DOI: 10.21088/jd.2582.3582 10225.3

RFP Journal of Dermatology
Volume 10 Number 2, July - December 2025

CASE REPORT

Pachydermoperiostosis, Complete Form: A Case Report of Rare Occurrence

Ketki Shekhar Bhoite[1], Siddhi Rajesh Patadia[2], Sunanda Arun Mahajan[3]

HOW TO CITE THIS ARTICLE:

Ketki Shekhar Bhoite, Siddhi Rajesh Patadia, Sunanda Arun Mahajan, *et al.* Pachydermoperiostosis, Complete Form: A Case report of Rare Occurrence. RFP Jr of Drea 2025; 10(2): 59-62.

ABSTRACT

Pachydermoperiostosis is a rare genetic disorder featuring a triad of pachydermia, periostosis and digital clubbing. It is either caused by the mutations in the HPGD (AR inheritance) or SLCO2A1 gene (AD inheritance), resulting in elevated prostaglandins E2 levels. It can be classified on the basis of presence or absence of underlying cardiac, pulmonary or hepatic disease as primary or secondary. We hereby report a case of complete form of primary pachydermoperiostosis.

KEYWORDS

• Pachydermia • Periostosis • Hypertrophic Osteoarthropathy.

INTRODUCTION

Pachydermoperiostosis, also called as Touraine-Solante-Gole syndrome, is a rare genetic disorder inherited by an autosomal dominant trait, characterised by a triad of pachydermia, periostosis and digital clubbing. It is either caused by the mutations in the HPGD (AR inheritance) or SLCO2A1 gene (AD inheritance), resulting in elevated prostaglandins E2 levels which induces cytokine-mediated tissue remodelling and vascular stimulation, thereby causing hyperhidrosis, periostitis, pachydermia and arthritis. It is a condition of Primary idiopathic form of hypertrophic osteoarthropathy without any underlying pulmonary, cardiac or hepatic causes which occurs in secondary hypertrophic osteoarthropathy. It is predominantly seen in adolescent males with an approximate male to female ratio of 7:1.[3] We hereby describe this case of rare occurrence.

AUTHOR'S AFFILIATION:

[1] Assistant professor, Department of Dermatopathology, Seth GS Medical College and Kem Hospital, Mumbai, India.

[2] Senior Resident, Department of Dermatopathology, Seth GS Medical College and Kem Hospital, Mumbai, India.

[3] Professor, Department of Dermatology, Seth GS Medical College and Kem Hospital, Mumbai, India.

CORRESPONDING AUTHOR:

Sunanda Arun Mahajan, Professor, Department of Dermatology, Seth GS Medical College and Kem Hospital, Mumbai, India.

E-mail: sunandamahajan@gmail.com

➢ **Received:** 22-05-2025 ➢ **Accepted:** 27-09-2025

CASE REPORT

A 31 year old male, born of third degree consanguineous marriage, presented to the OPD, with complains of drooping bilateral eyelids. On enquiry, there was also thickening of the skin on his face since 6 years, lengthening of both upper and lower extremities and widening of the joints since 10 years with intermittent joint pain. It was associated with hyperhidrosis and seborrhoea of the face, scalp and axillae with foul smell. He complained of easy fatigability since few years with recent onset breathlessness on exertion. He was also a known case of hypothyroidism controlled on medications.

On examination, the patient had coarse facial features with pronounced skin folds over his forehead and nasolabial grooves with drooping of eyelids and an enlarged nose (Figure 1). Skin over the face was greasy with acneiform lesions and flaking over the scalp due to seborrhoea. His hands and feet showed skin thickening with grade 4 digital clubbing (hypertrophic osteoarthropathy) of all digits (Figures 1). There was an increase in the arm span with column like legs.

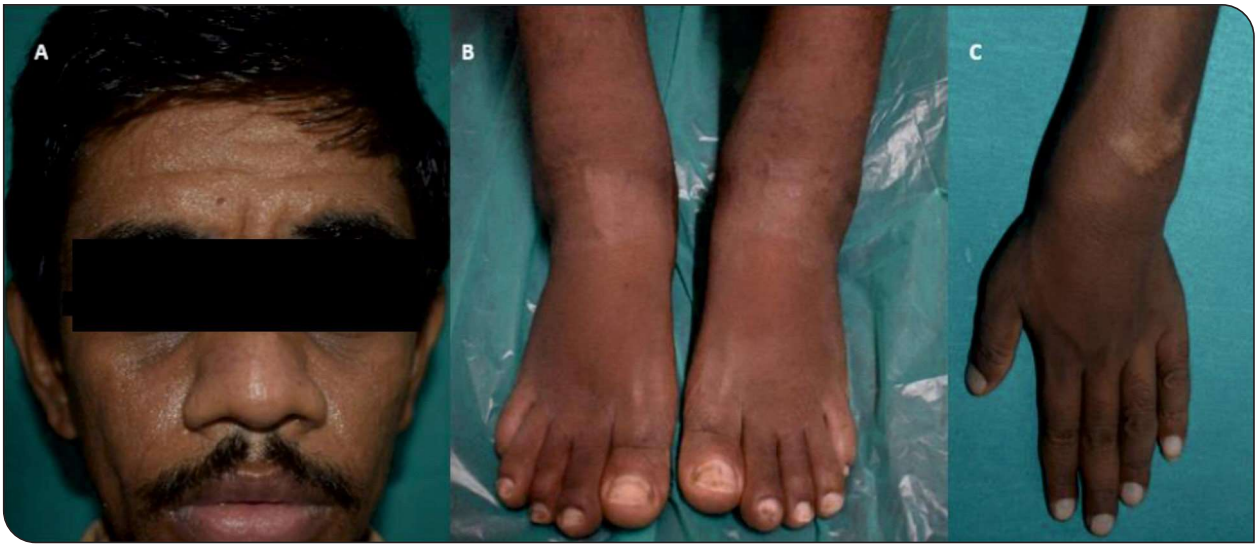

Figure 1: A. Face shows diffuse thickening of the skin and furrowing along skin folds with enlarged nose and drooping of eyelids; **B.** Bilateral ankle joint shows enlargement; **C.** Grade 4 digital clubbing is seen, with widening of the wrist joint.

On investigating it was found that the haemoglobin was low (7.4 gm%) and the peripheral smear showed hypochromic RBCs. The serum iron levels were low, with normal levels of transferrin saturation, Total Iron Binding Capacity and haemoglobin electrophoresis suggesting iron deficiency anaemia secondary to chronic kidney disease (erythropoietin deficiency). Renal function tests and abdominal ultrasound were suggestive of chronic kidney disease secondary to NSAIDS taken for recurrent joint pain. On further evaluating for his joint pain we found the CRP and ESR to be elevated with a positive rheumatoid factor but anti-CCP antibodies and anti-tissue transglutaminase antibodies were not detected and no other rheumatological cause was found. X-rays of bilateral hands, wrist, ankles, knees, pelvis and hips showed symmetric shaggy subperiosteal bone formation with the widening of diaphysis and distal end of long bones (figure 2), suggestive of hypertrophic osteoarthropathy. In order to rule out the underlying causes for it we investigated further and found a normal chest HRCT with normal growth hormone and insulin like growth factor levels. A skin biopsy was performed which showed thickening of dermal collagen fibres and sebaceous and eccrine hyperplasia (figure 3) confirming the clinical findings of pachydermia. There was no history of similar presentation in his family. He was thus diagnosed as pachydermoperiostosis, the complete form of primary hypertrophic osteoarthropathy with iron deficiency anemia secondary to chronic kidney disease with hypothyroidism. Genetic testing could not be done due to the socio-economic condition of the patient. He was treated with capsule isotretinoin 20mg once a day with oral antihistamines, vitamin D3 and calcium supplements with topical moisturisers.

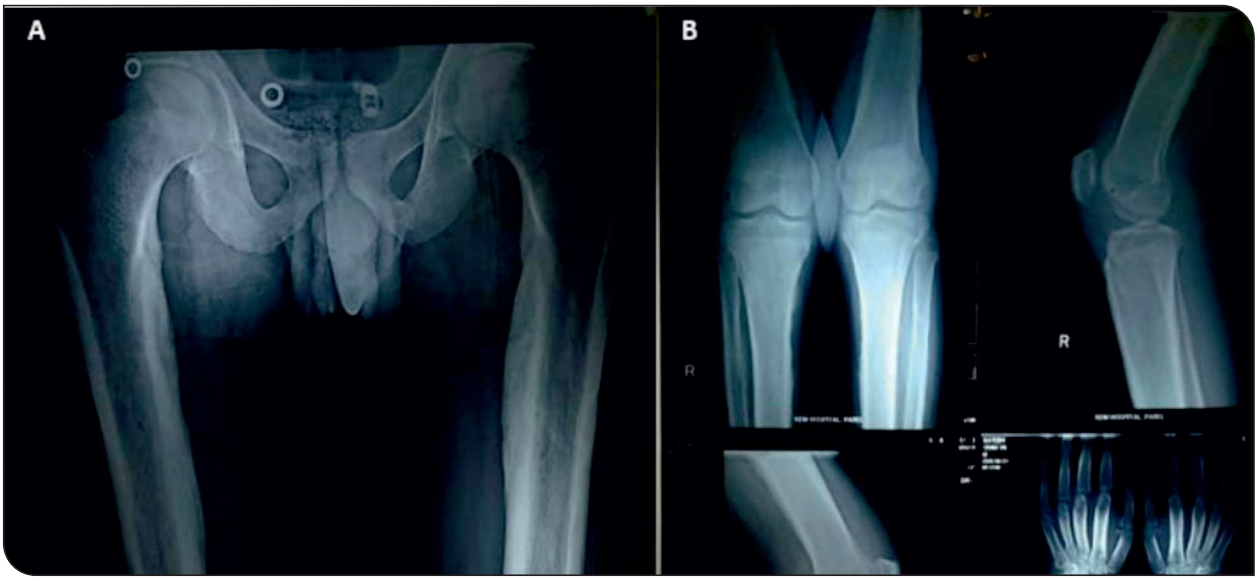

Figure 2: A. X-ray of bilateral Hip joint, antero-posterior view- shows enlargement of diaphysis of bilateral femur with sub periosteal reaction and soft tissue swelling; **B.** X-ray of bilateral knee joint, antero-posterior and lateral views showing enlargement of the knee joints with subperiosteal reaction. X-ray of bilateral hand, wrist joint and forearm, antero-posterior view showing widening of the joints and subperiosteal reaction.

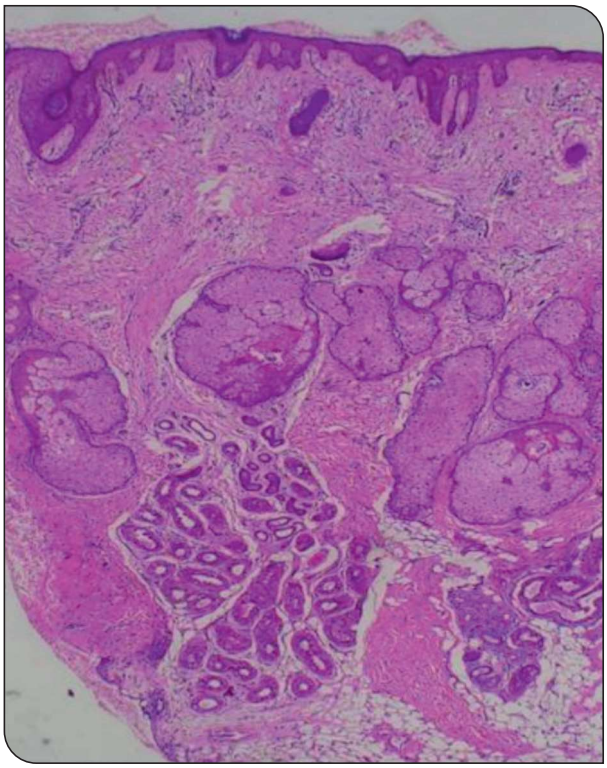

Figure 3: Histopathology of skin biopsy - hematoxylin and eosin stain at 10X magnification showed mucin deposition and interwoven thick collagen in the dermis with hyperplasia and hypertrophy of sebaceous and eccrine glands

DISCUSSION

Pachydermoperiostosis (PDP) or primary hypertrophic osteoarthropathy (PHO) was first described by Nikolaus Friedreich in 1868.[1] The clinical presentations are as follows: the complete form, with all three major symptoms; the incomplete form, with periostosis but without pachydermia; the forme frustra with pachydermia and minimal or no skeletal anomalies. It begins during childhood or adolescence and progresses gradually over the next 5 to 20 years before stabilizing. The disease has been mapped to chromosome 4q33-q34 and mutations in HPGD (15-hydroxyprostaglandin dehydrogenase,) the enzyme of prostaglandin degradation.[5,6] Increased levels of (PGE2) resulting from defective degradation due to gene mutation HPGD and SLCO2A1, appear to contribute to the pathogenesis of PDP. PGE2 can mimic the activity of osteoblasts and osteoclasts, which may be responsible for the acro-osteolysis and periosteal bone formation and can explain digital clubbing. Other manifestations include coarse facial features, seborrhoea, and symmetrical polyarthritis in 20 to 40% patients due to endothelial cell activation, palpebral ptosis d/t sebaceous gland hyperplasia and dermal mucin deposition, hyperhidrosis, acne, cutis verticis gyrate, acro-osteolysis of long bones, corneal leucoma, pre-senile macular dystrophy, cataract formation.[1,2] Although it is known that one third of PDP patients have a family history, the absence of the same in this case and presence of consanguinity makes autosomal recessive inheritance or an autosomal mutation a possible cause.

A normal level of IGF-1 is strong evidence that the patient does not have acromegaly. Other conditions which must be ruled out include syphilitic periostitis, thyroid acropachy, lepromatous leprosy. Thyroid achropachy is a hyperthyroid state which occurs in 0.3% patients with graves disease and can lead to similar manifestations years after treatment but no such history was obtained in our patient. Secondary HOA could be associated with chronic kidney disease but that wasn't the case here as patient had chronic NSAIDs usage. The diagnosis is based on the presence of two of the four criteria by Borochowitz, which are a history of familial transmission; pachydermia; digital clubbing; and skeletal manifestations, such as pain or radiographic signs of periostitis. Presently there is no specific treatment for this disease. Non-steroidal anti-inflammatory drugs and corticosteroids are used in symptomatic patients to alleviate polyarthritis. Retinoids play an important role for alleviating the seborrheic symptoms. Rhytidectomy is indicated for aesthetic reasons to change the appearance of the facial and scalp furrows. Bilateral blepharoplasty, tarsal wedge resections can be helpful in ocular involvement.[4] The diagnosis of PDP is clinical but due to its rarity the patient remained undiagnosed for many years. Our patient was referred to the Dermatology department for eyelid complaints, it is thus important to consider PDP as a possibility earlier in the course of the disease. Such patients need to be followed up chronically. Considering the pathogenesis, there might not be a complete cure however patient can be given symptomatic relief on and off with monitoring of causes of HOA and adverse effects of retinoids.

CONCLUSION

Pachydermoperiostosis is a rare genetic condition diagnosed clinically on the basis of its clinical features. However, the secondary form has to be ruled out with the help of investigations to look for any underlying systemic disorders. Thus the management involves a multimodality approach.

Conflict of interest: Nil

REFERENCES

1. Sandoval AR, Flores-Robles BJ, Llanos JC, Porres S, Dardón JD, Harrison RM. Cutis verticis gyrata as a clinical manifestation of Touraine-Solente-Gole' syndrome (pachydermoperiostosis). BMJ Case Rep. 2013, 2013. doi: 10.1136/bcr-2013-010047
2. Lee S, Park SY, Kwon HJ, Lee CH, Kim OH, Rhee Y. Identification of the mutations in the prostaglandin transporter gene SLCO2A1 and clinical characterization in korean patients with pachydermoperiostosis. J Korean Med Sci. 2016;31:735---4 doi: 10.3346/jkms.2016
3. Abdullah NRA, Jason WLC, Nasruddin AB. Pachydermoperiostosis: a rare mimicker of acromegaly. Endocrinol Diabetes Metab Case Rep. 2017:2017. DOI: 10.1530/EDM-17-0029
4. Monteiro E, Carvalho P, Silva A, Ferraro A. Frontal rhytidectomy: Anew approach to improve deep wrinkles in a case of pachydermoperiostosis. Plast Reconstr Surg 2003;112:1189-90.
5. Uppal S, Diggle CP, Carr IM, Fishwick CW, Ahmed M, Ibrahim GH, et al. Mutations in 15-hydroxyprostaglandin dehydrogenase cause primary hypertrophic osteoarthropathy. Nat Genet. 2008;40:789---93.
6. Nakazawa S, Niizeki H, Matsuda M, Nakabayashi K, Seki A, Mori T, et al. Involvement of prostaglandin E2 in the first Japanese case of pachydermoperiostosis with HPGD mutation and recalcitrant leg ulcer. J Dermatol Sci. 2015;78:153-5.