

## A case of primary hypertrophic osteoarthropathy

### *Osteoartropatía hipertrófica primaria: a propósito de un caso*

To the Editor:

Hypertrophic osteoarthropathy (HOA) is a rare syndrome characterised by arthritis, bilateral and symmetric periostitis, as well as bilateral clubbing in the hands and feet. It can be classified as primary (hereditary or idiopathic, which corresponds to less than 5% of cases) or secondary (frequently associated with intrathoracic neoplasms and inflammatory conditions).<sup>1,2</sup> We present a primary HOA case, interesting because there are classical clinical and image findings of this rare disease, and because of the prolonged period of symptom evolution until diagnosis.

The case is a 45-year-old male who consulted the Rheumatology Department complaining of polyarthralgia, mainly in the wrists, knees and ankles. This was associated with increased fingertip length and width from nearly 30 years ago. He indicated that there were no other constitutional or systemic complaints. He had no other relevant medical history and denied alcohol abuse or smoking. Physical examination revealed that he was in good condition, of normal colour, well hydrated, with no disturbances in the heart and lung auscultation; abdominal examination was normal. The neurological examination showed no changes. The patient also presented clubbing and “watch glass” fingernails (Figure A) and toenails (Figure B). He had an increased number of skin folds on the scalp and frontal region, as well as intensification of skin oiliness.

The patient was submitted to laboratory tests, which showed no relevant changes. The radiological study of the long bones showed cortical thickening with periosteal reaction (Figure C). Primary HOA was diagnosed from the clinical picture and imaging examinations, and after carefully excluding secondary causes. Treatment was started with non-steroidal anti-inflammatory drugs, which improved the

arthralgia. The patient is currently asymptomatic, with out-patient follow-up.

Primary HOA, also known as pachydermoperiostosis, was initially described by Friedreich in 1868. It is an autosomal dominant disease, which predominantly affects males (9:1), manifesting itself in a presentation peak during the first year of life and with another one during adolescence.<sup>3</sup> Clinically, it is translated into clubbing with “watch glass” nails, increase in limb volume, arthralgia, myalgia, coarse facial features, with wrinkled and oily skin in the frontal area, as well as cutis verticis gyrata (excess growth of the skin on the scalp, with furrows similar to cortical brain twirls).<sup>2,4</sup>

The characteristic radiological findings are cortical thickening and long bone periostitis, which is bilateral and symmetric. Periosteal reaction is irregular and often involves epiphysis; there is preservation of joint spaces and absence of erosions or periarticular osteopenias.<sup>1</sup> It is necessary to carefully reject a series of pulmonary and extrapulmonary conditions to be able to differentiate from secondary HOA. The pulmonary causes include bronchiectasis, pulmonary fibrosis, cystic fibrosis, sarcoidosis, primary or metastatic (the most common) neoplasm, mesothelioma and infectious diseases such as tuberculosis and chronic empyemas.<sup>4,5</sup> The extrapulmonary causes consist of some neoplasms (Hodgkin's disease, carcinoma of the oesophagus), as well as cyanotic congenital heart disease, infective endocarditis, polycythemia vera, liver cirrhosis, pyelonephritis and ulcerative colitis, among others.<sup>4,5</sup>

The differential diagnosis of primary HOA includes acromegaly, generalised cortical hyperostosis (Van Buchem disease), epiphyseal dysplasia, diaphyseal dysplasia and hypervitaminosis A, among other conditions.<sup>6-8</sup> In acromegaly, the patient's growth is disproportionate and craniofacial deformities occur, together with prognathism.<sup>7</sup> In Van Buchem disease, clubbing or cutaneous changes are not observed and, unlike HOA, periosteal reaction does not involve epiphysis.<sup>6-8</sup> Another pathology used for differential diagnosis is thyroid clubbing, but here exophthalmos, myxoedema (without skin thickening) and other hyperthyroidism findings commonly occur.<sup>7</sup>



**Figure.** A) Clubbing and “watch glass” nails. B) Increase in the tips of the toes and “watch glass” nails. C) Radiograph of the left forearm shows cortical thickening in the radius and ulna, as well as periosteal reaction (arrows).

There is no specific treatment, with the use of NSAIDs being indicated in symptomatic patients; octreotide and pamidronate have been used in refractory cases. In cases of secondary HOA, there can be symptom and manifestation relief after successful treatment of the underlying cause.<sup>3</sup>

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