## Case 14469

# Eurorad ••

#### Pachydermoperiostosis - A rare

#### radiologic case

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DOI: 10.1594/EURORAD/CASE.14469 ISSN: 1563-4086 Section: Musculoskeletal system Area of Interest: Musculoskeletal system Procedure: Diagnostic procedure Imaging Technique: Ultrasound Imaging Technique: Digital radiography Special Focus: Inflammation Pathology Case Type: Clinical Cases Authors: Dr. Akanksha Jain1, Dr. Shital K Turakhia2, Dr. Pratik Tak3 Patient: 60 years, male

#### **Clinical History:**

The patient had complaints of clubbing, swelling of multiple joints and thickening of the facial skin since adolescence. The patient complained of excessive sweating and feeling of heat and burning sensation in palms and soles. Thyroid profile, growth hormone assay, tests for syphilis and smears of skin for AFB were unremarkable. **Imaging Findings:** 

Symmetric, exuberant, shaggy subperiosteal bone formation was seen in both forearms and legs. There was expansion of the distal ends of radius and ulna and the proximal ends of tibia and fibula, with a reduction in the radiocarpal and femorotibial joint spaces. There was reduction in joint spaces of the proximal and distal interphalangeal joints. Periarticular osteopenia and resorption of the distal phalanges was noted with associated soft tissue swelling of distal fingers and toes. Widening of the base of distal phalanx of all fingers of both hands was seen. Enlargement and remodelling of sesamoid bones in both hands and feet was noted.

The images showed collapse of calcaneum and calcification of tendoachilles. There was a symmetric and shaggy periosteal reaction along the lower lateral aspect of the iliac bones.

There was evidence of metaphyseal widening of the bilateral femur with cortical thickening and widening of the shafts.

There was mild hyperostosis of the skull bones in the parietooccipital region. The sella turcica appeared normal. **Discussion:** 

PDP is a rare form of hypertrophic osteoarthropathy with no known cause and hence it is called idiopathic or primary hypertrophic osteoarthropathy (PHO). PDP accounts for 3–5% of cases of hypertrophic arthropathy and affects males more often than females (7:1).

In up to one-third of the patients, PDP occurs as an autosomal dominant hereditary disease. Some case reports suggest that it may be an X-linked disease. [1]

Deficiency of the prostaglandin transporter (SLCO2A1) has been characterized as the main cause of PHO. Touraine et al. described three forms of PDP, viz, classic or complete form, with skin and skeletal changes; incomplete form, with skeletal changes but no dermal findings; and forme fruste with dermal changes but no skeletal findings. [2]

Facial involvement occurs in the form of thickening of the facial skin and scalp, with prominent folds on forehead and

cheeks. Sometimes, the scalp takes on an undulating appearance and shows prominent grooves, the appearance called cutis verticis gyrata. Cutis verticis gyrata can also be seen in a variety of other conditions, including neurofibromatosis, DM, myxoedema, acromegaly, etc., and in syndromes including Turner's syndrome, tuberous sclerosis, etc., hence it is not pathognomonic for PDP. [3]

Skeletal findings include symmetric, shaggy subperiosteal bone formation in the long bones, especially of forearm and leg. Involvement of the epiphyseal region distinguishes it from the secondary form, in which epiphyses are usually spared. [4] There is widening of the ends of bones, especially at wrist and knee joints. A prominent feature is enlargement of distal part of the digits with resorption of distal phalanges and calcification of ligaments and interosseous membranes. In later stages, cortical thickening with narrowing of medullary cavity may be seen. Enlargement of sinuses may be seen uncommonly. Bone scintigraphy may reveal increased tracer uptake by the cortex in diaphyseal and metaphyseal regions. [5, 6] Hyperostosis of the calvaria and skull base bones is common. [4]

Joints affected in PDP show swelling due to joint effusion, with evidence of chronic nonsuppurative inflammation. There is reduction in joint spaces, with relative preservation of articular surfaces. Late-onset deformities may occur, especially in the digits. [5]

Spinal manifestations are unusual but have been described.

Variants of PDP include Rosenfeld-Kloepfer syndrome, Currarino idiopathic osteoarthropathy and a localized form with only the radiographic features of PDP in the lower extremities. [6]

Treatment is limited to NSAID, steroid, or colchicine therapy to alleviate arthralgias and retinoids for dermal changes. Surgical treatment is limited to plastic surgery for cosmetic indications. [5, 6, 7].

**Differential Diagnosis List:** Pachydermoperiostosis, Secondary hypertrophic osteoarthropathy, Thyroid acropachy, Acromegaly, Van Buchem's disease, Syphilitic periostosis

Final Diagnosis: Pachydermoperiostosis

#### **References:**

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Athappan G, Unnikrishnan A, Chengat V et al (2009) Touraine Solente Gole syndrome: the disease and associated tongue fissuring. Rheumatol Int 29:1091-93 (PMID: <u>19050893</u>)



**Description:** Symmetric, shaggy and exuberant subperiosteal new bone formation. There is expansion of the proximal end of the tibia with a reduction in the femoro-tibial joint spaces. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** Exuberant and shaggy subperiosteal new bone formation. There is expansion of the distal end of the radius and ulna with a reduction in the radio-carpal joint space. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** There is reduction in PIP and DIP joint spaces. Periarticular osteopaenia and acroosteolysis with associated soft tissue swelling and contracture of distal fingers. Widening of the base of the distal phalanx of all fingers. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** There is evidence of collapse of calcaneum and calcification of Achilles tendon. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** Symmetrical and shaggy periosteal reaction along lower lateral aspect of iliac bones. There is metaphyseal widening of both femurs. Cortical widening and widening of femoral shafts is seen. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** Mild hyperostosis of the skull bones in parieto-occipital region. Sella turcica is normal. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** There is reduction in PIP and DIP joint spaces. Periarticular osteopaenia and resorption of the distal phalanges. Associated soft tissue swelling and contractres of toes. Enlargement and deformities of sesamoid bones. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India



**Description:** Mild effusion with thickened synovium bilaterally. The effusion was seen to extend into suprapatellar bursae. **Origin:** Akanksha J, Department of Radiology, BJMC, Ahmedabad, India