Primary hypertrophic osteoarthropathy (incomplete form) in young adults: a case report and review of literature

Hemanta K Nayak, Vangipuram Deepak Rajkumar, Naresh Kumar, Premashis Kar

Department of Medicine,
Maulana Azad Medical
College, New Delhi, India

Correspondence to

Dr Hemanta K Nayak,
drhemantanayak@gmail.com

SUMMARY

Hypertrophic osteoarthropathy is characterised by digital clubbing and periosteal reaction of long bones. Most cases are associated with malignancy or other conditions such as congenital heart disease, liver cirrhosis, pulmonary fibrosis, biliary atresia and inflammatory bowel diseases. We report a middle-aged man found to have 15-year history of clubbing of the fingers and toes on his routine check-up for dyspepsia. Skiagram of hand joints showed periosteal apposition without any periosteal reaction of long bones. The search for a secondary cause of clubbing remained negative. The primary or idiopathic form is rare and has a good prognosis and has to be differentiated from secondary form. He was eradicated successfully with Pylori kit for his antral predominant *Helicobacter*-induced gastritis.

BACKGROUND

Hypertrophic osteoarthropathy is a condition characterised by digital clubbing of the hands and feet, extremity enlargement secondary to bone and peri-articular tissue proliferation, joint pain and oedema, bilateral eyelid ptosis, leonine face and thickening of the skin.¹

It is classified as either primary (pachydermoperiostosis) or secondary. The primary or idiopathic form is considered rare, corresponding to 3–5% of all cases.

Here, we describe the clinical and radiological manifestations of a patient with the primary form of hypertrophic osteoarthropathy without any evidence of long bone involvement irrespective of long-standing nature of the disease. In cases of arthralgia/arthritis together with clubbed fingers, consideration must be given to hypertrophic osteoarthropathy. The primary or idiopathic form is rare and has a good prognosis and has to be differentiated from secondary form.

CASE PRESENTATION

A 30-year-old man on his routine health check-up for his dyspepsia was found to have pan-digital clubbing. He had no history suggestive of chronic lung disease or malignancy, rheumatic or congenital heart disease, gastrointestinal disease or hepatic disease. There was no history of a similar condition in his family.

Physical examination showed pan-digital clubbing with watch-glass nails, and diffuse swelling of the hands and feet (figures 1–3). Cardiopulmonary and abdominal examinations were normal.

INVESTIGATIONS

Laboratory reports revealed haemoglobin 120 gm/l (120–150), total leucocyte count 7600/mm³ (4000–11 000), differential leucocyte count polymorphs 74, lymphocytes 24, eosinophil 2, random blood sugar 5.77 mmol/l (normal 4.4–6.1), blood urea 7.1 mmol/l (normal range 2.6–7.5 mmol/l). Serum electrolytes, arterial blood gases and renal and liver function tests were within normal limits.

His chest x-ray, ECG and echocardiogram were normal. His erythrocyte sedimentation rate (ESR) was 50 mm in the first hour. His C reactive protein, rheumatoid factor, antinuclear antibodies and anti-DNA antibodies were non-reactive. On repeating after 1 month the ESR was 30 mm in the first hour.

Skiagram of the patient's hands and wrists showed an increase in soft tissue content, an enlargement of the middle and distal phalanges and associated thickening of the cortex (figure 4). Skiagram of the tibia and fibula were grossly normal with normal periosteum.

Secondary causes of hypertrophic osteoarthropathy were excluded in view of a normal chest skiagram, normal echocardiogram, normal contrast echocardiography and normal arterial blood gas analysis reports.

He also underwent oesophogastroduodenoscopy for his dyspepsia and was found to have *Helicobacter pylori*-related antral predominant gastritis that was successfully eradicated with anti-pylori kit.

DIFFERENTIAL DIAGNOSIS

- ▶ Pandigital clubbing due to cardiac cause
- ▶ Primary hypertrophic osteoarthropathy

TREATMENT

- ▶ Conservative
- ▶ Anti-*H pylori* kit

Figure 1 Photograph showing digital clubbing of hand.

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Figure 2 Photograph showing digital clubbing of hand.

OUTCOME AND FOLLOW-UP

The patient received an *H pylori* kit for antral predominant gastritis and successfully eradicated *H pylori* infection. He followed up regularly for his digital clubbing and was found to have been completely asymptomatic for his clubbing at 1-year follow-up.

DISCUSSION

Primary hypertrophic osteoarthropathy (PHOA) may present with varied clinical manifestations. The most common are clubbing of digits, periostitis and skin thickening. It must be differentiated from the secondary form, triggered by lung, heart or neoplastic diseases.¹

The disease is classified as either primary (hereditary or idiopathic) or secondary. The idiopathic form (also known as pachydermoperiostosis, primary hypertrophic osteoarthropathy or Touraine-Soulente-Golé syndrome) is a rare disease with unknown aetiology, and represents 3–5% of all cases of hypertrophic osteoarthropathy. The diagnosis of pachydermoperiostosis is based on the presence of at least two of the four criteria set by Borochowitz—a history of familial transmission; pachydermia; digital clubbing and skeletal manifestations such as pain or signs of radiographic periostitis. Our patient has the incomplete form of PHOA, with hyperostosis and finger clubbing and no pachydermia.²

Most patients with idiopathic hypertrophic osteoarthropathy have normal development until adolescence, when skin thickening and joint deformities began to occur. These changes progress for many years, then usually stabilise. In the case reported here, there is no evidence of any skin thickening and bony deformity, although clubbing was present for the past 15 years.

Figure 3 Photograph showing digital clubbing of foot.

Figure 4 Skiagram of the patient's hands and wrists showed an increase in soft tissue content, an enlargement of the middle and distal phalanges and associated thickening of the cortex.

The disease affects males predominantly (sex ratio 9:1) and is considered to be familial (25–40% of cases). No family member of this patient has similar disease. The combination of thickened skin and bony enlargement can result in great thickening of the extremities, which is the most striking physical finding. Seborrhoea, hyperhidrosis, acne and folliculitis are part of complete form of PHOA but notably absent in this case.¹

The pathogenesis of the Touraine-Soulente-Golé syndrome has not been fully elucidated. A brief study on patients with the disease showed abnormal peripheral blood supply when compared with non-affected individuals; however, additional studies failed to reproduce these findings. Biopsies of skin and bone marrow show an exacerbated proliferation of fibroblasts, associated with diffuse epidermal hyperplasia, partial occlusion of vascular lumen and lymphohistiocytic infiltration with collagen deposition. An isolated study showed normal fibroblastic proliferation, but with a decrease in total protein rate and collagen synthesis, with significant increase of proteoglycans and sulphated glycosaminoglycan.³

The clinical condition typically begins in adolescence, evolving chronically and insidiously, and alternating exacerbation with asymptomatic periods. Usually, the disease evolves for about 10 years to stabilise spontaneously. Although the evolution of the idiopathic form is less satisfactory (unlike the patient described in the case), it usually does not interfere with the patient's life expectancy.⁴

The bony changes consist of symmetric, irregular periosteal hypertrophy with new bone formation. These changes are most severe in the extremities and can involve any bone, although the skull and the vertebral column are rarely affected. Radiographs reveal diffuse periostitis along the length of bones, including epiphyses, in 80–97% of cases, and often with irregular contours. The importance of periosteal apposition increases with disease duration. Acro-osteolysis has also been reported in 78% of cases. However, there were no clinical symptoms of inflammatory arthropathy and laboratory markers of inflammation were negative.⁴

Cutaneous alterations in PHOA include seborrhoea, blepharoptosis, acne, palmar and plantar hyperhidrosis, eczema, erythematous lesions in joints and burning sensation in hands and feet. The cosmetic and functional deformities can improve with simple surgical procedures, such as resection of stratum skin and adjacent tissue, followed by primary healing.³ However, in this case no skin involvement was observed.

In conclusion, the identification of secondary osteoarthropathy is relatively common among clinicians. However, the primary or idiopathic form of hypertrophic osteoarthropathy is

uncommon and again incomplete form is extremely rare in long-standing primary hypertrophic osteoarthropathy cases. In cases of arthralgia/arthritis together with clubbed fingers, consideration must be given to hypertrophic osteoarthropathy. The primary or idiopathic form is rare and has a good prognosis.

Learning points

- ▶ In cases of arthralgia/arthritis together with clubbed fingers, consideration must be given to hypertrophic osteoarthropathy.
- ▶ The primary or idiopathic form of hypertrophic osteoarthropathy is uncommon and again incomplete form is extreme rare in long-standing primary hypertrophic osteoarthropathy cases.
- ▶ The primary or idiopathic form is rare and has a good prognosis.

Competing interests None.

Patient consent Obtained.

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