

CASE REPORT

PACHYDERMOPERIOSTOSIS: COMPLETE FORM WITH RARE DENTAL AND NEW OCULAR CHANGES

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ABSTRACT: Pachydermoperiostosis or Primary Hypertrophic Osteoarthropathy is a rare genetic disease affecting both the skin and bones. It is characterized by pachydermia, digital clubbing and periostosis. Its pathogenesis is uncertain and the condition affects mainly men. Here, we report a complete form of pachydermoperiostosis with rare dental changes and new ocular findings.

KEYWORDS: Pachydermoperiostosis, Primary Hypertrophic Osteoarthropathy, Ocular findings, Periodontitis, Alveolar bone disease

INTRODUCTION: Pachydermoperiostosis (PDP), the primary or idiopathic form of hypertrophic osteoarthropathy (HOA), is characterized by pachydermia, clubbing of digits of both hands and feet, and periostosis. It is a rare genetic disorder with autosomal dominant transmission with variable penetrance or autosomal recessive transmission. Approximately 5 percent of HOA are primary and predominantly occurs in men (7–9:1). Cases of secondary HOA are more common and are associated with underlying cardiopulmonary diseases and malignancies.¹⁻⁵ We report a case of complete form of PDP with rare dental features and new interesting ocular features.

CASE REPORT: A 32 year old unmarried male, born out of third degree consanguineous marriage, presented with distinctive thickening and exaggeration of skin folds over the face and scalp, enlargement of hands and legs, and pain and swelling of knees and ankles. He noted the changes since adolescence. He also had loss of few teeth in both jaws since 25 years of age. All these physical changes disturbed his quality of life, prevented him from getting married and finally made him seek medical advice. No other family members were affected. On physical examination, pachydermia and furrowing of skin over forehead and chin, seborrhoea, cutis verticis gyrata of the scalp, bilateral clubbing of fingers and toes were seen (Figure 1).

Increased volume of both upper and lower limbs was observed. Both knee and ankle joints were swollen and tender. Oral examination revealed absence of all upper incisors, lower right and left central incisors, right second and third mandibular molars and left mandibular molar. Patient uses fixed partial dentures. Ophthalmological examination revealed thickened eyelids with mechanical ptosis and straightening of eyelashes. Vision was normal.

Fundus examination revealed tortuous retinal veins with hyperaemic optic disc with hypopigmented peripheral retina. (Figure 2) All other systems were normal. Hematological and biochemical investigations were normal, except his hemoglobin was 10.9 grams per decilitre and peripheral smear showed predominant normocytic normochromic picture with few microcytic hypochromic red blood cells.

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Hormonal assays such as thyroid profile, growth hormone and parathormone were normal. Skin biopsy specimen showed normal epidermis with follicular plugging with sebaceous hyperplasia and focal hyalinization of dermal collagen with scanty perivascular and periadnexal inflammatory infiltrate.

A complete radiological examination of the skeletal system revealed the following findings:

1. Cortical thickening in the skull vault;
2. Periostosis of distal ulna, radius, metacarpals, proximal and middle phalanges;
3. Enlarged metaphysis with cortical thickening of femur bilaterally;
4. Periostosis of distal fibula, tibia, calcaneum and talus with thickening of interosseous membrane;
5. Early osteoarthritic changes of knee and ankle joints.
6. MRI of knees and legs show altered medullary signal intensities, periosteal and endosteal reaction with cortical and interosseous membrane thickening.
7. Oral pantogram showed absence of upper four incisors, lower central incisors, two lower molars on right side and with impacted wisdom tooth on the left side. There is severe generalized vertical alveolar bone loss between the teeth suggestive of periodontitis. (Figures 3a, 3b, 3c, 3d, 4a, 4b, 4c, 4d, 5) Other radiological investigations like chest radiograph, ultrasound of abdomen and echocardiogram were normal.

DISCUSSION: PDP was first reported by Friedrich in 1868 and was named and characterized as the primary form of HOA by Touraine, Solente and Golé in 1935. The disease presents with three possible presentations: a complete form showing pachydermia, periostosis and acropachy; the incomplete form showing only periostosis and acropachy; and the forme fruste or minimal/mild form showing only pachydermia with minimal or absent bone modifications.⁴

The disease typically appears in infancy and adolescence, progresses for a number of years (5–20 years), and then stabilizes.¹⁻³ PDP is defined by major and minor criteria. The three major criteria are pachydermia, periostosis, and digital clubbing. The minor criteria include seborrhoea with sebaceous hyperplasia, folliculitis, acne, hyperhidrosis, and cutis verticis gyrata.¹ Pachydermia, which predominantly affects the face and limbs, is the most frequent skin symptom.¹⁻³

The principal complications concern the joints, with arthralgia, arthritis, hydrarthrosis and haemarthrosis, which may occur symmetrically in the major joints and in the small joints of the hands and feet.⁴ Radiologically, acro-osteolysis, periosteal changes of short, long and flat bones, and ossification of ligaments and interosseous membranes have been reported. Additional features such as myelofibrosis, gastrointestinal abnormalities (peptic ulcer, chronic gastritis, Crohn's disease, Ménétrier's disease) and a number of occasional findings including compressive neuropathy, corneal leukoma, hypoplastic internal genitalia, gynecomastia, periodontal and alveolar bone abnormalities have also been reported in single patients.⁵

The exact pathology is unknown. The skin alterations could be secondary to the platelet and fibroblast dysfunction, alteration of glycosaminoglycans and proteoglycan synthesis.⁶ The agent responsible for PDP could be a unique circulating substance (such as a hormone) which abnormally rises in the plasma of affected individuals – such as osteocalcin, endothelin – 1, β – thromboglobulin, Platelet Derived Growth Factor, von Willebrand factor and Vascular Endothelial Growth Factor.

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Mutations in the gene coding for 15-hydroxyprostaglandin dehydrogenase located on chromosome 4q33 – 4q34, which leads to high maintained concentrations of prostaglandin E2, a mediator in some process involved in digital clubbing, skin thickening and periostosis.⁵⁻⁷

Our case is a complete form of this syndrome with presence of all the characteristic clinical and radiological findings. Although this syndrome has association with heredity,¹⁻⁴ but in this case, there is no history of any of the patient's relatives being affected with one or more similar features. Curiously, our patient had unique ophthalmic features like increased tortuosity of retinal veins, hyperaemic optic disc and hypopigmented peripheral retina.

These changes did not result in any visual disturbances to the patient. Such changes were not described in the literature. We are not sure whether these ophthalmic features are a part of this syndrome or incidental findings. We also report a case of teeth loss secondary to alveolar bone loss and periodontitis which was reported in a single patient.⁵

Though PDP is a benign condition, the disease creates social stigma and leads to considerable reduction in patient's quality of life. Hence these patients should be counselled by a team that includes dermatologists, rheumatologists and psychiatrists.

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Fig. 1: Prominent pachydermia with furrowing over the face and clubbing of all the fingers.



Fig. 1

Fig. 2: Hyperaemia of the fundus with venous engorgement and tortuosity.



Fig. 2

Fig. 3 'a': Radiograph of the skull showing frontal bone thickening.



Fig. 3a

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Fig. 3 'b': Radiograph of hand and wrist showing osteitis with enlargement of distal ulna and radius, metacarpals, phalanges.



Fig. 3b

Fig. 3 'c': Radiograph oblique view of both legs and ankle showing interosseous membrane thickening, ossification of ligaments, irregular periosteal proliferation, cortical thickening of distal tibia, fibula, calcaneum and talus with osteoarthritis of ankle joint.



Fig. 3c

Fig. 3 'd': Radiograph of both legs shows cortical thickening without narrowing of medullary cavity, periosteal thickening.



Fig. 3d

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Fig. 4 'a': MRI sagittal proton density sequence with fat suppression in knee joint picture showing altered medullary signal intensities.

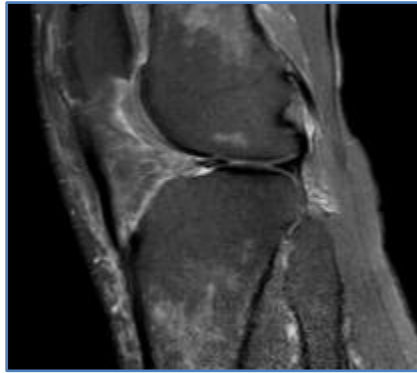


Fig. 4a

Fig. 4 'b': MRI coronal proton density sequence knee joint showing altered medullary signal intensities and endosteal reaction.



Fig. 4b

Fig. 4 'c': MRI proton density fat suppressed axial image right leg showing periosteal reaction, cortical thickening and interosseous thickening.

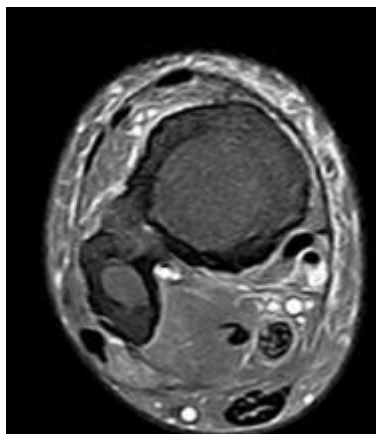


Fig. 4c

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Fig. 5: Oral pantogram showing absence of all upper incisors, lower central incisors, right lower lateral two molars and impacted lower molar on the left side with generalized severe alveolar bone loss – periodontitis.

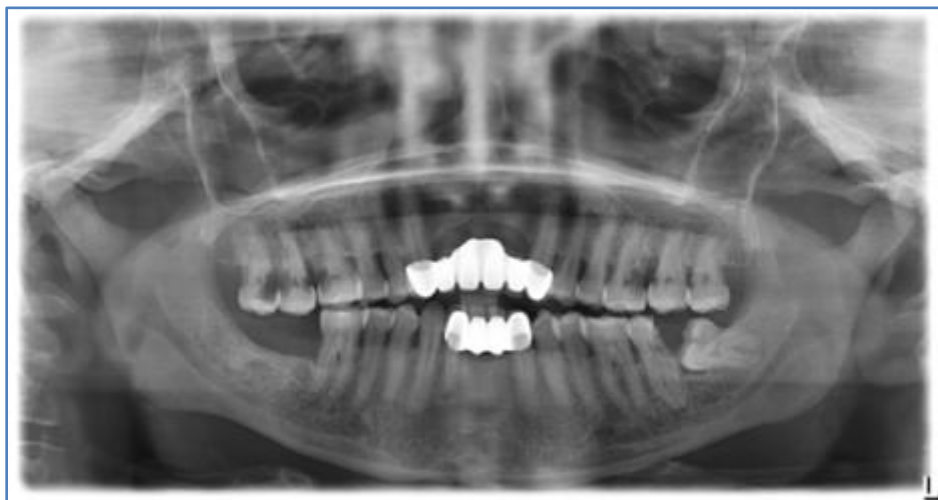


Fig. 5

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