Pachydermoperiostosis: The Elephant Skin Disease

CASSANDRA HONG, MBChB, MRCP, MRCP (Rheum), MSc (Rheum), Department of Rheumatology, Guy's and St. Thomas' UK National Health Service (NHS) Foundation Trust; NATASA DEVIC, MBBS, MRCS, FRCR, Diploma in SEM RCSED, Department of Radiology, Guy's and St. Thomas' NHS Foundation Trust; ALI ZAVAREH, MD, FRCR, Department of Radiology, Guy's and St. Thomas' NHS Foundation Trust; AMIDEVI DESAI, MBBS, BSc (Hons), MRCP, FRCR, Department of Rheumatology, Guy's and St. Thomas' NHS Foundation Trust; NORA NG, BmedSci, BMBS, MRCP(Rheum), MD, Department of Rheumatology, Guy's and St. Thomas' NHS Foundation Trust. Address correspondence to Dr. C. Hong, Department of Rheumatology, Guy's Hospital, Great Maze Pond, London SE1 9RT, UK. E-mail: cassandra.hong@nhs.net. Ethics approval was not required in accordance with the policy of the authors' institution. J Rheumatol 2017;44:1680–1; doi:10.3899/jrheum.170102

Pachydermoperiostosis (PDP) was first described in 1868. Three forms have been identified: complete (clubbing, periostosis, pachyderma, and cutis verticis gyrata), incomplete (without cutis verticis gyrata), and forme fruste (pachyderma with minimal skeletal changes)[1]. Two genes have been associated, *HPGD* and *SLCO2A1*[2].

A 27-year-old British Pakistani man presented with clubbing, hyperhidrosis, palmoplantar hyperkeratosis, and painful swollen knees and ankles since childhood that responded to intraarticular injections. He had pronounced facial skinfolds, excessive facial oiliness but no ptosis. His parents were first cousins. He had symmetrically enlarged edematous legs with ankle swelling and underlying joint deformity (Figure 1). Blood tests showed raised inflammatory markers.

The incidence of PDP is unknown, but it is more common in Asia. Estimated prevalence is 0.16%[3] with increased severity in males[4]. Diagnostic criteria require at least 2 of family history, clubbing, hypertrophic skin changes, bone pain, or radiographic changes[5]. Although autosomal dominant trait with incomplete penetrance and variable expression has been proven, autosomal recessive, X-linked inheritance, and consanguinity have been suggested. Osteoarthropathy usually begins during childhood, but does not affect life expectancy. However, dermoskeletal changes persist for life. Skeletal involvement includes arthralgia, joint enlargement, subperiosteal ossification (Figure 2), acroosteolysis, and osteoporosis. Management is symptomatic. The patient fulfilled diagnostic criteria for primary PDP, but was negative for *HPGD* and *SLCO2A1*, raising the possibility of

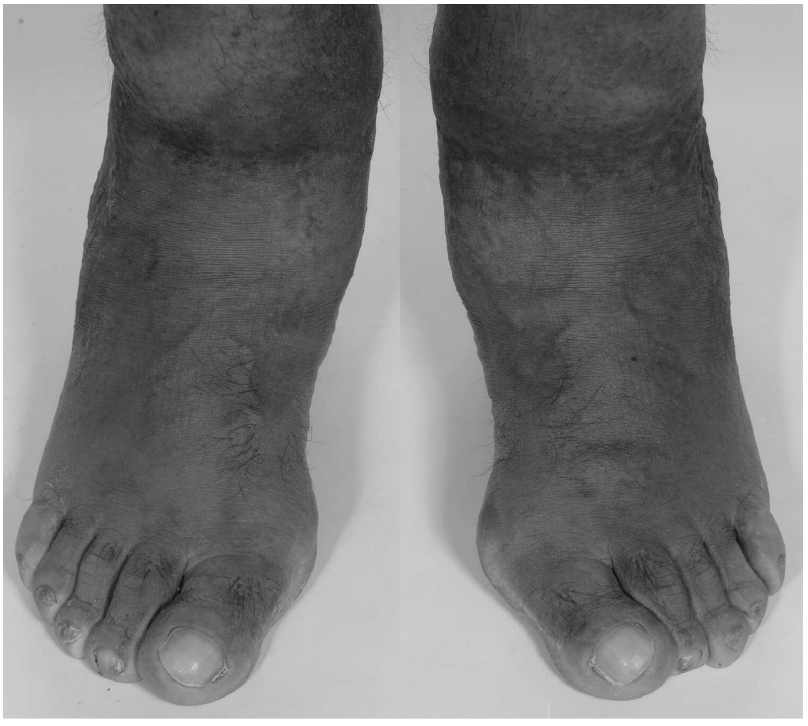

Figure 1. Hypertrophy of feet and ankles with edema and underlying foot deformity.

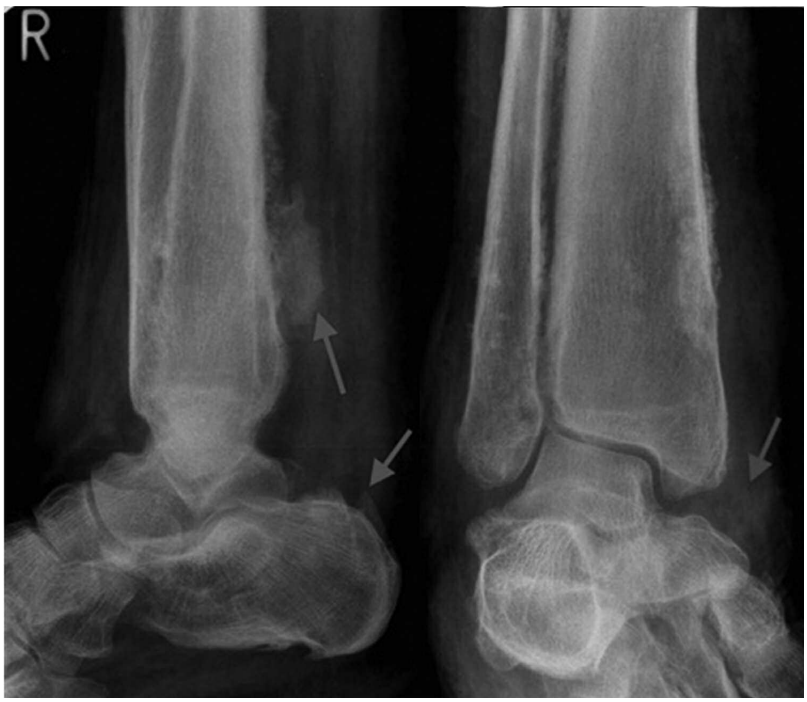

Figure 2. Anteroposterior and lateral radiographs of the right ankle show florid subperiosteal bone formation at the distal tibia and fibula, and also the calcaneum (arrows).

an undiscovered mutation. Zoledronate acid provided some benefit. Other management options include knee synovectomy; however, the patient declined.

PDP is a rare entity that should be differentiated from secondary hypertrophic osteoarthropathy, thyroid acropachy, acromegaly, and chronic inflammatory rheumatic diseases.

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