Case Report

Pachydermoperiostosis: A Rare Genetic Disorder

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Abstract

Pachydermoperiostosis (PDP) is a rare genetic disorder. Finger clubbing, skin changes and bony changes are the main three features of it. Here we present a 26-year-old male patient who was admitted for some other neurological disorder and diagnosed clinically as PDP. We diagnosed the condition clinically as gene analysis is not available in Bangladesh for a confirm diagnosis.

Key words: *Pachydermoperiostosis, periostosis, clubbing.*

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Introduction

Pachydermoperiostosis (PDP) is a rare genetic disorder of autosomal dominant variant.[1]It is characterized by "HOA triad" of digital clubbing, periostosis and pachydermia. It is a primary hypertrophic osteoarthropathy, where secondary hypertrophic osteoarthopathy has an underlying disease (cardio pulmonary disease, malignancy or paraneoplastic syndrome).[2] We report a case of PDP (Fruste form) with Guillain-Barré syndrome **(**GBS).

Case Report

A 26-year-old man presented with progressive weakness of both lower limbs for 3 weeks which he first noticed difficulties in standing from sitting position. His weakness gradually spread to upper limbs. Patient was unable to close his eyes and completely open his mouth. He did not complain any sensory deficit and his bowel and bladder habit was normal. Since last 4 years he experienced broadening and thickening of hands and feet with thickening of the skin of face and forehead.Patient did not experience any fever, shortness of breath, fatigability, headache, skin rash, diplopia, urinary incontinence, chest infection or weight loss.

On examination he was conscious, oriented and cooperative.His blood pressure was 120/80 mmHg, pulse-80b/min, R/R:16 breaths/min.Temp-98.4 F. There was bilateral facial nerve palsy (lower motor type) with 3/5 muscle power and loss of deep tendon reflex in all 4 limbs. Patient had pachydermia involving hands and feet with coarse architecture and large in size (Figure 1). There was digital clubbing, leonine facies and cutis verticis gyrate (Figure 1 and 2). Skin was oily and greasy. Sebborheic dermatitis was present in scalp. Patient did not have any joint tenderness.There was wasting of small muscle of hand and positive tinea sign. Other systemic examination findings were within normal limit.

Investigations showed CBC-8300/mm3, S. Creatinine -0.9mg/dl, S. electrolytes: Na-137mmol/L, K-4.1mmol/L,Cl-109, HCO3-27. FBS-4.43mmol/L, 2hABF-5.04mmol/L.S. Prolactin-10.10 ng/ml. Growth hormone-0.05ng/ml.S.TSH-2.3, X-ray skull showed-Bones are prominent, no evidence of osteoporosis, sclerosis or erosion. Sella was normal. X-ray of hand B/V showed thickness of cortex with expansion of the bone, soft tissue was swollen. CXR-normal. NCV was suggestive of AIDP. MRI of brain revealed normal. All these clinical findings and investigations supported our diagnosis.

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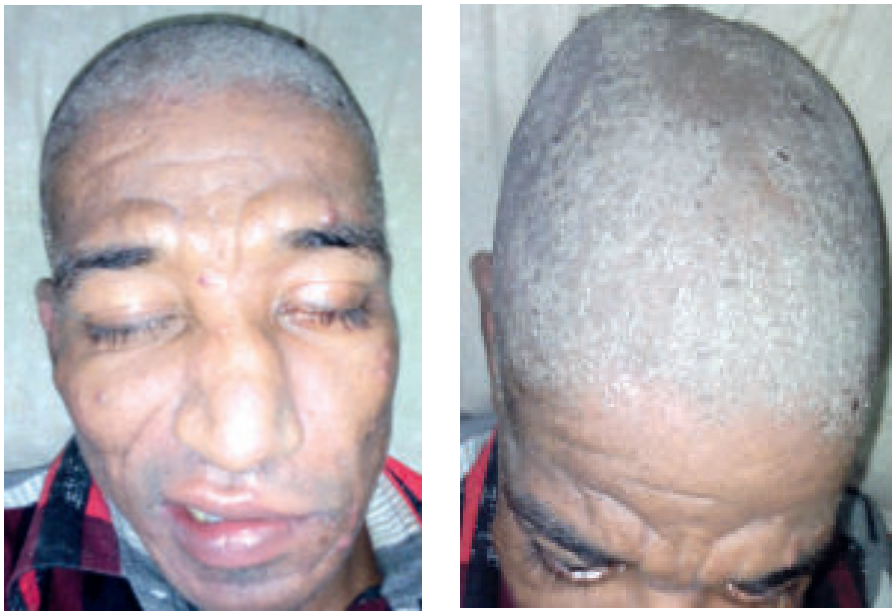

Figure 1a and 1b. *Facial and scalp involvement with pachydermia and cutis gyrata*

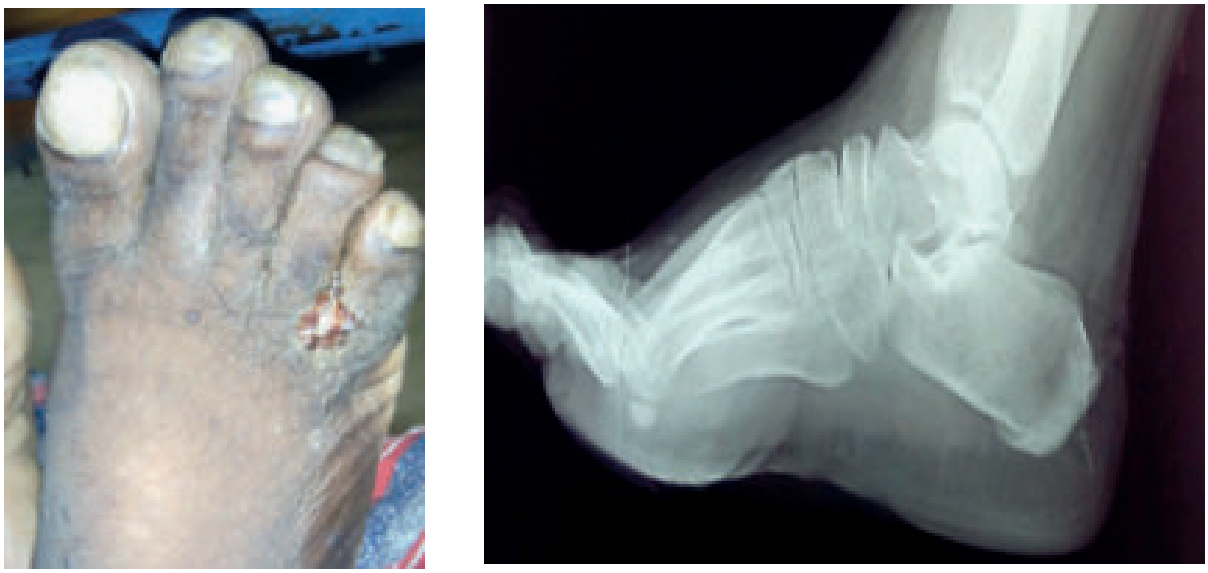

Figure 2a and 2b. *Finger clubbing, periostosis and increase heel pad thickness*

Discussion

Pachydermoperiostosis is a primary HOA, an autosomal dominant rare disorder which is mainly characterized by finger clubbing, skin thickening and excessive bone formation.This disease affects relatively more men than women. After onset, the disease stabilizes after about 5-20 years. Less than 250 cases have been reported world wide of this disease. PDP can be grouped into three a) Complete form in which patient has all the symptoms but mainly Pachydermoperiostosis & finger clubbing also known as full blown phenotype b) Incomplete form characterized by having mainly effect on bones & thereby skeletal changes and limited cutaneous manifestation and c) The fruste form in which

mostly by cutaneous symptoms with minor skeletal change.[2]

The causes of this disease are still unknown but 2 theories have been suggested. The neurogenic theory proposes that stimulation of vagus nerve leads to vasodilation, increased blood flow and PDP.

Common clinical features are pachydermi, course oily skin, thick hand & foot skin, leonine facie, cutis verticis gyrate, seborrhoeic dermatitis, periostosis, thick fingers and toe bone, digital clubbing, hyperhidrosis. All these features were present in our patient. Other less common features include drooping eyelid, arthralgia, joint diffusion, muscle discomfort, peptic ulcer etc. The easiest way to diagnose is pachydermia, finger clubbing & periostosis of long bone. Sub periosteal new bone formation can be detected by radiograph of long bone. To diagnose this disease we must exclude secondary causes of HOA (any sign of cardiovascular ,pulmonary, hepatic, intestinal & mediastinal disease & by investigations).[3]

Skin biopsy is another way to diagnose PDP, but it is not very specific method, because other diseases share the same skin alteration with PDP, such as mixed edema and hypothyroidism. In order to exclude other diseases, hormonal studies are done. Since PGE2 levels are co-related with PDP, urinary PGE2 can be useful biomarkers for this disease.For following up of PDP disease activity, bone formation markers such as TAP, BAP, BGP, carbodyterminal peptide of type 1 pro-collagen or NTX can play an important role.PDP usually progresses for 5-20 years until it becomes stable.Life expectancy may be normal, despite patient getting functional and cosmetic complications.[4,5]

Treatment are symptomatic. Rheumatologic symptoms can be improved by treating with bisphosphonate (pamidronate or risedronate). It acts by inhibiting osteoclastic bone resorption and therefore reduce bone remodeling and alleviate painful pathway. In isolated cases Tamoxifen was effective.Retinoids are used to improve skin manifestations. Colchicine also be used. Botulinum toxin type-A (BTX-A) improves leonine faces of the patient. Surgical cases may also require to improve facial appearance and to relieve entrapment neuropathy.[6]

This is the first case in the world that the patient presents with PDP with GBS. We want to draw attention if there is any clinical correlation between this two diseases or not or there is any other disease.

Conflict of interest: Nothing to declare.

References

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