Case Report

Turner's syndrome presenting as metabolic bone disease

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ABSTRACT

Turner's syndrome is a genetic disorder with a complete or partial absence of one X chromosome with characteristic phenotypic features. The prevalence of renal anomalies in turner syndrome is 30–40%. However, the renal function is usually normal. We report a case of Turner's syndrome presenting with chronic kidney disease and renal osteodystrophy.

Key words: Renal osteodystrophy, secondary hyperparathyroidism, Turner's syndrome

Introduction

Turner's syndrome is the most common female sex chromosomal disorder with an estimated prevalence of 1 in 2500.[1] Renal anomalies are seen in 30–40% of these patients and screening for the same is currently recommended for all patients.[2] Although various renal anomalies ranging from horseshoe kidney to multicystic kidney disease have been described, the renal function is usually normal. The renal malformations increase the risk of recurrent urinary tract infections. Decreased bone mineral density has been documented in Turner's syndrome and can occur independent of estrogen deficiency.[3]

Case Report

A 16-year-old girl was admitted with progressive thinning and weakness of both upper and lower limbs with severe myalgia for 1 year duration. She had become bed bound in the last 3 months. She also had poor appetite and nausea. She was unable to flex or extend the neck. She also had primary amenorrhea. She was the second born girl of second-degree consanguineous marriage and her only sibling had died due to undiagnosed illness.

On examination, her height was 131 cm (<3rd centile), weight was only 23 kg (< 3rd centile), and had webbing of neck. She also had severe restriction of neck flexion and hyperconvex nails. There were features suggestive of rachitic rosary [Figure 1a], anterior bowing of tibia [Figure 1b], and proximal myopathy. The LH was 157.3 mIU/ml and FSH was 138.9 mIU/ml. Her karyotype showed 45 XO pattern, confirming Turner's syndrome. Her biochemical features showed hypocalcemia (6.5 mg/ dl), normal phosphorus (3.6 mg/dl), and raised alkaline phosphatase level (1813 IU/l). Serum parathormone was greatly elevated (1652.5 pg/ml, normal 14–74 pg/ml) and 25-hydroxy vitamin D was decreased. (17.6 ng/ ml, normal >30 ng/ ml). She also had uremia (serum urea: 124 mg/dl and serum creatinine: 3.8 mg/dl) and her ultrasound showed bilateral shrunken kidneys.

Her arterial blood gases revealed an uncompensated metabolic acidosis with pH of 7.19, HCO_3 of 11.4 mmol/l and pCO_2 of 31 mmHg. The anion gap was elevated (−19.6). X-ray of spine showed non-union of the ring apophyses [Figure 2] with the body of the vertebra and those of long bones and skull revealed features of osteosclerosis and metaphyseal widening [Figure 1c]. Tc99m bone scan showed metabolic bone disease with no renal uptake, i.e. metabolic superscan [Figure 3]. Her parathyroid MIBI

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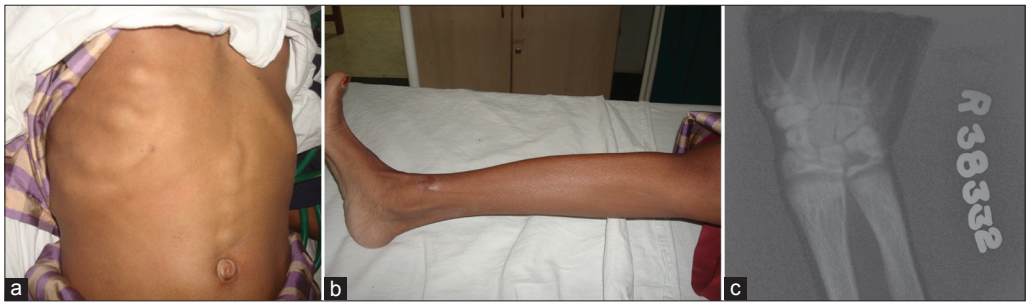

Figure 1: (a) Figure showing rickety rosary. (b) Figure showing anterior bowing of tibia. (c) Radiograph of right hand with wrist of a 16-year-old female patient with secondary hyperparathyroidism and renal osteodystrophy shows widening of the growth plate with indistinctness of metaphyseal margins and frayed appearance at the lower end of radius ulna and proximal metacarpals. Also seen is intracortical bone resorption with coarsened trabeculae.

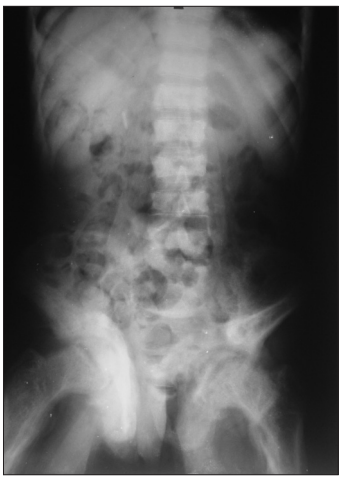

Figure 2: Plain radiograph AP view of pelvis of a 16-year-old female with renal osteodystrophy with lumbar spine shows deformed pelvis with bilateral protrusio acetabulae and triradiate configuration. There is increased density of bones with ground glass appearance, extensive cortical thickening of pelvic bones, and proximal femur more so involving the ileopectinate line with coarsened trabeculae. The physis plates of proximal femur are wide and irregular with deformed femoral epiphysis thus showing feature of both rickets and osteosclerosis in this patient.

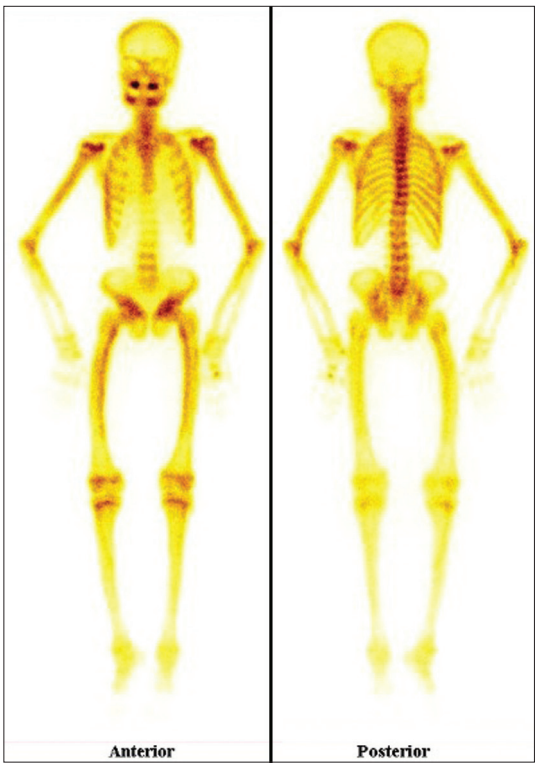

Figure 3: Tc99 Bone scan image of the patient showing avid tracer uptake by the entire skeleton with negligible renal uptake (metabolic superscan).

scan did not show any uptake by parathyroid glands. Echocardiography and audiometry were normal. Her thyroid function was normal.

She was diagnosed to have Turner's syndrome with renal osteodystrophy. Her chronic kidney disease was deemed to be due to chronic glomerulonephritis and was managed conservatively. She was given 3,00,000 IU bolus injection of cholecalciferol and followed with supplementation of calcium 1500 mg/day and calcitriol 0.75 mcg/day. With treatment, her calcium improved to 7.5 mg/dl. She also had significant reduction in bone pain and PTH dropped to 627 pg/ml.

Discussion

Turner's syndrome is a chromosomal disorder which is usually recognized due to characteristic phenotypical features in preadolescence or due to delayed puberty/primary amenorrhea later on. The presence of renal anomalies in Turner's syndrome is well known but cases of frank renal failure and its complications have not been reported so far. The incidence of urinary tract infections is increased because of obstruction. Di Pinto *et al* studied the renal malformations in 24 patients with Turner's syndrome in which none had chronic kidney disease.[4] In a larger series, Lippe *et al.* studied 141 patients of which 33% had renal anomalies but none had bilateral contracted kidneys.[5] The presence of hyperparathyroidism in Turner's syndrome has been reported in only five cases in the literature and all had primary hyperparathyroidism unlike secondary hyperparathyroidism in our case.[6] Her low calcium and high parathormone explain the significant contribution of coexistent vitamin D deficiency to her condition. The rise in

calcium levels after calcium and calcitriol supplementation supports this fact.

The classical skeletal X-ray finding in renal osteodystrophy is "rugger jersey" spine, which is osteosclerosis of the superior and inferior ends of the body of vertebra.[7] Our patient paradoxically had an appearance that was the reverse of Rugger jersey spine (sclerotic center of the vertebra and lucent bands in the superior and inferior margins).

The reason for preserved renal function in Turner's syndrome has not been investigated previously. It is probably due to earlier detection with remedial measures in majority of the cases. The poor socioeconomic status and limited access to healthcare resources could be the cause of late detection of the disease in some cases like the present one. The mortality rate in these patients is increased and is primarily due to cardiovascular cause.[8] Our patient did not have any congenital heart disease. Since this was an atypical presentation of an uncommon syndrome, it was regrettably overlooked by local physicians. To our knowledge, this is the first reported case of renal osteodystrophy in Turner's syndrome in the literature.

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