Rare disease

Touraine–Solente–Gole' syndrome

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Summary

Touraine–Solente–Gole' syndrome, also known as pachydermoperiostosis or primary hypertrophic osteoarthropathy is a rare familial disorder generally seen in males. Although it presents with characteristic morphological and radiological features, this is an uncommon diagnosis and is entertained only once other causes of secondary hypertrophic osteoarthropathy are carefully excluded.

BACKGROUND

Pachydermoperiostosis accounts for only 3–5% cases of hypertrophic osteoarthropathy.¹ Patients with pachydermoperiostosis may present to multiple clinical specialities like orthopaedics, rheumatology, endocrinology and dermatology. The general physician is very occasionally confronted with such a case in a lifetime. Such cases are intriguing, challenging to diagnose and difficult to treat.

CASE PRESENTATION

An 18-year-old male presented to us with moderate intensity pain and swelling of both wrists and ankles for last 6 months. He also had gradual enlargement of the fingers and toes, furrowing of skin over the forehead, facial acne and excessive sweating of the palms and soles. He also reported low-grade fever but there was no history of skin rash, sore throat or urinary complaints. He had no headache or visual disturbances. He did not narrate any history pertaining to pulmonary, cardiovascular or gastrointestinal disease. No other sibling or family members were afflicted with similar complaints.

Clinical examination revealed coarsening of facial features, shiny upper eyelids, deepening of forehead lines (figure 1), palmar line thickening with ripped appearance (figure 2), spade-like enlargement of hands (figure 3),

swollen ankle joints (figures 4 and 5) and palmo-plantar hyperhidrosis. There was no evidence of digital clubbing. Systemic examination was unremarkable.

INVESTIGATIONS

Patient's haematological investigation revealed haemoglobin of 10.9 g/dl, total leucocyte count 13 700/mm³, platelets 4.2 lac/mm³, mean corpuscular volume 72 fl, mild anisopoikilocytosis and mild hypochromia. His biochemical parameters showed normal values of serum sodium 136 mEq/l, serum potassium 4.7 mEq/l, blood urea 15 mg/dl, serum creatinine 0.8 mg/dl, serum uric acid 4.8 mg/dl, serum calcium 7.8 mg/dl, serum phosphate 3.4 mg/dl, serum bilirubin 0.8 mg/dl, aspartate aminotransferase 28 IU/l, alanine aminotransferase 17 IU/l, total serum proteins 7.6 g/dl, serum albumin 3.9 g/dl.

Hormonal assays were carried out to rule out underlying thyroid, pituitary disease or abnormal bone metabolism. The growth hormone level was 10.2 ng/ml (normal range 0.5–55 ng/ml), thyroid stimulating hormone 3.42 uIU/ml (normal range 0.35–5.5 uIU/ml), T3(F) 3.46 pg/ml (normal range 2.3–4.2 pg/ml), T4(F) 1.11 ng/ml (normal range 0.89–1.76 ng/ml), serum prolactin 7.01 ng/ml (normal range 0–20 ng/ml) and luteinising hormone 2.3 mIU/ml (normal range 1.5–9.3 mIU/ml). 25-hydroxy D3 level was 47.29 nmol/l (normal range 47.70–144 nmol/l)

Figure 1 Clinical photograph showing deep furrows on forehead.

Figure 2 Clinical photograph showing coarsening of palmar creases.

Figure 3 Clinical photograph showing large spade-like hands with broadened wrists.

Figure 4 Clinical photograph showing enlargement of the distal part of the lower extremities.

and parathyroid hormone was 45.9 pg/ml (normal range 8–51 pg/ml).

Patient was further investigated and antistreptolysin O titre was less than 200 IU/ml. Rheumatoid antibody and antinuclear antibody were negative; however, the C reactive protein was 26.9 mg/l (normal range 0.2–3 mg/l). Venereal Disease Research Laboratory testing was also non-contributory. Skull x-ray, chest radiograph, abdominal ultrasonograph and echocardiography did not reveal any abnormality. Radiographs of the long bones revealed subperiosteal new bone formation characterised by irregular outline of the bony surfaces (figures 6–8). Bone scan revealed active inflammation with increased osteoblastic activity in the knee joint and increased osteoblastic activity in the appendicular skeleton. On the basis of the clinical features and the radiological findings, a diagnosis of pachydermoperiostosis was made.

TREATMENT

The patient was started on indomethacin 75 mg twice a day after food for amelioration of the pain. Oral risenedronate

Figure 5 Clinical photograph showing ankle joint arthritis.

Figure 6 Radiograph of the leg (tibia and fibula) depicting irregular bony surfaces.

35 mg weekly was added to the regimen as his pain did not remit on analgesics alone. However, patient's pain did not improve on the above regime. So, he returned after a period of 6 months. This time patient was given a trial of zoledronic acid 5 mg by intravenous route.

OUTCOME AND FOLLOW-UP

He is being followed up regularly.

DISCUSSION

Pachydermoperiostosis or primary hypertrophic osteoarthropathy was first described in 1868 by Friedrich in two male siblings.² Touraine–Solente–Gole' syndrome refers to the characteristic constellation of symptoms and signs described in detail by Touraine *et al* in 1935.³ Since then many isolated reports of similar cases have been published, but the disease is largely uncommon and thus fascinating to identify. Its pathogenesis is unknown but inflammatory factors and increased bone remodelling have been implicated.

Figure 7 Radiograph of the foot depicting subperiosteal new bone formation.

Pachydermoperiostosis generally presents in peripubertal young adults, with a predilection for males. This condition is often idiopathic and inherited in an autosomal dominant fashion⁴ or as X-linked trait.⁵ However, in various other case reports, family history could not be identified.⁶ It manifests as gradual thickening and enlargement of the extremities, arthritis and arthralgias and clubbing of digits. In addition, there is coarsening of facial features, seborrhoea, facial acne and hyperhidrosis. In extreme cases of pachydermia, cutis verticis gyrata (undulating grooved and thickened scalp) or leonine facies may become apparent.

The diagnosis is clinched with the assistance of radiological investigations revealing subperiosteal bone formation in the long bones and ossification of interosseous membranes or ligaments. Since the morphological features overlap with secondary hypertrophic pulmonary osteoarthropathy, the latter must be excluded by thorough examination.

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 use of non-steroidal anti-inflammatory drugs for pain relief. Bisphosphonates like pamidronate or risedronate and have been used extensively in pachydermoperiostosis due to their antiresorptive and osteoclast inhibitory properties.^{7,8}

Figure 8 Radiograph of the forearm (radius and ulna) depicting irregular outline.

Learning points

- ▶ Patients of pachydermoperiostosis have features similar those of pulmonary hypertrophic osteoarthropathy, acromegaly or thyroid acropachy.
- ▶ Characteristic morphological and radiological features facilitate the diagnosis.
- ▶ Disease is self limited but may result in physical disability.

Competing interests None.

Patient consent Obtained.

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