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Case Study

Touraine–Solente–Golé Syndrome (Primary Hypertrophic Osteoarthropathy) Presenting With Chronic Kidney Disease: A Rare Case Report

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ABSTRACT

Touraine–Solente–Golé syndrome (TSGS), also known as primary hypertrophic osteoarthropathy, is a rare genetic condition that is characterized by pachydermia, digital clubbing, and periostosis. We present a case of a 31-year-old male patient with a diagnosis of TSGS and hypertension who came to us with complaints of bilateral ankle swelling, foamy urine, and worsening kidney function. He was diagnosed with chronic kidney disease stage 3 with a suspected intrinsic renal pathology, for which a CT-guided renal biopsy was planned. The patient was treated by giving supportive care, blood pressure optimization, and monitoring of his renal function closely. We conclude that this case demonstrates the significance of identifying kidney manifestations in patients suffering from TSGS and emphasizes the need for kidney evaluation when proteinuria and worsening kidney dysfunction are noted.

INTRODUCTION

Touraine-Solente-Golé syndrome (TSGS) or primary hypertrophic osteoarthropathy (PHO) or pachydermoperiostosis is a rare genetic disorder characterized by three features: digital clubbing, pachyderma (skin thickening), and periostosis (formation of new bone). This disorder makes up 3-5% of all hypertrophic osteoarthropathy cases, with the rest stemming from the presence of other diseases concerning cardiovascular, gastrointestinal, liver, or malignancies. This syndrome was first cited by Touraine, Solente, and Golé in 1935 and tends to be passed through either autosomal dominant or recessive ways with different degrees of penetrance. [**Error! Reference source not found.**,**Error! Reference source not found.**]

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TSGS generally manifests itself in the teenage or early adult years and has a greatly higher prevalence in men than women. The mechanism behind this syndrome arises from mutations in the genes HPGD or SLCO2A1, which results in poor degradation or transport of prostaglandin E_2 (PGE_2). The levels of PGE_2 are high, which leads to the overgrowth of periosteal bone, connective tissue, and blood vessels, and thus the appearance of these clinical manifestations. Most patients report gradual enlargement of their hands and feet along with digital clubbing, altered mimicry of the face, hyperhidrosis, pains in the joints, and skin thickening. [**Error! Reference source not found.**]

Touraine–Solente–Golé syndrome is often missed or misdiagnosed due to its rarity and performance that overlaps with ailments such as acromegaly, thyroid acropachy, and secondary hypertrophic osteoarthropathy. [**Error! Reference source not found.**] The diagnosis is based on specific clinical signs, imaging studies that show periostitis, ruling out other possible conditions, and genetic testing if possible. Treatment is aimed at relieving symptoms only and includes using NSAIDs, rehabilitation, and other treatment that is not surgical or aesthetic, which will be used only in specific cases.

CASEPRESENTATION

The 31-year-old man who had a history of Touraine-Solente-Golé syndrome which is commonly known as primary hypertrophic osteoarthropathy or pachydermoperiostosis, as well as eight years of hypertension, presented with bilateral edema of the ankles for the last six months. He had initially gone to the doctor and was diagnosed with a condition known as acute kidney injury (AKI) where he went unnoticed for about three consecutive months until he started showing signs of frothy urine indicating a possible excessive amount of proteins in the urine and generalized skin rash. The patient stated that he had no previous pain history nor any sudden problem in the lower part of his abdomen during this period.

The patient was considered as a person suffering from primary hypertrophic osteoarthritis (pachydermoperiostosis). He was also known to have chronic kidney disease stage 3 along with kidney function that has been getting worse with time. Hence, at this point, his doctors discussed that he requires further kidney evaluation with the intention of conducting kidney biopsy. Upon examining the patient, we found him to be conscious, alert, and stable in terms of his cardiovascular health. A thorough examination uncovered signs related to pachydermoperiostosis, such as thickening of the skin. Therefore, a review of his neurological state revealed that all higher brain processes as well as cranial nerves functions were intact, while movement and balance were normal. In addition to prior ailments of the patient, tests that indicated skin conditions were positively related to pachydermoperiostosis.

Our tentative diagnosis states that the patient has developed pachydermoperiostosis along with chronic kidney disease stage 3. It was decided to conduct a CT-guided kidney biopsy. This procedure involves certain risks but is useful in confirming the diagnosis. However, the prognosis is expected to be poor, so the patient was warned about various complications. The patient was subject to additional nephrologic assessment and provided with appropriate care, blood pressure regulation, and regular monitoring of kidney function pending diagnosis from renal biopsy results. The future management strategy was determined on the basis of biopsy results and worsening of underlying chronic renal failure.

Table 1: Urine Examination

Investigation	Observed value	Reference range	Interpretation
Urine protein	+++	Negative	Significant proteinuria
Blood	+	Negative	Positive
RBCs	5–6/HPF	0–5/HPF	Mildly increased
Granular casts	+	Absent	Abnormal
Hyaline casts	+	Absent	Abnormal

Table 2: Renal Function Tests

Investigation	Observed value	Reference range	Interpretation
Serum creatinine	1.50 mg/dL	0.66–1.25 mg/dL	Elevated
eGFR	61.05 mL/min/1.73 m²	>90	Reduced
BUN	16.0 mg/dL	9.0–20.0 mg/dL	Normal
Urea	34.2 mg/dL	19.0–43.0 mg/dL	Normal

Table 3: Viral Serology

Investigation	Observed value	Reference range	Interpretation
HIV 1 & 2 Ab + p24 Ag	0.29 COI	<0.90	Non-reactive
HBsAg	0.48 COI	<0.90	Non-reactive
Anti-HCV antibody	0.08 COI	<0.90	Non-reactive

Table 4: Renal Doppler Findings

Parameter	Finding
Right kidney	10.2 × 4.1 cm
Left kidney	11.2 × 4.2 cm
Cortical echogenicity	Bilaterally increased
Corticomedullary differentiation	Loss of corticomedullary differentiation
Hydronephrosis	Absent
Renal calculi/mass	Not detected
Renal artery stenosis	No significant stenosis
Intrarenal resistive index (RI)	Approximately 0.70 bilaterally
Impression	Bilateral medical renal parenchymal disease with mildly elevated intrarenal resistive indices.

DISCUSSION

Touraine-Solente-Gole syndrome (TSGS), also called pachydermoperiostosis (PDP) or primary hypertrophic osteoarthropathy (PHO), is a rare genetic condition with marked clinical characteristics, which are histologically described as the classical triad of pachydermia, clubbing of fingers, and periostosis. Males are more frequently affected by this condition, which has its onset mostly during adolescence or early adulthood. The mutations of HPGD and SLCO2A1 genes, which take part in the metabolism of prostaglandin E2, are known to be involved in the etiology of the disease. [**Error! Reference source not found.**]

This case is striking in that the patient is a 31-year-old man with a documented diagnosis of

pachydermoperiostosis/TSGS who presented with bilateral ankle swelling and renal impairment. Patient had hypertension history and was diagnosed with acute kidney injury in the past. He then developed frothy urine and urinalysis revealed +++ proteinurea, microhematuria, and granular and hyaline casts. Blood creatinine level was 1.50 mg/dL and GFR was 61.05 mL/min/1.73m² which indicated that the renal function is impaired. The Doppler examination of kidneys revealed increased cortical echogenicity and loss of corticomedullary differentiation with mildly increased resistive indices which indicates presence of renal parenchymal disease. It should be noted that there were no indications of renal artery stenosis. The involvement of the kidneys is not typically seen as an important feature of TSGS. It does, however, make this patient's kidney condition unique. Nonetheless, some cases of kidney disease have been reported for patients who have pachydermoperiostosis. Cases like that of a patient with amyloidosis of the kidneys or chronic disease of the kidneys have been noted. In the case of one particular patient, the kidneys failed and were caused by some kind of injury due to pain relief so treatment of that disorder may not be due to TSGS. [**Error! Reference source not found.**,**Error! Reference source not found.**]

The existence of considerable proteinuria and kidney damage resulted in the decision to carry out CT-guided renal biopsy. The significance of histopathological assessment would be of utmost importance in identifying the renal pathology, either primary renal disease, hypertensive nephrosclerosis, drug-related tubulointerstitial damage, amyloidosis, or any other glomerular disease. Due to the fact that no biopsy had been performed and histopathological evidence had not been provided, it is impossible to identify the renal pathology in the particular case. The case signifies an important clinical aspect: patients suffering from some unusual systemic conditions like TSGS must undergo nephrological assessment whenever the symptoms of these conditions appear, such as proteinuria, edema, or deterioration of renal function. [**Error! Reference source not found.**]

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CONCLUSION

The condition known as Touraine-Solente-Gole syndrom, known as pachydermoperiostosis, occurs as a rare hereditary variant of primary hypertrophic osteoarthropathy characterized by having pachydermia, digital clubbing, and periostitis. The case reports the unusual manifestations of TSGS presenting with both bilateral ankle swelling, proteinuria, and renal failure classified as CKD stage 3. The occurrence of renal complication is not a typical feature of TSGS; hence, proteinuria and renal impairment requires careful assessment for an independent kidney disease. The approach in this patient using renal Doppler revealed the presence of medical renal parenchymal disease and a lack of serious

renal arterial stenosis. Renal biopsy was scheduled to investigate the etiology of nephropathy. The case shows the importance of being attentive to the diagnosis of TSGS and always maintaining a high level of alertness regarding the potential existence of systemic complications.

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