Case Report

Sacrococcygeal Agenesis with Sacral Lipoma in a Child of a Non-Diabetic Mother

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Abstract

Sacrococcygeal agenesis or caudal regression syndrome is an uncommon disorder in which there is partial or total agenesis of the sacrum with or without lumbar involvement. Very few cases have been reported in the literature. We report a case of a 7-year-old child of a non-diabetic mother who presented with complaints of urinary and faecal incontinence since birth. Clinical examination revealed a soft, fluctuant swelling at the lower aspect of the back. The sacral and coccygeal bones were not palpable. CT scan and MRI confirmed sacrococcygeal agenesis with a sacral lipoma. He was being managed with pelvic floor training and physiotherapy with minimal improvement before being lost to follow up.

Keywords: Sacrococcygeal Agenesis; Sacral Lipoma; Non-Diabetic Mother.

Introduction

Sacrococcygeal agenesis is an uncommon disorder in which there is partial or total agenesis of the sacrum with or without lumbar involvement.[1] The frequency of the condition is 1 in 60,000 births and commonly affects boys than girls (3:1).[1] The exact aetiopathogenesis of sacral agenesis is not known but genetic predisposition like mutations of HLBX9, CYP26A1 and Wnt3a are implicated.[2]

Clinically, sacrococcygeal agenesis presents with range of anomalies including urologic, gastrointestinal, neural tube and orthopaedic abnormalities.[3] Prenatal ultrasound scan can be helpful in identifying fetuses with sacrococcygeal agenesis.[4] Plain radiograph, computed tomography (CT) and magnetic resonance imaging (MRI) identify dysplastic and osseous anomalies.[4]

A case of a 7-year-old boy with sacrococcygeal agenesis and sacral lipoma is thus reviewed with discussion of the aetiology, associations, clinical and radiological features, literature and conservative management.

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Case Report

Y. A is a 7-year-old boy that presented to the paediatric outpatient clinic of Aminu Kano Teaching Hospital with complaints of inability to achieve urinary and faecal continence. He also passes urine about ten times in 24 hours. He was a product of full-term gestation, the seventh child of his mother. There was no such abnormality in his siblings. His mother is not a known diabetic or hypertensive. His pregnancy and delivery were uneventful. He achieved all milestones at the expected time.

On examination, he was found to be clinically stable. A soft, fluctuant swelling at the lower aspect of the back was palpated. The sacral and coccygeal bones were also not palpable. His penile and anal openings were normal. Other systemic examinations were unremarkable.

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Abdominopelvic scan showed a sonographically normal urinary bladder with clear urine content within it (fig. 1). There was no calculus or pelvic mass seen. The kidneys were also sonographically normal (fig. 2). Other systems were sonographically normal.

Computed tomography scan showed absence of all the sacral and coccygeal bones with non-fusion of the ilium, ischium and the pubic bones but with widened symphysis pubis (fig. 3). The lumbar vertebral bodies were however preserved. There was also a hypodense structure with a fatty attenuation seen in the sacral region due to a lipoma (fig. 4).

Magnetic resonance imaging showed a normal spinal cord (fig 5). There was however a fatty structure seen in the sacral region due to lipoma but no evidence of syrinx, tethered cord or diastematomyelia. The sacral and the coccygeal bones were not visualized but the lumbar vertebrae were intact. Based on these findings, an assessment of type II sacrococcygeal agenesis with a concealed sacral lipoma was made. He was placed on pelvic floor training and physiotherapy though with little improvement. He was being planned for urinary and faecal diversion through a colostomy site but was lost to follow up after 3 months.

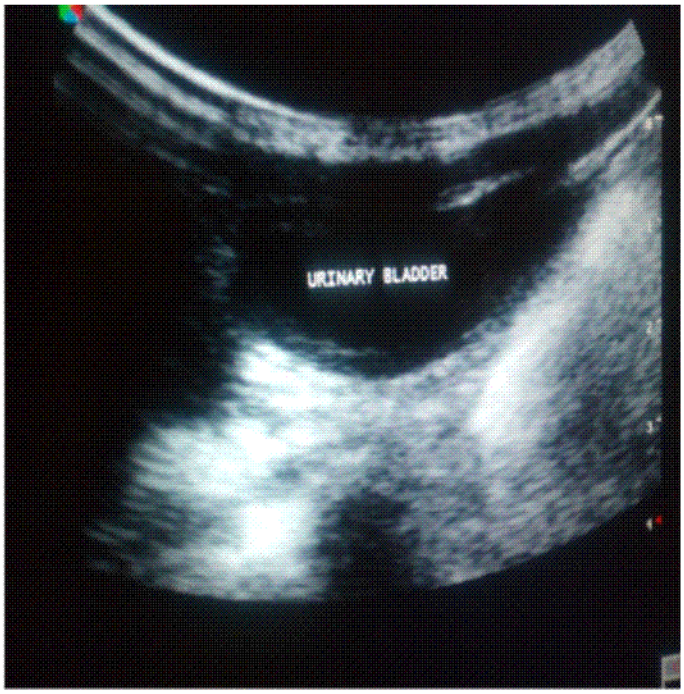

Figure 1: Grey scale transverse sonogram of the child showing the urinary bladder

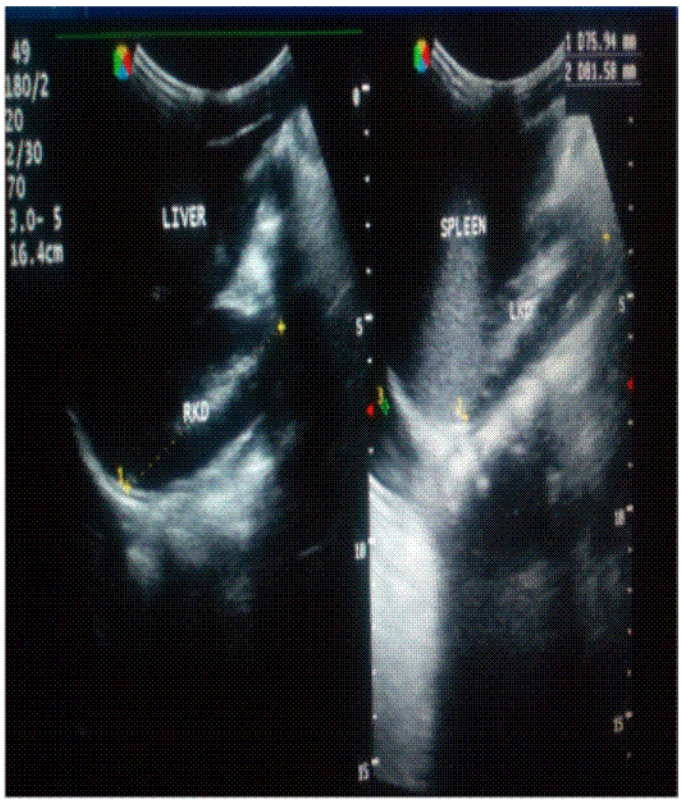

Figure 2: Gray scale longitudinal sonogram showing sonographically normal upper urinary tract system. RKD = Right Kidney, LKD = Left Kidney

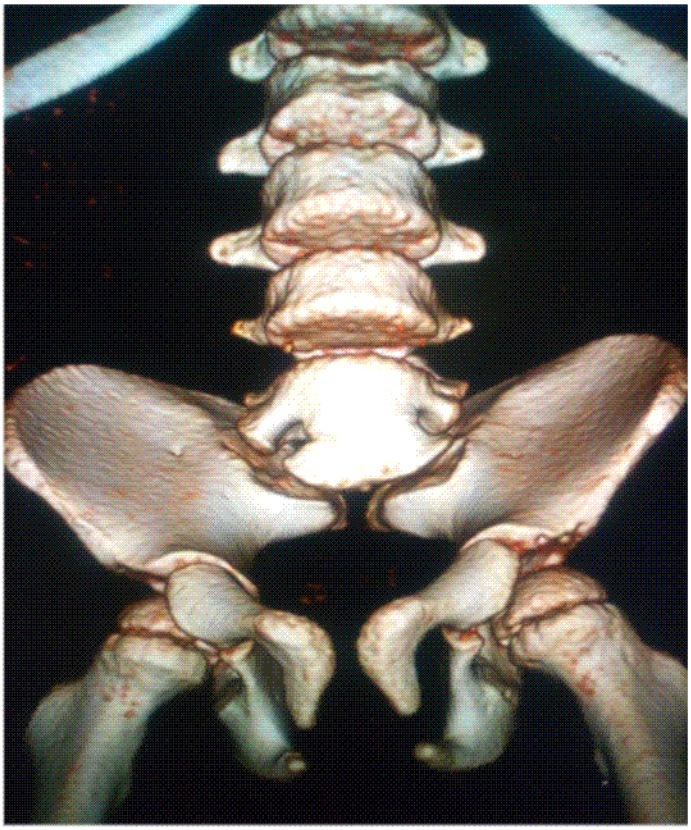

Figure 3: 3D volume rendered CT scan of the patient showing absence of all the sacral and the coccygeal bones.

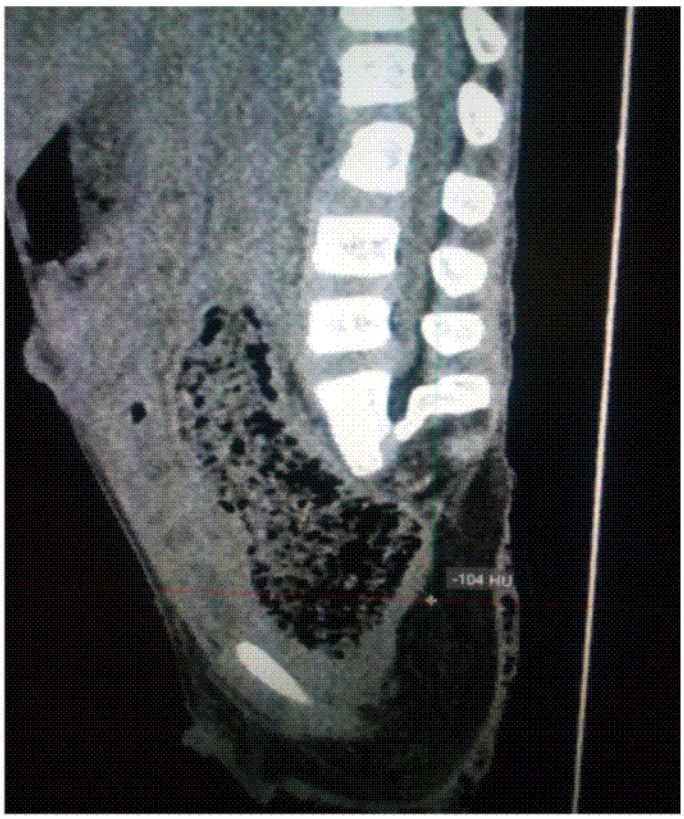

Figure 4: Sgaittal computed tomogrpahic image of the patient showing absence of the sacrococcygeal bones. There is a lipomatuous structure (fatty attenuation) seen at the sacral region.

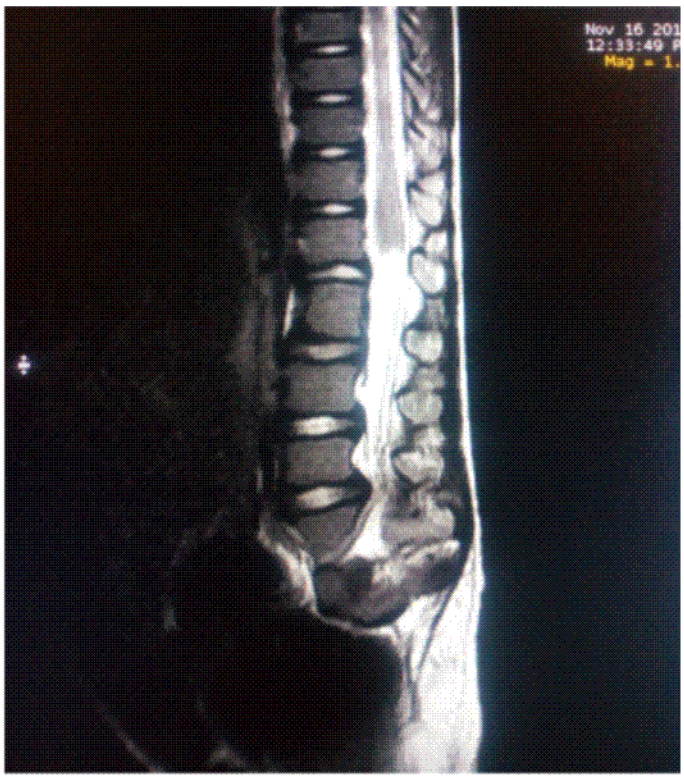

Figure 5: Sagittal T2 weighted MRT of the patient showing normal spinal cord. A hyperintense structure is seen at the sacral region.

Discussion

Sacrococcygeal agenesis or caudal regression syndrome (CRS) is a condition in which there is abnormal development of the caudal end of the fetal spine along with many associated anomalies.[5] The severity varies from absence of the coccyx to sacral or lumbosacral agenesis. Sacrococcygeal agenesis was classified by Renshaw into four types.[6] Type I is total or partial unilateral sacral agenesis. Type II sacral agenesis is total or partial bilateral, symmetric agenesis and is the most common type. Type III is total sacrococcygeal agenesis with variable lumbar agenesis with the last lumbar segment articulating with the ilia. Type IV is total sacrococcygeal agenesis with variable lumbar agenesis with the last formed vertebral segment resting on fused ilia or on an ilial amphiarthrosis.[6] The index case is a boy and presented with total and symmetric agenesis of all the sacral and the coccygeal bones but with preserved lumbar bones. This corresponds to the type II sacrococcygeal agenesis.

CRS is a consequence of abnormal development of the structures derived from the caudal mesoderm of the embryo before the 9th week of gestation.[5] The anomaly is thought to be due to defect in the HLBX9 homeobox gene, which is found in chromosome 7q36.[5] This gene is also expressed in the pancreas.[5] Therefore, association between diabetic hyperglycemia and CRS is proposed. Infants of diabetic mothers have a 200-fold increased risk of developing this condition compared to the general population and approximately 15-20% of the patients have diabetic mothers.[5] Other factors that are implicated in CRS include chromosomal abnormalities, toxins, infections, vascular hypoperfusion and excessive retinoic acid in the mother. It is not known what caused this condition in this case as his mother is not a known diabetic which is in contrast with most other reports.[5] Also, the mother could not recall ingesting medications that are known to be teratogenic during the pregnancy. However in this case report, chromosomal analysis was not done due to the fact that it is not readily available in our environment and this constitutes a limitation to the case report.

Patients with sacral agenesis present with many clinical abnormalities.[7] The most common are the genitourinary where neurogenic bladder is the commonest, seen in about 80% of patients.[7, 8] Renal defects include renal agenesis, horse-shoe kidney, ureteral duplication and bladder extrophy.[7] The index case presented with urinary incontinence likely from

neurogenic bladder, defective pelvic floor musculature and poor sphincteric activity. However, definitive diagnosis of neurogenic bladder with voiding cysto-urethrography was being planned when the patient was lost to follow-up. Other renal anomalies were absent in this patient. Associated congenital anomalies include gastrointestinal anomalies such as imperforate anus, malrotation; orthopaedic anomalies such as scoliosis, club feet; central nervous anomalies such as hydrocephalus, lipoma, lipomeningocoele; cardiac anomalies like atrial septal defect.[7] The index case presented with faecal incontinence and sacral lipoma but other anomalies were not found.

Although patients with sacral agenesis present at an early time, a significant proportion is detected later in childhood at near or after the time of expected toilet training.[8] This may be related to subtle physical signs noted on a careful history and or physical examination.[8] In this case, the patient presented for the first time in the hospital because he did not achieve urinary and faecal continence at the age of seven years. Also, the delay in presentation was probably due to absence of most of the associated anomalies. Another factor that might have contributed to the delayed presentation is poor parental knowledge of the child's condition.

Ultrasonography is usually employed initially to assess patients with sacrococcygeal agenesis to rule out other associated anomalies such as renal agenesis and horse-shoe kidney.[9] However, such associated anomalies were not found. Plain lumbosacral radiograph is employed to assess the lumbosacral spine.[9] Though CT scan gives adequate information of the bony abnormalities, MRI is the goal standard in evaluating patients with sacral agenesis.[10] In the index case, CT scan and MRI of the lumbosacral spine were used in the diagnosis.

Treatment of sacrococcygeal agenesis involves a team approach.[10] Urinary incontinence is treated with anticholinergic agents.[10] Other measures include pelvic floor training, urinary and faecal diversion as well as construction of sphincters.[10] The index case is currently on pelvic floor training and physiotherapy with possibility of considering other options until he was lost to follow up. Faecal incontinence is treated with sacral nerve stimulation.[11] The long-term morbidity consists mostly of a neurogenic bladder dysfunction, resulting in progressive renal damage and disabling neuromuscular deficits of the lower extremities.[10]

Differential diagnoses of sacrococcygeal agenesis include Curarino triad syndrome, femoral hypoplasia-unusual facies syndrome and sirenomelia.[6] Curarino triad syndrome is characterized by a hemi-sacral defect, autosomal dominant inheritance and not associated with infants of diabetic mothers. Femoral hypoplasia-unusual facies syndrome is highly associated with infants of diabetic mothers. It consists of severe femoral hypoplasia bilaterally, cleft palate, hypoplasia of the ala nasi and a long philtrum. Sirenomelia is a very rare case of fusion of the lower extremities into a single limb and variable lumbosacral agenesis. Bilateral renal agenesis and Potter's syndrome are almost always present in this condition.[6]

Key Lessons

- Sacrococcygeal agenesis is a rare condition although it should be suspected in a child of diabetic mother who presented with urinary and faecal incontinence.
- Its management requires multi-disciplinary approach.
- Good parental knowledge and health seeking behaviour are key to its management.
- As a limitation to this case report, chromosomal analysis and voiding cysto-urethrography were not done in this case report.

References

1. Chawla GS, Agrawal MP, Bajwa KS. A rare case of caudal regression syndrome in a fetus of non-diabetic mother: A case report. *Pol J Radiol.* 2017; **82**:621-624.
2. Temizkan O, Abike F, Ayvaci H, Yilmaz A, Demirag E, Sanverdi I. Prenatally diagnosed caudal regression syndrome. *Open Journal of Obstetrics and Gynaecology* 2013; **3**:227-231.
3. Bicakci I, Turgut ST, Turgut B, Icagasioglu A, Egilmez Z, Yumusakhuylu Y. A case of caudal regression syndrome: walking or sitting? *Pan Afr Med J*. 2014; 18: 92.
4. Boruah DK, Dhingani DD, Achar S, Prakash A, Augustine A, Sanyal S, et al. Magnetic resonance imaging of caudal regression syndrome and concomitant anomalies in paediatric patients. *J Clin Imaging Sci*. 2016; 6: 36.
5. Knight B. Caudal regression syndrome: A case report. AANA Journal. 2011; 79(4): 281-282.

6. Herman TE, Siegel MJ. Type IV lumbosacracral agenesis in an infant of diabetic mother. *J Perinatol*. 2008; **28**:163-165.
7. Al-Ghananim R, Awaysheh F, Maaytah M, Hadidi A, Al-Hamiedeen N. Lumbosacral agenesis in an infant of diabetic mother: A case report. *J Royal Med Serv*. 2014; **21**:75-78.
8. Cho PS, Bauer SB, Pennison M, Rosoklija I, Bellows AL, Logvinenko T, et al. Sacral agenesis and neurogenic bladder: Long-term outcomes of bladder and kidney function. *J Paediatr Urol*. 2016; **12**:158.
9. Kemer S, Eroglu A, Cuce F, Demiray O, Meral C, Atabey C. An incidentally detected sacral agenesis and associated anomalies: Case report. *Firat J Med*. 2017; **22**:149-52.
10. Kumar Y, Gupta N, Hooda K, Sharma P, Sharma S, Kochar P, et al. Caudal regression syndrome: A case series of a rare congenital anomaly. *Pol J Radiol*. 2017; **82**:188-192.
11. Thomas G, Nicholls R, Vaizey C. Sacral nerve stimulation for faecal incontinence secondary to congenital imperforate anus. *Tech Coloproctol.* 2013; **17**: 227-229.