autosomal dominant inheritance with marked variability, phenotypically more severe in males. Familial incidence has been observed in more than 50% of cases reported.

Lazarus[14] and Galloway reported in 1973 pachydermo periostosis in a male

aged 38 years from U. S. A. Neiman, Gompels and Martel reported detailed roentgentelogical and clinical manifestations in 1974 in a male aged 22 years. The patient had painless effusion of the knees, slight gynaecomastia with little facial hair. There were myeloid metaplasia in the spleen. Gharpury[15], Virani and Mistry from India reported in .973 pachydermo periostosis, in a 21 year old male.

Many including Virchow[4] have made mistaken clinical diagnosis of acromegaly mainly because of the facial features and spade like hands and feet. It is likely to be mistaken for Paget's disease of the bones. Pulmonary osteoarthropathy could be eliminated in the absense of recognisable bronchiectasis, suppurative lesion or benign or malignant neoplasm of the lungs.

Pathologic changes involve soft tissue and skeleton. Distal part of the long tubular bones, carpals, metatarsals and phalangeal bones show diffuse periosteal ossification increasing the circumference of the bone without otherwise altering the shape of the bones. Except the cranium all bones may be involved in severe case. Bones are thickened and not deformed. Ossification of the ligaments, tendons, and interosseous membranes occur leading to ankylosising of joints and spine. Hyperkeratosis and hyperplasia of sweat and sebaceous glands and thickening of corium by hyaline fibrous tissue are seen. Hyperplasia of elastic fibres and hyalinisation separated by foci of necrosis and cellular infiltration are observed. Sebaceous glands are enlarged and secretory ducts are elongated. Hair follicles are hypertrophied.

The rarity of published case reports of pachydermo periostosis in the Indian literature has prompted me to record below, findings of a case.

Case Report

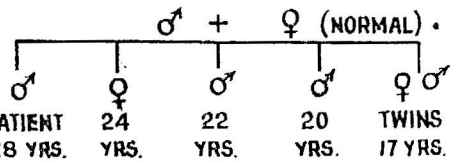
A male aged 28 years was admitted in the Dermatology ward of Govt. General Hospital, Guntur, on 10-4-1977 for thickening of the face and extremities of seven years duration and marked thickening of both the eye lids leading to ptosis and obstruction to vision for 3 years.

History of present illness

At the age of 21, patient observed the changes in the extremities which gradually progressed. There was symmetrical bilateral swelling of knee, ankle and wrist joints associated with pain. There was considerable thickening of both forearms, hands, legs and feet. Thickening of the skin of the forehead, face, extremities and of the upper eyelids associated with conjunctival discharge was present for 3 years. Patient did not sweat excessively. He had not lost weight and had no cough and was not dyspnoic. He had suffered from bronchial asthma 3 years prior to the onset of the present malady. He had not been doing any work due to pain in the joints.

Family History

Patient was born of consanguinous parents.



All the children except the patient and the parents were normal. The patient was the first born.

Physical Examination

Revealed a moderately nourished male of medium stature. The skin of the entire face and scalp was grossly thickened with coarse facial features leading to furrowing of the scalp and face and oiliness of the skin (Fig. 1 & 2.) Both upper eye lids were greatly thickened resulting in mechanical ptosis.

Sebaceous glands showed hyperplasia with wide open sebaceous pores. There was bilateral symmetrical thickening of the knee and ankle joints. (Fig. 3.) The synovia of the knee joints appeared thickened. The skin over the forearms, hands, legs and feet were thickened and so also the underlying bones. The hands and feet were spade like. There was well marked clubbing of the fingers (Fig. 4) and toes. Cardiovascular, respiratory, gastrointestinal and central nervous system were normal. Examination of eyes revealed lenticular opacities, temporal optic atrophy and blurring of disc margins. A-V ratio was normal.

Investigations

Blood picture, urine analysis and Ba meal series were normal. X-Ray of chest and skull were also normal.

There was increased width of the long bones due to extensive periosteal reaction and cortical thickening. (Fig. 5). X-Ray of hands showed no periosteal reaction. (Fig. 6) X-Ray of foot showed some disorganisation on the medial aspect of navicular and cuniform bones. The terminal phalanges of great toes showed mild exostosis (Fig. 7).

Biopsy of the skin

Showed atrophy of the dermis with lymphocytic infiltration at the dermoepidermal junction. There was moderate collegenisation of the dermis. Adnexial glands appeared normal.

The clinical features of the case were pathognomonic of pachydermo periostosis and have enabled to rule out the possibility of acromegaly, myxoedema, syphilis, leprosy and Paget's disease of the bones.

Fig. 1 Photograph illustrates "Cutis verticis gyrata".

Treatment

Blaskovic's operation with wedge resection of both the upper eyelids was done and the patient was able to see well.

Discussion

Barring the history of bronchial asthma 3 years prior to the insiduous onset of the condition, there was no significant past illnesses. History of consanguinity among the parents supports earlier observations made by others. It is well established that pulmonary osteoarthropathy and pachy-dermo periostosis are independent conditions but perhaps genetically related. Nothing is known about the aetiology of pachydermo periostosis, except that of an underlying genetic factor. Pathogenetic mechanism responsible for thickening of the skin and clubbing of fingers remains

to be clarified, although neurogenic and circulatory causes have been considered.

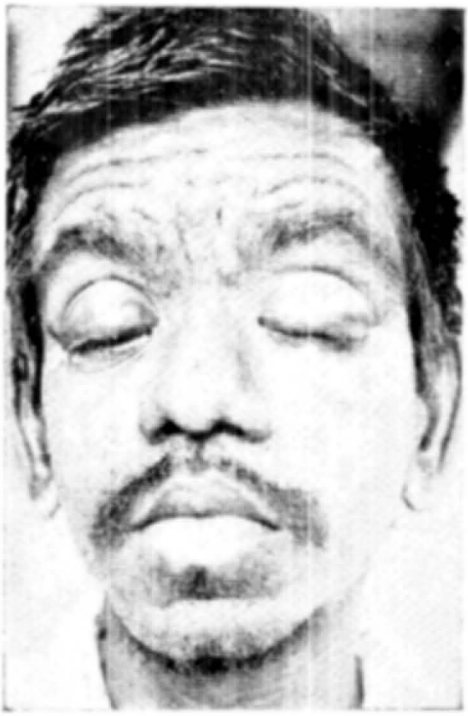


Fig. 2 Photograph illustrates drooping, heavy and thickened eyelids, folds, and furrows of the forehead.

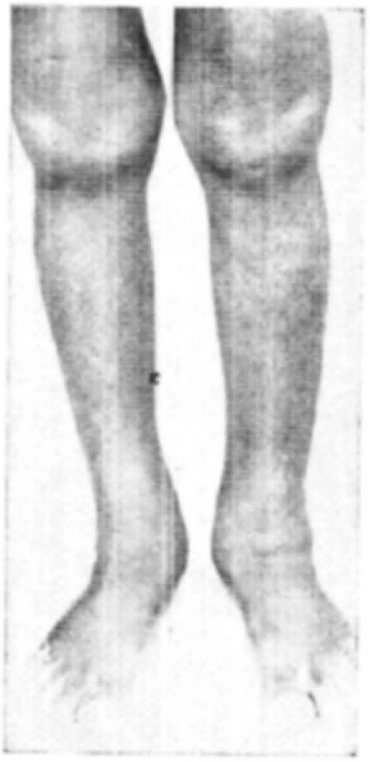


Fig. 3 Photograph illustrates bilateral swelling of knee and ankle joints and thickening of the skin.

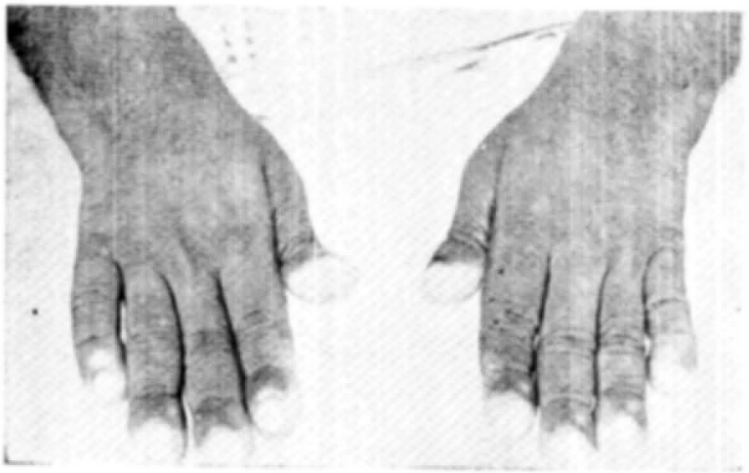


Fig. 4 Photograph illustrates marked clubbing of the fingers.

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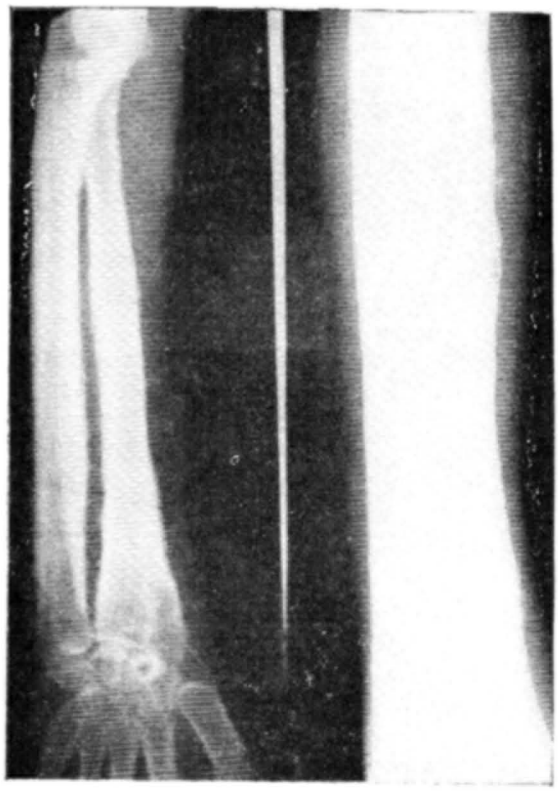

Fig. 5 X-ray of forearm and leg – periosteal reaction along the medical length of radius in lower 2/3rds. Corte is thickened in both ulna and radius in lower 2/3rds. X-ray leg shows periostitis in tibia of the lower 2/3rds. Cortex thickened along the whole length of the lower half.

Fig. 6

X-ray of hand – Soft tissue of the fingers especially of the phalanges is thickened. No evidence of periosteal reaction.

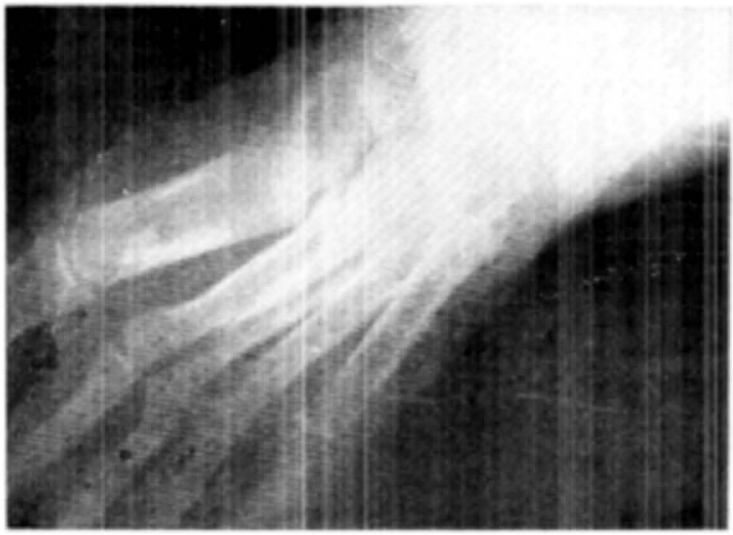

Fig 7

X-ray foot tarsal bones - some amount of disorganisation of bone on the medial aspect of navicular and cuniform bones which is of periosteal reaction type. The terminal phalanges of the great toes show evidence of mild exostosis.

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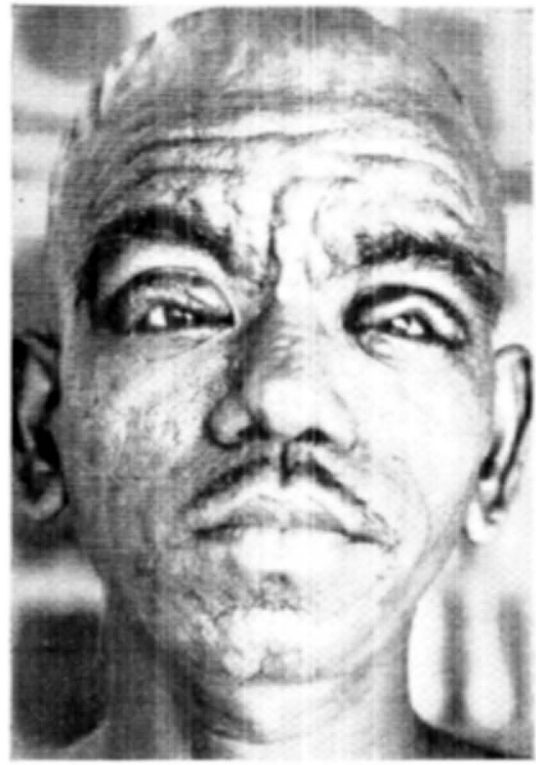

Fig. 8 Same patient after Bloskovic's operation with wedge resection of the both upper eyelids. Patient is able to see well now.