Pachydermoperiostosis: A Rare Case Report

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Abstract:

Pachydermoperiostosis (PDP) is a rare disorder with variable presentation and it is characterized by clubbing of the fingers (acropachia), skeletal changes (periostosis), thickening of the skin (pachyderma), and excessive sweating (hyperhidrosis). Clinical presentations of PDP can be confused with secondary hypertrophic osteoarthropathy, psoriatic arthritis, rheumatoid arthritis, thyroid acropachy, and acromegaly. We report a patient with complete PDP.

Keywords: Pachydermoperiostosis, Pachyderma, Clubbing of fingers

Introduction:

In 1868, Friedreich first described Pachydermo periostosis as 'Hyperostosis of the entire skeleton'. Unna, in 1907 called 'cutis verticis gyrate' for thick, transversely folded skin of scalp and forehead.[1] Three dermatologists, in 1935,Touraine, et al,[2] explained it as familial condition with three forms: complete (pachyderma and perisotosis), incomplete (without pachyderma) and the forme fruste (pachydermia with minimal skeletal changes). Rimoin noticed affected persons in successive generations.[3]

As per Borochowitz,[3] the diagnosis should only be made when at least two out of a family history, clubbing, hypertrophic skin changes, and bone pain/radiographic changes are present. Jajic estimated the prevalence of the disease is 0.16%.9,10 Symptoms usually appear around puberty, with a male to female ratio of 7:1, and males are severely affected.[4] The main features are digital clubbing, skin changes (flushing, blanching, hyperhidrosis and hypertrophy) causing coarse facial features with thickening, furrowing and excessive oiliness of the skin of the face and forehead. Bone and joint involvement includes arthritis, arthralgia, periosteal new bone formation, subperiosteal ossification, acro-osteolysis and osteoporosis. Gastric hypertrophy, gastric ulcer and other endocrine abnormalities have been described.

An infantile form has been described which is characterized by early presentation with enlargement and delayed closure of the cranial sutures, patent arterial duct and skin manifestations.[4] This condition progresses slowly for a few years and is self-limiting thereafter. It typically appears presents during childhood or adolescence, often around the time of puberty, and progresses slowly.

Case report:

A 29 year old unmarried non-alcoholic, non-smoker male born out of consanguineous marriage presented to dermatology outpatient department with progressive thickning skin folds of face, scalp and oily skin since 20 years of age.

On examination patient had cutis vertis gyrate (Fig: 1), coarse facial wrinkles, profuse seborrhoea and sweating. Pectus carinatum was seen (Fig: 2). Grade 4 clubbing of all fingers (Fig: 3) and toes was noted along with bilateral knee joint swelling and bilateral pedal oedema. (Fig: 4 & 5)

BMI of the patient was normal. Both the upper and lower limbs were in proper proportion. There was no history of morning stiffness and no abnormality seen in tongue.

Hormonal assay and other laboratory investigations were within normal limits. Histopathological examination of forehead skin done.(Fig: 6 & 7)

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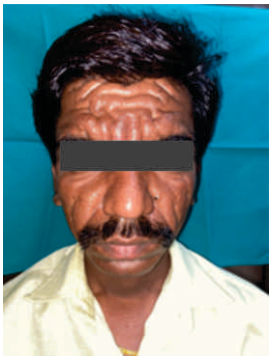
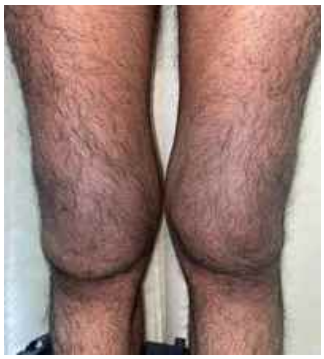


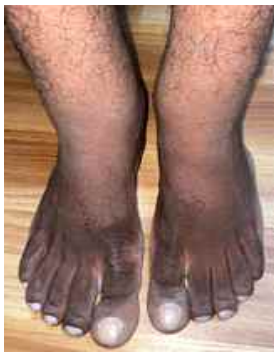
1) Cutis vertis gyrate

2) Pectus carinatum

3) Clubbing



4) Knee swelling



5) Pedal edema

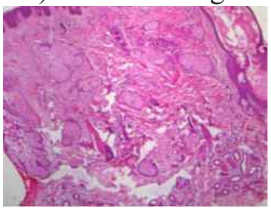


Fig: 6

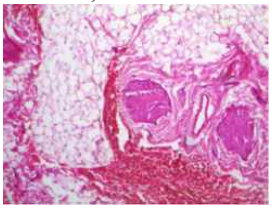


Fig: 7

Fig 6 & 7: mild epidermal hyperplasia with elongated rete ridges. Papillary dermis shows sparse perivascular infiltrate of lymphocytes and melanophages with occasional plasma cells. Reticular dermis is thickened. Follicular infundibulae are dilated and plugged with stratum corneum. There are multiple hyperplastic sebaceous globules lying freely along with increased arrector pili muscles within reticular dermis. Deep dermis shows increased number of eccrine glands and horizontally oriented thickened nerves. Similar thickened nerves are also seen in subcutaneous fat.

Discussion:

Pachydermoperiostosis is an autosomal dominant variant with incomplete penetrance but in some cases it is autosomal recessive with its relation to consanguinity. The pathogenesis of PDP is not fully known. Anionic solute carrier transporter 2A1 and 15-hydroxyprostaglandin dehydrogenase gene are associated with PDP.[5] Study conducted by Sasaki et al.[6] showed that, severity of pachydermia and histological changes are related to the serum levels of PGE2. Increased levels of PGE2 are assumed to cause vascular stimulation and tissue remodelling by action of cytokines which leads to pachyderma, arthritis, periostosis, acro-osteolysis and hyperhidrosis. As per the study conducted by Martinez-Lavin,[7] pachydermoperiostosis is caused by actively proliferating fibroblasts which causes infinite collagen fibre proliferation.

Wegrowski[8] proposed that, increased decorin protein synthesis and dysregulated deposition of matrix is the cause of pachydermoperiostosis.

Pachydermoperiostosis is diagnosed with the help of following diagnostic criteria:[5,9]

Major criteria: Pachyderma, periostosis, finger clubbing.

Minor criteria: hyperhidrosis, arthralgia, gastric ulcer, cutis verticis gyrate, blepharoptosis, joint effusion, column-like legs, edema, seborrhea, acne, flushing.

The most frequent skin symptom of this disease is pachydermia affecting limbs and face.

The pachyderma is graded as follows –

1.Grade 0:- absence

2.Grade 1:- mild to moderate involvement (cutaneous thickening without puckering),

3.Grade 2:- severe (cutaneous thickening and puckering), seborrhea (90% of cases), acne, folliculitis, dilated pores, hyperhidrosis of the palms and soles (44–67% of cases) with occasional flushing, thickened eyelids (30–40% of cases), cutis vertices gyrate (24% of cases), and reduced facial and pubic hair. 89% of patients shows digital clubbing and nail bed capillaroscopy shows capillary enlargement and increased tortuosity. 80-97% patients shows irregular periosteal ossification in distal ends of long bones. 41% joint effusion and 20-22% cases have arthritis.[10]

Pachydermoperiostosis has been found in association with some malignancies like facial epidermoid carcinoma, peptic ulcer diseases, gastric adenocarcinoma, Cohn's disease and myelofibrosis. Due to increase in soft tissue bulk and hyperostosis complications like ptosis, nerve ending compression,

kyphosis, hearing problems, arthrosis and carpal tunnel syndrome.

This syndrome can be distinguished from acromegaly on the basis of clinical features and laboratory findings. In contrast to PDP, acromegaly presents clinically with larger bones in the face, skull and limbs, jaw prognathism, along with elevated insulin-like growth factor-1 levels and positive oral glucose tolerance test. In our patient, closer scrutiny in the clinical examination coupled with the negative biochemical markers for acromegaly effectively allowed us to rule out this differential.

Our patient had all three major criteria i.e., hyperostosis, finger clubbing, and pachyderma.

Similar findings were seen in case report by Supradeeptha C et all.[11]

We started this patient on colchicne 0.5mg BD/day for articular symptoms, pachyderma and folliculitis and on isotretinoin 20mg HS/day for symptoms related to pachyderma and cutis vertis gyrate.

Conclusion:

The diagnosis of pachydermoperiostosis is done on the basis of pachyderma, clubbing of digits and periostosis in absence of any systemic diseases. And as the prognosis, management and long term sequel differs needs to be differentiated from thyroid acropachy, secondary hypertrophic osteoarthropathy, acromegaly, syphilitic periostosis and chronic inflammatory rheumatic disease. Patient needs close follow-up because these patients can develop long-term complications.

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