Pachydermoperisotosis

Sir,

We read with a great interest the article entitled "Enlarged hands and feet-Not always acromegaly" by Gnatnatti *et al.*, published in IJEM 2012;16(Suppl 2):S318-20, and would like to add our experience about the fact that pachydermoperiostosis (PDP) can be easily mistaken for acromegaly.

PDP also called primary hypertrophic osteoarthropathy (PHO), or idiopathic hypertrophic osteoarthropathy (IHO), or hereditary hypertrophic osteopathy (HHO), or Touraine-Solente-Gole syndrome is a rare genodermatosis[1] which is apparently inherited in a dominant pattern with variable penetrance. But, both autosomal recessive and X-linked inheritances have been suggested.[1] Recently, novel mutations have been reported in some Turkish,[2] and Chinese[3] families. Clinical modifications are thought to be related to excessive collagen formation and dysregulation of matrix proteins because of fibroblastic hyper activation.[4]

PDP real incidence is unknown, but is prevailing in males with a sex ratio equal to 9:1. The disease mainly affects skin, but also bones. It is characterized by a thickening of the skin or pachydermia, excessive bone formation (or periostosis), and finger clubbing. Fingers modifications are represented by tissue swelling with loss of normal angle between nail and nail bed. Other manifestations are painful arthritis, cutis verticis gyrate (CVG) which is a condition of the scalp consisting of deep grooves and convolutions that resemble the surface of the brain. Other signs are hyperhydrosis, seborrhea, edema, and eyelid hypertrophy.

There are two forms: The primary PDP in which no cause can be found, and the secondary form which is associated to several diseases and neoformations. The last one is considered as a paraneoplastic syndrome[5] which should be excluded before thinking to genetic form.

For the management, as there is not any available specific treatment, genetic counseling should be offered to patients and their family to prevent new cases. Plastic surgery and botulinum toxin type A may help in complete forms. Joints pain can be treated by non steroidal anti-inflammatory agents or with corticoids. Biphosphonates are sometime used with success.

Progressive skin and bones modifications raise differential diagnosis with acromegaly. But, contrary to acromegaly face and head bones are usually not involved in PDP and growth hormone (GH) is normal as in the following observation:

A young man aged 26, with a family history of

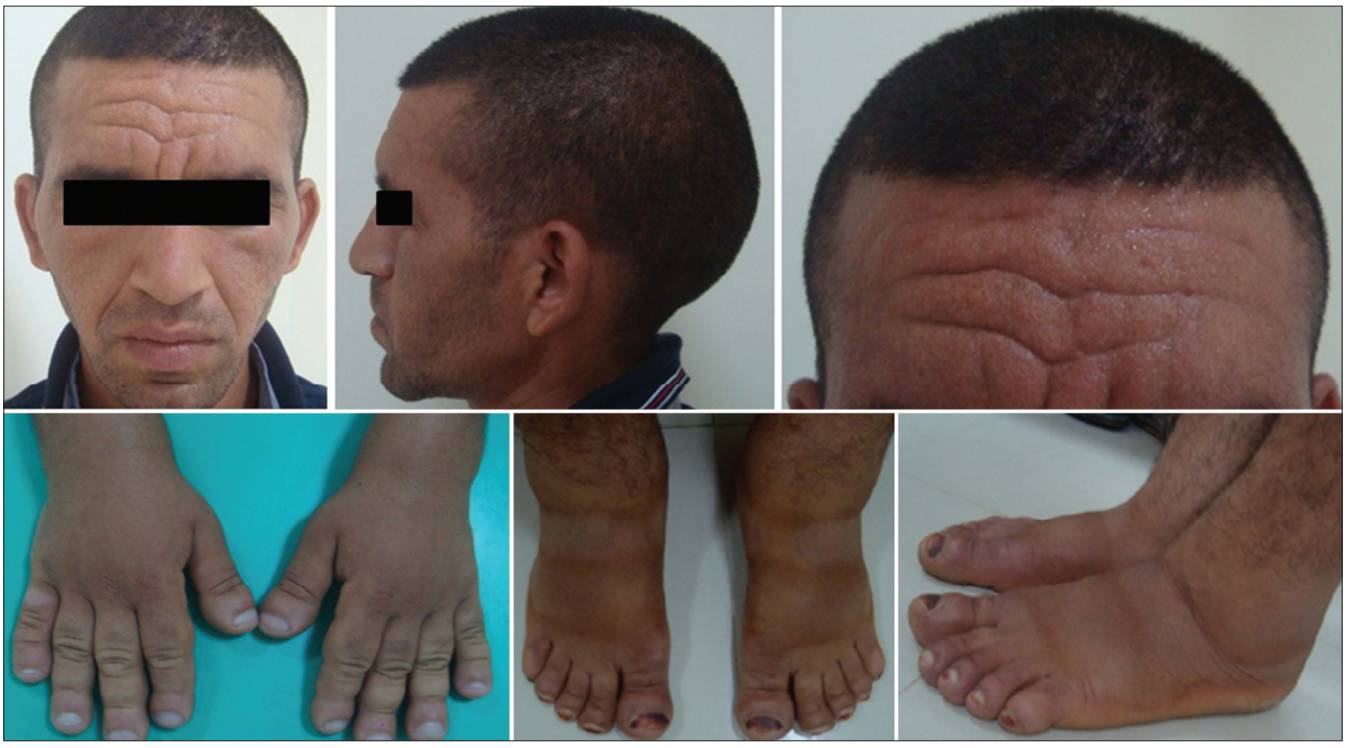

Figure 1: Pachydermoperiostosis (PDP) with hands and feet enlargement with finger clubbing. Remark the nondysmorphic face but with mild cutis verticis gyrate and hyperhydrosis

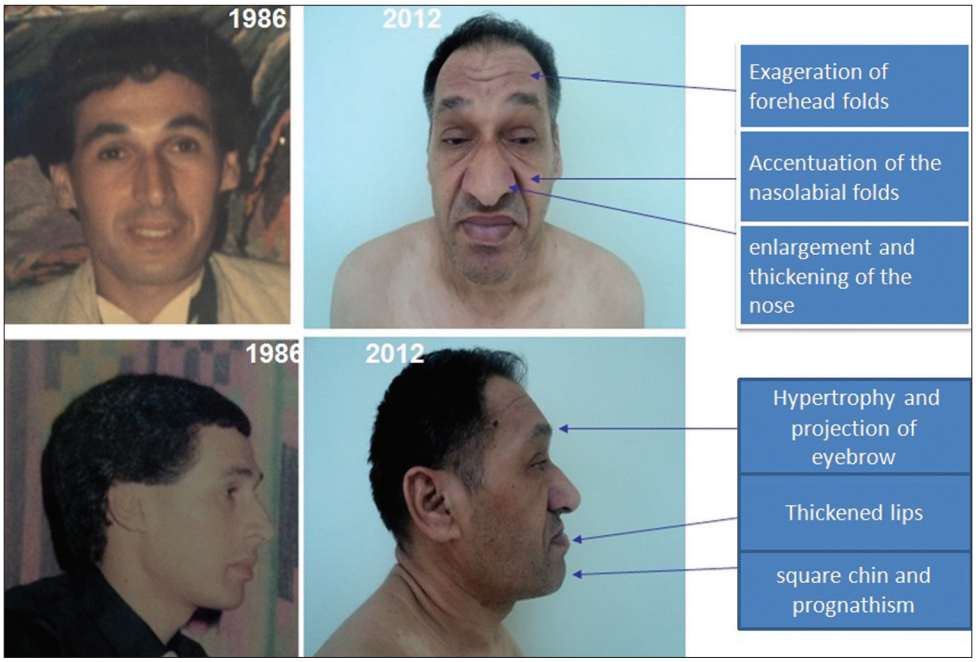

Figure 2: Acquired modifications of the face in acromegaly. Remark the difference with PDP

diabetes mellitus, high blood pressure, renal lithiasis, hyperhydrosis, and large hands (one brother), was sent to our unit for suspicion of acromegaly. His disease began when he was 10 marked by an enlargement of his hands and feet with hyperhydrosis. During the last 3 years, another phenomenon has appeared as he suffered from joint pains and exaggeration of hyperhydrosis. Medical history and clinical examination were against acromegaly and pleaded for PDP [Figure 1], because except for skin abnormalities the face was not concerned by bone hypertrophy. Research for other diseases and neoformations was negative. Biological assessment showed normal GH = 0.07 ng/ml contrary to acromegaly where GH is increased leading to acquired dysmorphic syndrome concerning hands and feet, but also the face as in the Figure 2.

Farida Chentli, Leyla Rabhi, Nassima Belhadj-Aissa, Said Azzoug

Department of Endocrinology and Metabolism, BabEl Oued Universitary Hospital, Algiers, Algeria

Corresponding Author: Prof. Farida Chentli, Department of Endocrinology And Metabolism, Bab El Oued Hospital, 5 boulevard Said Touati Algiers, Algeria.
E- mail: chentli.f@gmail.com

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