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CASE REPORT

Touraine–Solente-Gole Syndrome: A Case Report

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Abstract:

Touraine–Solente-Gole Syndrome (TSGS) or pachydermoperiostosis is a rare disorder characterized by pachydermia, periostosis and digital clubbing. It is a clinical variant of primary hypertrophic osteoarthropathy, for which the etiopathogenesis is not fully known. It is seen more often in males and I have not found a female case in the literature. The case presented here is of a female patient who presented with digital clubbing in the hands and feet, periostosis, cutis verticis gyrate, and arthralgia.

Keywords: Touraine Solente Gole syndrome, Pachydermoperiostosis, Hypertrophic osteoarthropathy.

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Introduction

Touraine–Solente-Gole Syndrome (TSGS) is a rare disorder characterized by pachydermia, periostosis and digital clubbing [1]. The male-female ratio has been reported to be 9:1 [2], and estimated prevalence is approximately 0.16% [3]. TSGS may be idiopathic or inherited. Although autosomal dominant inheritance with incomplete penetration and variable expression has been confirmed, both autosomal recessive and X-linked inheritance have been suggested [1]. It was initially described by Friedreich [4] in1868 and then by Touraine, Solente and Gole [5] in 1935, who recognized its familial nature. Although the etiopathogenesis is not fully known, the 15-hydroxyprostaglandin dehydrogenase gene and the solute carrier organic anion transporter family member 2A1 have been found to be associated with TSGS [6,7]. It is more common in men, and I have not found a female case in the literature. The case presented here is a female patient who presented with arthralgia and was diagnosed with TSGS.

Case

A 51 year old female presented at the polyclinic with complaints of pain in the hands and feet. The complaints had started approximately 30 years ago and joint pains had increased in the last 4-5 years. There was nothing remarkable in the patient's own history or family history. There was nothing determined in the rheumatological investigation. There was no psoriasis. In the physical examination of the patient, deep folds known as cutis verticis gyrata were observed in the scalp (Figure 1).

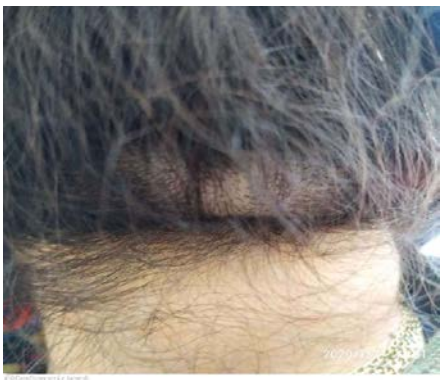

Figure 1. Cutis verticis gyrata

There was mild pachydermia on the forehead. Widening and clubbing was present in the distal of the fingers and toes (Figure 2,3). Joint range of motion was full. There was sensitivity in the joints of the hands and feet but there was no arthritis.

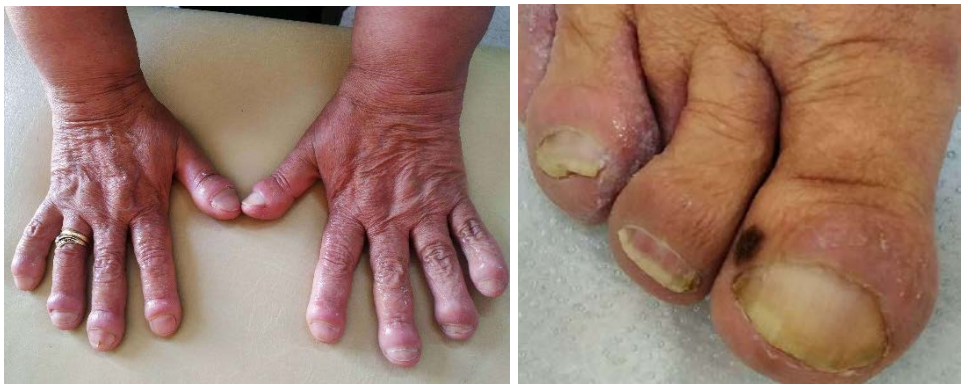

Figure 2,3. Clubbing was present in the distal of the fingers and toes

The blood work up including hemoglobin, hematocrit, red blood cell, white blood cell and blood biochemistry (glucose, alanine aminotransferase, aspartate aminotransferase, alkaline phosphatase, gamma-glutamyl transferase, serum creatinine, urea) results were in the normal ranges. Erythrocyte sedimentation rate (ESR): 25 mm/h and C-reactive protein (CRP): 3 mg/L(normal <5 mg/L). P-ANCA, C-ANCA, ANA, RF, anti-CCP, cryoglobulins were all negative. Serum iron, ferritin, TSH, T4 (free), growth hormone, immunoglobulins (IgM, IgG, IgA), and complements (C3, C4) were normal. Serology results were negative for viral hepatitis (VDRL, TPHA, HBsAg, anti-HIV, anti-hepatitis C virus), TORCH, antibodies against streptococcus, Epstein-Barr virus and the Write test for brucellosis. Urinalysis revealed no changes.

Abdominal ultrasound, electrocardiogram and chest X-ray examinations revealed no pathology. The plain radiographs of the wrists and feet showed significant metaphyseal and diaphyseal periosteal reactions. Increased cortical thickening was seen in the hand bones and in the tibia (Figure 4,5).

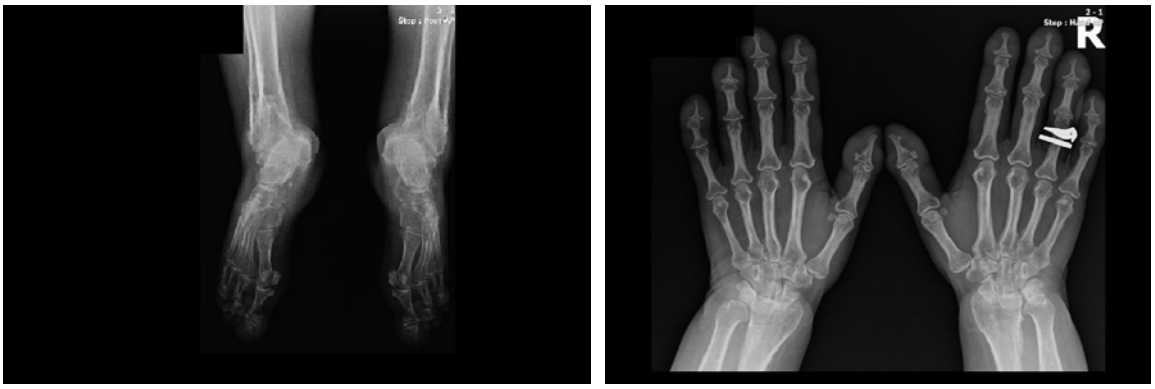

Figure 4,5. Radiographs of the wrists and feet showed periosteal reactions and increased cortical thickening.

A diagnosis of TSGS was made, and treatment was started of non-steroidal anti-inflammatory drugs (NSAID) and colchicine. The pain decreased significantly during follow-up. Written consent was obtained from the patient

Discussion

TSGS is an infrequently seen disease. A previous study over a 17-year period showed TSGS at the rate of 0.03% in rheumatismal diseases in patients presenting at a university hospital [8]. It includes 3-5% of all patients with hypertrophic osteoarthritis, it affects males more than females and phenotypes are more severe in males [9]. There are family clusters in 25%-38% of cases [3]. No differences have been reported between ethnicities [10]. Although the pathogenesis is not fully known, there may be a mutation in the gene encoding the 15-hydroxyprostaglandin dehydrogenase enzyme in TSGS patients, and this leads to an increase in prostaglandin E2 (PGE2) in the blood. By mimicking osteoblast and osteoclast activity, PGE2 can lead to acro-osteolysis and periosteal bone formation [11].

Clinically, the disease is defined in 3 different forms [5].

1. Complete; pachydermia, clubbing and periostitis together
2. Incomplete; skeletal changes are present but no pachydermia
3. Fruste; pachydermia is strongly evident, and mild skeletal changes are seen

The diagnostic criteria of TSGS are classified as major (pachydermia, periostosis, digital clubbing), or minor (hyperhidrosis, cutis verticis gyrate, gastric ulcer, blepharoptosis, arthralgia, joint effusion, column-like legs, edema, seborrhea, acne, hyperhidrosis, flushing) [5].

In the current case, there was clubbing in the fingers and toes, periostosis, cutis verticis gyrate, arthralgia and mild pachydermia. When pachydermia is severe, there may be a lion face appearance [10]. In this patient, the pachydermia was significant on the scalp but mild on the forehead. There is no specific laboratory test for TSGS [10].

Bulbous deformities at the distal fingers, abnormal nail curvature, and soft-tissue swelling can be present on radiographs. Periostosis seen on imaging is characteristic of hypertrophic osteoarthropathy. The shafts of tubular bones are especially affected. Involvement of the epiphysis is more frequent in primary hypertrophic osteoarthropathy [12]. The histopathological findings of pachydermia include dermal edema, mucin deposition, elastic fiber degeneration, dermal fibrosis and adnexal hyperplasia [13].

Secondary hypertrophic osteoarthropathy, thyroid acropachy, acromegaly and syphilitic periostitis are included in the differential diagnosis. There is no specific treatment. NSAIDs, steroids,

pamidronate, risedronate, colchicine, and tamoxifen can be used in treatment [14]. Infliximab has been reported to be of partial benefit in resistant bone pain and arthritis [15]. Surgical interventions are rarely applied [10].

In conclusion, although TSGS is a benign disease, differentiation from secondary causes such as malignancies is important.

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