*Case Report*

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# Pachydermoperiostosis mimicking acromegaly: A case report

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**Abstract**

Pachydermoperiostosis (PDP) is a rare clinical condition that is characterized by pachyderma (thickened facial skin), skeletal changes (periostosis), excessive sweating (hyperhidrosis), and acropachia (digital clubbing). Making a diagnosis followed by a proper care plan can be extremely challenging even for experienced clinicians due to the rarity of this disease. It mimics the clinical and radiographic manifestations of acromegaly. Acral enlargement, cutis verticis gyrate, facial coarsening, hyperhidrosis, seborrhea, and acne are common in pachydermoperiostosis as well as in acromegaly. Therefore, it should be considered one of the differential diagnoses in evaluating acromegalic patients. In this case report, we discuss a 22-year-old man with progressive enlargement of his hands and feet along with sweating but there was no biochemical evidence of acromegaly and was finally diagnosed as a case of pachydermoperiostosis. *[J Assoc Clin Endocrinol Diabetol Bangladesh, July 2023; 2 (2): 71-74]*

**Keywords:** Pachydermoperiostosis, acromegaly

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## Introduction

Pachydermoperiostosis (PDP) is a rare clinical condition affecting the skin and musculoskeletal system. It was first described in 1868 by Friedreich.[1] It usually presents with enlargement of acral body parts with clubbing, sweating, body ache, and skin thickening.[2] PDP is also called hypertrophic osteoarthropathy, Rosenfeld-Kloepfer syndrome, and Touraine-Solente-Gole syndrome[3] is a rare disease characterized by three main features - clubbing, skin thickening (pachyderma), and increased sweating (hyperhidrosis).[4] Abnormal vascular endothelial growth factor and/or genetically determined prostaglandins overexpression may play a key role in its pathogenesis.[5] There are two genes that have been associated with *PDP: HPGD* and *SLCO2A1*. It is usually transmitted in an autosomal dominant or autosomal recessive manner but a non-genetic form is also identified.[6] The actual incidence is not known but adolescent males are more frequently affected with a male: female ratio of 7:1.[7] The diagnostic criteria for PDP incorporate 3 major criteria comprising of pachyderma, periostosis, and finger clubbing as well as minor criteria counting hyperhidrosis, arthralgia, gastric ulcer, cutis verticis gyrata, blepharoptosis, joint effusion, column-like legs, edema, seborrhea, acne, and flushing.[8] There are three forms of the disease. The complete form consists of three major and some minor criteria, the incomplete form consists of two major and some minor criteria, and the 'forme fruste' consists of one major and some minor criteria.1 Pachydermoperiostosis is more commonly confused with acromegaly from which it can be differentiated with biochemical and radiological investigations (as most of the clinical features overlap). Characteristic morphological appearance, body habitus, and typical radiological features help clinch the diagnosis. Patients with PDP also frequently visit a rheumatologist, ophthalmologist, or dermatologist with complications. A multidisciplinary team involving an Endocrinologist is required to evaluate these patients. Treatment is usually supportive.

**Case Summary**

A 22-year-old man presented with enlarged hands and feet which were noticed since birth, slowly progressive, associated with the thickening of the skin of hands and feet. There was also excessive sweating of his hands which made him feel uncomfortable even in a well-ventilated room and in cold weather. He also complained about headache for the last 1 year which was dull aching, mild to moderate in intensity. It felt like a tightness across the forehead and on the sides and back of the head, not associated with visual disturbance, nausea, or vomiting and not aggravated by coughing, sneezing, or bending forward. He had no history of any weight change. His sleep pattern and bowel bladder habit were normal. There was no history of nausea, vomiting, abdominal pain, polyuria, polydipsia, or postural dizziness. He had no history of diabetes, hypertension or cardiac disease. His maternal uncle had a similar type of condition (enlarged hands and feet with thickened skin).

**Figure-1:** Facial features of the patient showing thick skin and enlarged nose, and lips.

On examination, height was 145 cm, weight 51 kg, body mass index (BMI) 24.3 kg/m$^2$, and waist circumference 89 cm. He had an enlarged nose and lips but no macroglossia or prognathism. His skin was thick and oily (Figure-1). His hands were large, warm, sweaty, doughy-feeling hands with spade-like fingers. Tinel's and Phalen's signs were negative. His feet were also large. He had clubbing of all the fingers and toes (Figure-2). Vital signs were normal (blood pressure: 115/80 mm Hg). Other system examinations revealed no abnormalities.

Investigations revealed random blood glucose was 4.8 mmol/L, serum alanine aminotransferase (ALT) 19 U/L, s. creatinine 0.72 mg/dl, basal growth hormone 0.898 ng/ml, and insulin-like growth factor-1 (IGF-1): 195 ng/ml. Growth hormone after oral glucose tolerance was less than 0.15 ng/ml. X-ray of hands

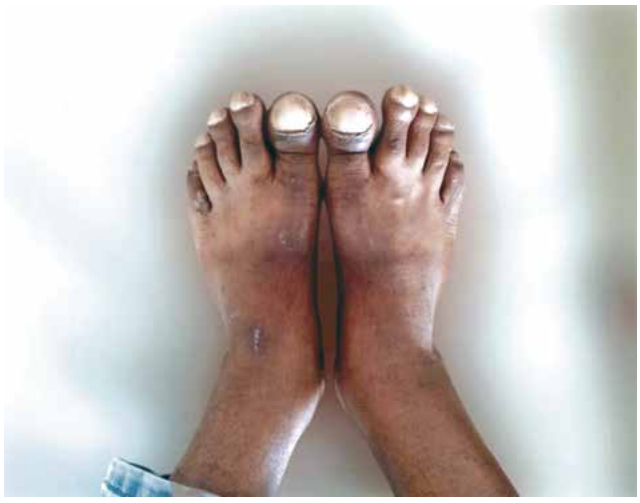

**Figure-2:** Clubbing of the fingers and the toes

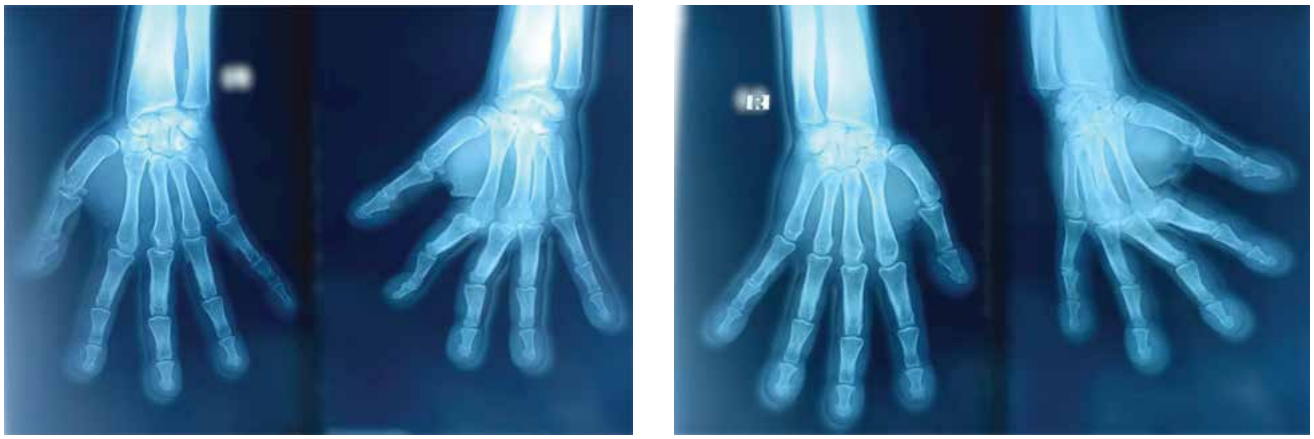

**Figure-3:** X-ray of the hands showing a periosteal reaction

showed terminal phalangeal tufts were hypertrophied having a spade appearance, soft tissue shadow was increased around the distal phalanx of all fingers, periosteal reaction was noted at the distal part of the radius, articular margins and joint spaces were regular (Figure-3). X-ray of the feet showed increased heel pad thickness (28 mm) (Figure-4). X-ray skull showed no abnormalities (Figure-5). ECG and echocardiogram were normal.

## Discussion

Our patient presented with an acromegaloid appearance for which there is a wide range of

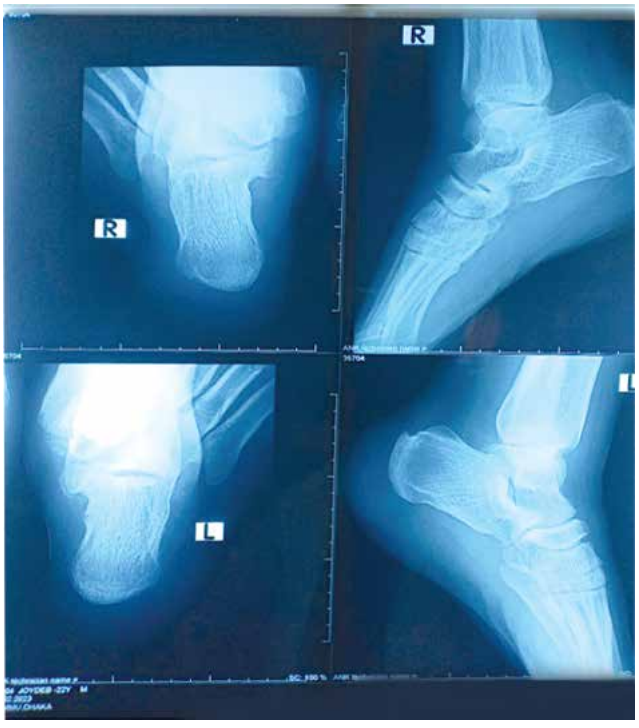

**Figure-4:** X-ray of the feet showing increased heel pad thickness

**Figure-5:** X-ray of the skull was normal

differential diagnoses like acromegaly, pseudoacromegaly due to severe insulin resistance, syphilitic periostitis, McCune Albright syndrome (MAS), psoriatic onycho-pachydermo-periostitis (POPP) and pachydermoperiostosis.[7] Many of the features of the patient like skin thickening, sweating and acral enlargement are common presentations in acromegaly. Other differential diagnoses are less likely from his clinical features. He had no features suggestive of insulin resistance like acanthosis or central obesity, no skin rash or joint pain suggestive of psoriasis or hormonal hypersecretion suggesting MAS. His insulin-like growth factor-1 and growth hormone 1 hour after oral glucose tolerance test were normal which excluded the possibility of acromegaly. The presence of clubbing in the fingers pointed to a possibility of pachydermoperiostosis. X-ray of his

hands showed periostitis of the radius and ulna. A diagnosis of pachydermoperiostosis was made as there were typical physical findings of acral enlargement, skin thickening, and sweating with radiological evidence of periostitis but no biochemical evidence of acromegaly. Genetic testing for mutation in *HPGD* and *SLCO2A1* is helpful for diagnosis but is not readily available in our country. The patient had an incomplete form of the disease as he had two major criteria of clubbing and periostosis. He had thick oily skin but had not developed pachyderma yet.

## Conclusions

In conclusion, clinical presentations of pachydermoperiostosis can be confused with multiple other diagnoses, especially acromegaly. The presence of clubbing and periostitis usually points towards pachydermoperiostosis as they are usually absent in acromegaly.

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**Conflict of Interest**

The authors have no conflicts of interest to disclose.

**Financial Disclosure**

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**Data Availability**

Any inquiries regarding supporting data availability of this study should be directed to the corresponding author and are available from the corresponding author on reasonable request.

**Ethics Approval and Consent to Participate**

Written informed consent was obtained from the patient. All methods were performed in accordance with the relevant guidelines and regulations.



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