CASE REPORT

Cutis verticis gyrata as a clinical manifestation of Touraine-Solente-Gole' syndrome (pachydermoperiostosis)

Aldo René Hurtarte Sandoval,¹ Bryan Josué Flores Robles,² Jaime Caceres Llanos,³ Salvador Porres,⁴ José David Penate Dardón,⁵ Rachael Marie Harrison⁶

¹Department of Internal Medicine, San Juan de Dios General Hospital, Guatemala, Guatemala

²Department of Rheumatology, Puerta de Hierro Majada Honda Hospital, Madrid, Spain

³Department of Rheumatology, San Juan de Dios General Hospital, Guatemala

⁴Department of Dermatology, San Juan de Dios General Hospital, Guatemala

⁵Department of Phlebology, Itzamna Day Hospital, Guatemala, Guatemala

⁶Department of General Medicine, San Juan de Dios General Hospital, Guatemala, Guatemala

Correspondence to

Dr Aldo René Hurtarte Sandoval,
aldohurtarte12@hotmail.com

SUMMARY

Cutis verticis gyrata is a descriptive term for a condition of the scalp consisting of deep grooves and convolutions that resemble the surface of the brain. We present a case of a 22-year-old man who presented with pain and swelling of both knees and hands. Enlarged wrists, ankles and feet were also noted, along with facial seborrhoea, thickening of the skin and deformity of the fingers. Physical examination of the scalp showed a cerebriform appearance with accentuating folds and deep furrows (cutis verticis gyrata), thickening in the face, frontal and parietal regions. Bone enlargement of the hands, knees, ankles and feet was also found. Secondary causes of pachydermoperiostosis were negative.

BACKGROUND

Cutis verticis gyrata (the least frequent skin manifestation of pachydermoperiostosis (PDP))¹ is a descriptive term for a condition of the scalp consisting of deep grooves and convolutions that resemble the surface of the brain. PDP is a rare syndrome as evidenced by the Medline search and we found a total of 286 cases reported. The diagnosis is established from clinical and radiological data.

CASE PRESENTATION

A 22-year-old man previously healthy, presented with pain and swelling of both knees and hands. Enlarged wrists, ankles and feet were also noted, along with facial seborrhoea, thickening of the skin and deformity of the fingers. He did not know of any history of similar conditions in family members. Physical examination of the scalp showed a cerebriform appearance with accentuating folds and deep furrows (cutis verticis gyrata), thickening in the face, frontal and parietal region (figure 1). Hyperkeratosis, palmoplantar hyperhidrosis and pachydermia of both hands with clubbing were also observed (figure 2). Swelling was observed in both knees (figure 3) with signs of joint effusion. Bone enlargement of the hands, knees, ankles and feet was also found.

INVESTIGATIONS

The radiograph of the hands and feet reported periosteal thickening in the tubular aspect of the metatarsals, femur and metacarpals (figures 4–7). The skin biopsy of the frontal region demonstrated elongated epithelial cones with increased collagen fibre content, which are the cutis verticis gyrata

Figure 1 Deep facial skin folds around the nose, mouth and on the forehead. Cerebroid folds on the scalp.

features described in PDP. Routine laboratory tests including thyroid function tests, adrenocorticotrophic hormone, cortisol, prolactin, rheumatoid factor, thoracoabdominal CT scan and arthrocentesis were normal. The diagnosis of PDP was made.

DIFFERENTIAL DIAGNOSIS

Acromegaly.

Figure 2 Clubbing and pachydermia of the hand.

To cite: Sandoval ARH, Robles BJF, Llanos JC, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2013-010047

Figure 3 Bilateral knee swelling.

OUTCOME AND FOLLOW-UP

The patient was treated with a keratolytic agent and non-steroidal anti-inflammatory drugs (NSAID) and referred to out-patient monitoring; however, he did not comply with the hospital.

DISCUSSION

Primary hypertrophic osteoarthropathy (HOA) (PDP) is a rare syndrome characterised by finger clubbing, periosteal new bone formation especially over the distal ends of long bones and coarsening of the facial features.²

First reported cases were the Hagner brothers, who had typical features of this syndrome. They were first described by Friedrich in 1868. It was only 45 years ago that PDP was first recognised as a distinct entity by Touraine, Solente and Gole (1935).¹ HOA is divided into primary and secondary forms. PDP, the primary form, accounts for 3–5% of all cases of HOA.

Figure 4 Metatarsals with periosteal thickening.

Figure 5 Bilateral diaphyseal expansion of tubular bones in the lower limbs.

Secondary HOA, also called pulmonary HOA, is associated with underlying cardiopulmonary diseases and malignancies.¹

Clinically, it is characterised by digital clubbing (89% of cases), pachydermia: thickening and wrinkling of facial features including the forehead and the nasolabial folds, with profound hypertrophy of the eyelids (30–40% of cases) and cutis verticis gyrata (24% of cases). The combination of thickened skin and bony enlargement can result in great thickening of the extremities, which is the most striking physical finding. Seborrhoea is noted in more than 90% of cases, with, sometimes, occurrence of acne lesions or folliculitis. Pubic and facial hair is almost always rare.¹ The diagnosis of PDP is based on the presence of at least two of the four criteria set by Borochowitz, which are a history of familial transmission, pachyderma, digital clubbing and skeletal manifestations, such as pain or signs of radiographic periostitis.¹

Gastric hypertrophy, gastric ulcer and other endocrine abnormalities have been described.⁴

Differential diagnosis must be with cases of HOAs are secondary to intrathoracic diseases. Thyroid acropachy is a rare manifestation of Graves' disease and is independent of the state of thyroid function. Clubbing usually coexists with exophthalmos

Figure 6 Outlining expansion of bilateral femurs without affecting the interarticular space.

Figure 7 Phalangeal and metacarpal widening.

and pretibial myxoedema. The presence of digital clubbing for several years in the absence of recognisable internal illness favours the diagnosis of PDP. Although some types of lung cancer may induce proximal skin hypertrophy, the occurrence of rough facial features or cutis verticis gyrata also favours the diagnosis of PDP. Acromegaly may superficially resemble PDP. The presence of clubbing and periostosis, plus the absence of prognathism, enlarged sella turcica or abnormal circulating concentrations of growth hormone products should lead to the correct diagnosis.⁵

Radiographs revealed diffuse periostosis along the length of bones, including epiphyses, in 80–97% of cases, and often with irregular contours.^{5 6}

In advanced cases, all tubular bones are affected: in addition to the diaphysis, the metaphysis and epiphysis are also involved and the periostosis takes on an irregular configuration. Periostosis has a symmetric distribution and evolves in a centripetal fashion. Typically it is preservation of joint space and the absence of erosions or para-articular osteopenia.^{5 7 8}

The disease is self-limited and usually stabilises after 5–20 years of onset, but may result in orthopaedic disability and neurological sequel. Standard treatment includes the use of NSAID for pain relief. Bisphosphonates like pamidronate or risedronate have been used extensively in PDP due to their anti-resorptive and osteoclast inhibitory properties.¹

Learning points

- ▶ Cutis verticis gyrata is a clinical manifestation of pachydermoperiostosis. This is a descriptive term for a condition of the scalp consisting of deep grooves and convolutions that resembles the surface of the brain.
- ▶ It can present in previously healthy individuals, with no family history. Physical manifestations may include pain and swelling of both knees and hands, enlarged wrists, ankles and feet; facial seborrhoea, thickening of the skin and deformity of the fingers.
- ▶ The diagnosis is established using a combination of clinical, histological and radiological findings.

Acknowledgements Thanks to Dr Ana Hualde Juvera.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Gupta M, Lehl SS, Singh R, *et al.* Touraine-Solente-Gole' syndrome. *BMJ Case Rep.* Published Online: 28 Sep 2011. doi:10.1136/bcr.08.2011.4605
- 2 Sinha GP, Curtis P, Haigh D, *et al.* Pachydermoperiostosis in childhood. *Br J Rheumatol* 1997;36:1224–7.
- 3 Christine P, Carr IM, Diggle, Zitt E, *et al.* Common and recurrent HPGD mutations in Caucasian individuals with primary hypertrophic osteoarthropathy. *Rheumatology* 2010;49:1056–62.
- 4 Rim A, Samar B, Fourati H, *et al.* Hypertrophy of the feet and ankles presenting in primary hypertrophic osteoarthropathy or pachydermoperiostosis: a case report. *J Med Case Rep* 2012;6:31.
- 5 Martínez-Lavin M. Pachydermoperiostosis. *Best Pract Res Clin Rheumatol* 2011;25:727–34.
- 6 Dyonísio Fernandes GC, Dos Santos Torres U, Fernandes TE Aídar, *et al.* A propósito de un caso Osteoartropatía hipertrófica Primaria. *Reumatol Clín* 2011;7:213–14.
- 7 Gómez Rodríguez N, Ibáñez Ruán J, González P, *et al.* Osteoartropatía hipertrófica primaria (paquidermoperiostosis). Aportación de 2 casos familiares. *Reumatol Clín* 2009;5:259–63.
- 8 Yu YL, Turck WPG. Pachydermoperiostosis (idiopathic hypertrophic osteoarthropathy). *Postgrad Med J* 1981;57:521–4.

Copyright 2013 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow