

# Touraine-Solente-Gole syndrome

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## DESCRIPTION

A 26-year-old man presented to our clinic with gradually progressive painful swelling of wrists, knees and ankles for the last 10 years. Since the last 2 years, he had also noticed an increase in thickness of skin over his forehead and an increase in sweating over his palms and feet. There was no history of consanguinity or similar illness in the family. On examination, he had coarse facial features with thick skin and increased grooves on the face, most notably on the forehead (**figure 1A**). Clubbing was noted in all fingers and toes, along with significant swelling and tenderness in bilateral wrists, knees and ankles (**figure 1B**). The plain radiographs of the wrists and knees showed significant metaphyseal and diaphyseal periosteal reactions, along with periostitis (**figure 2A,B**). The erythrocyte sedimentation rate (60 mm/hour) and the C reactive protein (36 mg/dL) were elevated. All other routine blood investigations were normal. Rheumatoid factor and antinuclear antibody were also negative. Chest X-ray and ultrasonographic study of the abdomen were normal. A synovial

biopsy from the knee joint showed chronic non-specific inflammation. A diagnosis of pachydermoperiostosis was suspected. A bone scan was done, which showed mildly increased flow and pool activity with increased tracer uptake in delayed phase in metaphyses of the bilateral femur, tibia and radius. To rule out associated gastric polyposis, an upper gastrointestinal (GI) endoscopy was done, which was normal. The patient was counselled regarding the condition and was put on non-steroidal anti-inflammatory drugs for symptomatic relief.

Touraine-Solente-Gole syndrome or pachydermoperiostosis is an extremely rare disorder characterised by a thickening of the skin (pachyderma), clubbing and hyperhidrosis.<sup>1 2</sup> It has three forms: complete (clubbing, periostosis, pachyderma and cutis verticis gyrata), incomplete (without cutis verticis gyrata) and forme fruste (pachyderma with minimal skeletal changes). Fulfilment of the diagnostic criteria requires at least two of the following: hypertrophic skin changes, clubbing, bone pain, radiographical changes and family history.<sup>3</sup> The most common genes associated with the disease are Hydroxyprostaglandin Dehydrogenase (HPGD) and SLCO2A.<sup>4</sup> Important differentials include secondary hypertrophic osteoarthropathy, acromegaly, thyroid acropachy and chronic inflammatory rheumatic diseases. Other rarer differential diagnoses are chronic recurrent multifocal osteomyelitis; synovitis, acne, pustulosis, hyperostosis and osteitis syndrome; Camurati-Engelman disease; and syphilitic periostosis. Secondary causes of hypertrophic osteoarthropathy (lung or GI malignancies, cirrhosis of the liver and infective endocarditis) should be excluded, especially where dermatological signs are not prominent. The management of this disease is mostly symptomatic, with anti-inflammatory and antiresorptive therapy. Response to treatment should be regularly assessed on follow-up as the disease may lead to debilitating complications where surgical



**Figure 1** (A) Coarse facial features with thick skin and increased grooves on the forehead. (B) Clubbing of fingers and toes with bilateral symmetrical swelling of the wrists, knees and ankles.



**Figure 2** (A) Plain radiograph showing significant bilateral periosteal reaction with thickening along the radius. (B) Plain radiograph showing significant bilateral periosteal reaction with thickening along the tibia and patella. R: Right.

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## Learning points

- ▶ Pachydermoperiostosis should be considered in patients with coarse thickened skin, clubbing and bone pain.
- ▶ Adequate symptomatic relief and prognostication are very important in such cases.
- ▶ Close monitoring of the patient's condition is advisable.

intervention may be required. Also, genetic counselling should be offered to patients and their families.<sup>5</sup>

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