



Pachydermoperiostosis in its Complete Family form

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Abstract

Introduction: Pachydermoperiostosis (PDP) or primary hypertrophic osteoarthropathy (OAH) is a rare hereditary disorder. We report the case of a family with PDP in its complete form.

Clinical Case: We report the case of a 25-year-old man suffering from polyarthralgia with palmoplantar hypertrophy. Its history began in 2015 with the progressive installation of diffuse polyarthralgia more pronounced in large joints (knees, ankles, elbows) with diffuse bone pain and a history of pathological fracture of the left 3 metatarsal without trauma. The patient described hyperhidrosis, seborrhoea and increased thickening of the forehead folds and palmoplantar hypertrophy during the last 3 years. Skin examination noted oily skin, glistening and puffy, pachydermia of the extremities with a pleated thickening of the forehead, without ptosis. Palmoplantar hypertrophy and digital clubbing were evident with pudgy fingers and toes. The patient shod 44 against 41 three years ago. The clinical examination also found palmoplantar hyperhidrosis and free joints, with no inflammatory signs. The diagnosis of PDP was retained, and the patient was put on colchicine. We noted consanguinity of the parents and the physical examination of the other members of the family found the same manifestations in the father and the brother with variable intensity.

Conclusion: PDP is a benign genetic affection that primarily affects the skin and bones. Its pathophysiological mechanism remains unclear.

Keywords: Pachydermoperiostosis; Primary Hypertrophic Osteoarthropathy; Cutis Verticis Gyrate

Introduction

The pachydermoperiostosis (PDP) or osteoarthropathy hypertrophic primitive (OAH) is an inherited disorder rare little known with a transmission mode discussed [1,2]. It is a radio clinical entity associating digital hippocratism, arthropathy, skin thickening and periostitis. The differential diagnosis can sometimes arise with acromegaly or secondary HAE but also with chronic inflammatory rheumatism due to joint manifestations [3]. We report the observation of a family with PDP in its complete form.

Clinical case

We report the case of a 25-year-old man referred to the internal

medicine consultation for polyarthrititis with palmoplantar enlargement.

Its history began in 2015 with the progressive installation of diffuse polyarthralgia more marked in the large joints (knees, ankles, elbows) with diffuse bone pain and a history of pathological fracture of the left 3 metatarsal without notion of trauma.

The patient described a sweating, a seborrhoea and accent thickening of forehead wrinkles and Palmoplantar hypertrophy at the end of the last 3 years (Figure 1).



Figure 1: Photographs of the patient's face at 23 and 25 years of age. Appearance of plicated pachydermia: thickening and widening of facial features.

Examination cutaneous - mucosal noted oily skin, shiny and puffy, a pachyderm ends with a pleated thickening the front, without ptosis. Hypertrophy Palmoplantar and clubbing were important (Figure 2). The patient wearing shoes 44 against 41 three years ago. The clinical examination also found palmoplantar hyperhidrosis and free joints, with no inflammatory signs.



Figure 2: Hands and feet of the patient. Enlargement of bone structures with a massive and stocky appearance associated with digital hypertrophy.

Standard radiographs showed periosteal thickening affecting long bones, metacarpals, metatarsals and even phalanges (Figure 3).

Biological assessment has not objectified abnormality: the erythrocyte sedimentation rate (ESR) was 10 mn, C-reactive protein (CRP) was 3 mg/L and the hormonal assays of T4, TSH were

normal as well as the hepatic and renal assessments. The growth hormone (GH) rate was 0.59 ng/ml (normal range for age reference: < 2.47 ng/ml) and serum Insulin-like growth hormone factor (IGF-1) level 162 ng/ml (normal range for age reference: 158~230 ng/ml).



Figure 3: X-ray of the hands showing periostosis in the phalanges.

The brain scan did not show any abnormality, notably the absence of pituitary adenoma. A thoraco- abdominal-pelvic computed tomography objectified beginning pulmonary emphysema (Figure 4), presence of three hypodense hepatic nodules (angiomas) and a dilated stomach with thickened wall (Figure 5). The endoscopy gastroesophageal showed a congestive bulbitis with presence of large fundic folds without loss of substance. The echocardiography did not show any abnormality.

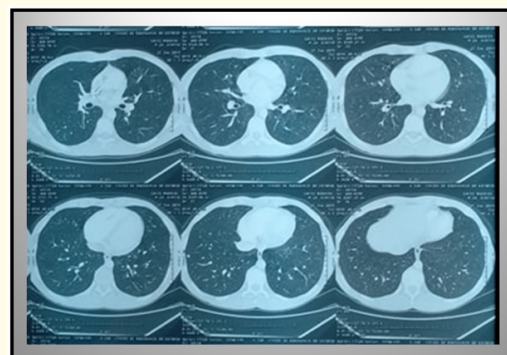


Figure 4: A chest CT (computed tomography) scan showing beginning pulmonary emphysema.

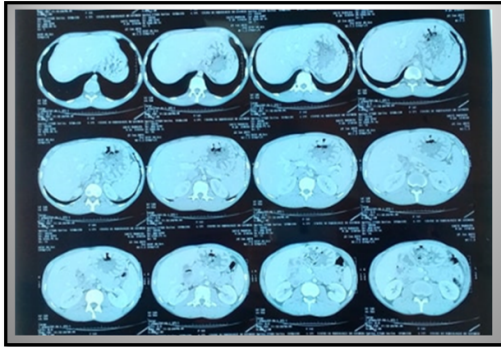


Figure 5: Abdominal CT (computed tomography) scan showing presence of three hypodense hepatic nodules (angiomas) and a dilated stomach with thickened wall.

The diagnosis of PDP was accepted, and the patient was put on colchicine.

The interrogation noted the notion of parents' consanguinity and the physical examination of the other family members found the same manifestations in the father and the brother with variable penetrance. Noting that the father appears describing the same symptomatology at the age of 22 years and was already tracking rheumatology for polyarthralgia broadcasts for which he was taking anti-inflammatory drugs and the painkillers for more than 7 years. The father had a more marked "cutis verticis gyrata" and a pachydermia (Figure 6 and 7).



Figure 6: Manifest pachydermia on the father's forehead.



Figure 7: "Cutis verticis gyrata" in the father.

Discussion

Primary PDP or OAH, described by Solente and Golé in 1935, is a rare affection of a familial nature (3.5% of OAH) [1,2] which is transmitted in an autosomal dominant mode with variable penetrance and phenotypic expressiveness [4] such is the case with our patients. Cases of autosomal recessive transmission have been reported as well as consanguinity [5]. all age groups can be affected, but especially around the age of 20 with a male predominance [6]. Its diagnosis is clinical radiology after having eliminated all other causes of secondary HAE. Sometimes dysmorphic syndrome can mimic acromegaly, but hormonal dosages correct the diagnosis.

Bronchitis proposed diagnostic criteria for PDP [7]. The presence of at least 2 of the following 4 criteria is necessary: the notion of family transmission; pachydermia; digital hippocratism; painful or radiological bone manifestations of periostitis. In our observations the 4 criteria were present.

Touraine, Solente and Golé have distinguished 3 clinical forms of PDP [8]:

- The complete form which is the most common and as observed in our cases. It includes a skin alteration such as placated pachydermia, "cutis verticis gyrata", sometimes a ptosis, an enlargement of the sebaceous and sweat glands, periostitis and digital Hippocrates with palmo-plantar hypertrophy
- The incomplete form, described by Currarino in 1961 [9], characterized by the presence of a primary osteoarthropathy with delayed closing of the fontanelles but without associated cutaneous involvement. The rough form associating an important pachydermia with minimal osseous signs. In this family it seems that we are facing a complete form of PDP with a typical skin and bone involvement.

- There is no unambiguous treatment for PDP, but rather it depends on the clinical forms and the degree of skin and bone damage. Conventional treatment includes analgesics, non-steroidal anti-inflammatory drugs and colchicine, sometimes even special preferences for Phenylbutazone [10, 11] In some cases cosmetic surgery may be useful in manifest facial dysmorphism [12]. A recent study has shown the effectiveness of etoricoxib and aescin in delaying the progression of pachydermia or even improving skin involvement [13].

Conclusion

The PDP is an affection benign gene that affects primarily the skin and bone. Its pathophysiological mechanism remains poorly understood. Radiology plays an essential role in positive diagnosis. But this rare entity poses diagnostic problems with secondary OAH and chronic inflammatory rheumatism.

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