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Case Report

Lumbopelvic Stabilization Outcome in Renshaw Type IV Sacral Agenesis: A Case Report and Review of the Literature

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Abstract

Background: Caudal regression syndrome (CRS), also known as sacral agenesis (SA), is a rare congenital disorder characterized by malformations of the caudal spine, lower limbs, urogenital, and anorectal systems. While the exact etiology remains unknown, a strong association with maternal diabetes mellitus (DM) has been observed.
Case Report: This retrospective study includes two patients diagnosed with Renshaw type IV CRS who underwent spinopelvic fusion. Both patients presented with back pain, severe thoracolumbar (TL) kyphotic deformity, and difficulty in sitting. Postoperative outcomes demonstrated improved sitting ability, enhanced mobilization, increased self-esteem, and better overall quality of life.
Conclusion: Orthopedic, neurological, and visceral anomalies are prevalent in patients with SA. Spinopelvic instability in type IV CRS significantly impairs sitting and mobilization. Surgical fusion can facilitate sitting and improve functional outcomes and cosmesis.

Keywords: Spinal Fusion; Sacral Agenesis; Caudal Regression Syndrome; Spinopelvic Fixation

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Background

Sacral agenesis (SA) is a rare congenital syndrome that affects the caudal parts of the spine and is commonly associated with scoliosis, kyphosis, spinopelvic instability, and lower extremity malformations such as clubfeet and knee and hip flexion contractures (1-11). Visceral malformations, including gastrointestinal, genitourinary, and cardiac anomalies, are frequently observed in this condition (12-15).

Caudal regression syndrome (CRS) is a rare anomaly with an incidence of one per 25000 live births (16). It is mostly non-familial and sporadic; however, specific variants of CRS, such as the Currarino triad, can be inherited in an autosomal dominant pattern (3, 13, 17-23). CRS has a strong association with maternal diabetes, but the causal relationship remains unknown. While gestational diabetes mellitus (DM) is observed in 16 to 49 percent of cases, caudal spinal anomalies are present in only one percent of children born to mothers with DM (8, 11, 16).

Renshaw classified this syndrome into four types based on the osteological defects between the spine and the sacrum (1, 6, 7, 11). Type I is total or partial unilateral SA. Type II, which is the most common type, involves partial SA with a normal or hypoplastic first sacral vertebra (6). Type III is characterized by variable lumbar and total SA, with the ilia articulating with the sides of the lowest vertebra. Type IV includes variable lumbar and total SA, where the caudal endplate of the lowest vertebra rests above fused ilia or an iliac amphiarthrosis (6). The severity of anomalies determines the extent of symptoms, comorbidities, and functional limitations.

This report presents two rare cases of Renshaw type IV SA managed through lumbopelvic stabilization. It aims to underscore the surgical value of spinopelvic fusion in enhancing mobility and functional outcomes in patients with this debilitating condition.

Case Report

Case 1: A four-year-old boy, born full-term via vaginal delivery to a mother with DM with no known history of congenital malformations, presented with sensory and motor abnormalities in the lower extremities (Figure 1). Clinical examination revealed a thoracolumbar (TL) kyphotic deformity, bilateral clubfeet, hip and knee flexion contractures, popliteal webbing, a shortened gluteal cleft, flattened buttocks, and bilateral sacral dimples. The patient was wheelchair-bound and unable to ambulate. Additionally, dextrocardia was noted.

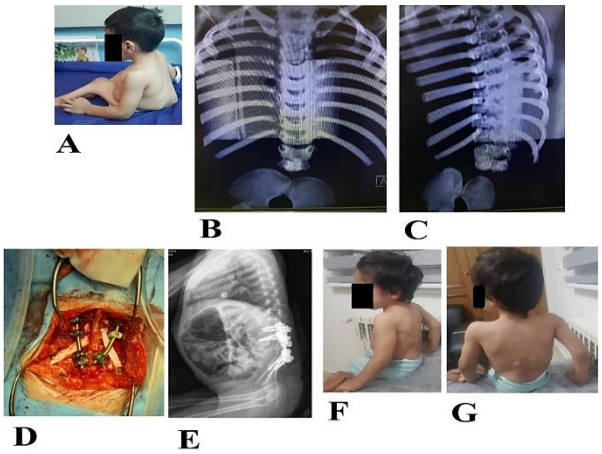

Figure 1. A) Preoperative photography of the patient that shows sever thoracolumbar (TL) kyphosis and knee webbing; B, C) Anteroposterior (AP) and lateral radiographies consistent with type IV Renshaw sacral agenesis (SA); D) The surgical instrumented fusion with fibular allograft; E) Postoperative radiography; F, G) Postoperative photography

Radiographic evaluation demonstrated complete absence of the L3, L4, and L5 vertebrae, a defective L1 vertebra, and total agenesis of the sacrum and coccyx, consistent with Renshaw type IV SA. Magnetic resonance imaging (MRI) revealed a tethered cord with the conus medullaris terminating at the L4 level. The ischium, ilium, and pubic bones were hypoplastic. The patient had impaired urinary control since infancy, characterized by persistent overflow incontinence. Clinical and ultrasonographic assessments confirmed bilateral undescended testes.

The patient underwent instrumented spinopelvic fusion using allograft material, along with surgical release of the tethered cord. Postoperatively, the patient achieved independent sitting ability and experienced significant improvement in bladder control.

Case 2: A three-year-old boy, born full-term via vaginal delivery to a mother with DM, presented to the orthopedic department with TL kyphosis, hip and knee flexion contractures, flattened buttocks, and bilateral lower limb atrophy. The patient was wheelchair-bound and unable to ambulate.

Radiographic evaluations revealed agenesis of the L3 to L5 vertebrae, defective L1 and L2 vertebrae, and complete absence of the sacrum and coccyx, consistent with Renshaw type IV SA (Figure 2). The patient had experienced poor urinary control since infancy, characterized by persistent overflow incontinence.

The patient underwent instrumented spinopelvic fusion using allograft material. Postoperatively, the patient exhibited improved sitting ability and gained better control over bladder function.

Discussion

SA is a rare congenital syndrome with unclear etiology; however, a strong association with maternal DM has been documented (1, 3, 11, 24). Phillips et al. reported that fifty percent of mothers of children with SA had gestational DM (25). Currarino et al. described an autosomal-dominant triad comprising sacral dysgenesis, anorectal malformation, and presacral mass (13).

SA is associated with various orthopedic abnormalities, including hip dislocation, knee and hip flexion contractures, foot anomalies, scoliosis, spinopelvic instability, and myelomeningocele (10, 11, 25). Jeelani et al. described a non-syndromic form encompassing total agenesis of the caudal spine from lower thoracic vertebrae to the coccyx (26). Visceral anomalies –gastrointestinal, genitourinary, and neurological – also frequently accompany SA (3, 5, 11, 22).

Emami-Naeini et al. evaluated fifty patients and recommended comprehensive neurologic and urologic assessment in all SA cases (4). Spinal deformities such as TL kyphosis, scoliosis, and spinopelvic instability occur commonly in SA (10).

Several surgical techniques have been proposed for correction. Griffet et al. described lumbopelvic distraction and stabilization (27), while Yazici et al. reported successful outcomes using posterior lumbopelvic instrumentation and fusion (28). Severe TL kyphosis and pelvic obliquity can impair sitting posture and lead to pressure sores (29). Surgical correction enhances sitting ability, cosmesis, patient care and hygiene, transfers, self-esteem, and overall quality of life.

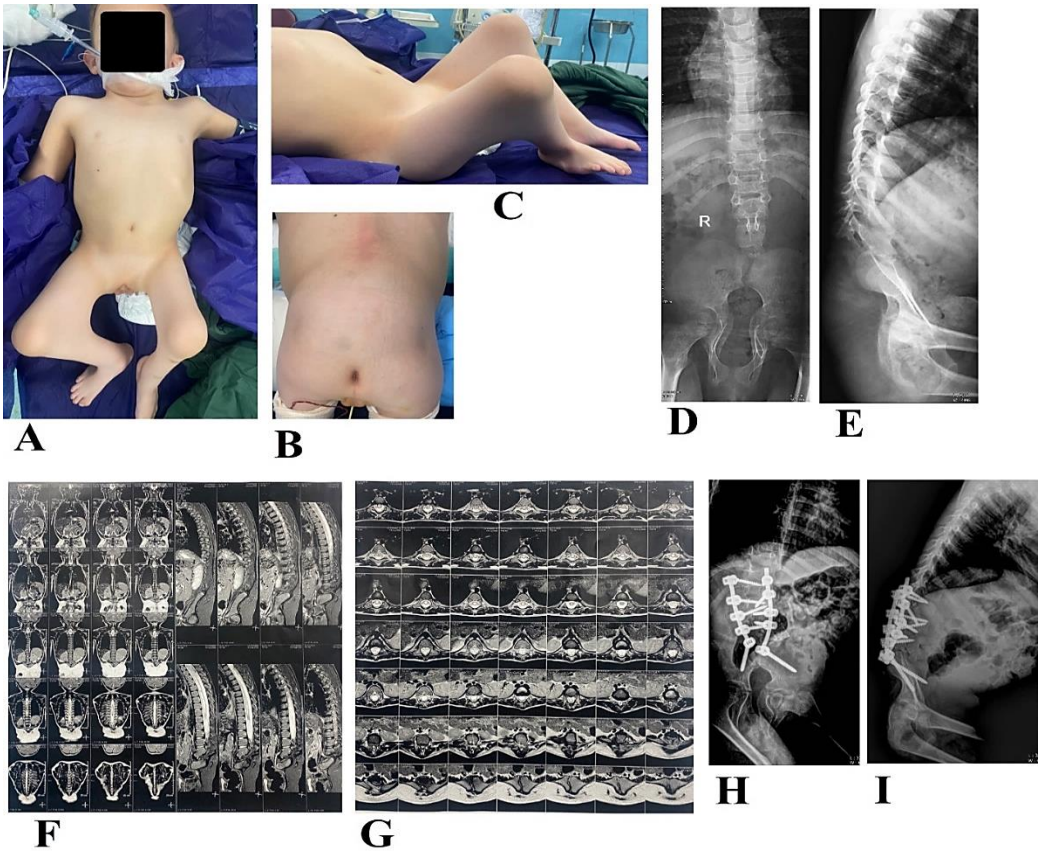

Figure 2. A, B, C) Preoperative photography; D, E, F, G) Preoperative radiographies and magnetic resonance imaging (MRI); H, I) Postoperative radiographies

Management should be individualized based on spinal, musculoskeletal, and visceral involvement. Mild sacral deformities are often managed conservatively, whereas symptomatic cases require surgery (1, 11, 28-30). Procedures may include tethered cord release or decompressive duraplasty for symptomatic dural stenosis (1, 11, 28-30).

Scoliosis is the most common spinal deformity in SA (1, 10, 11). Progressive scoliosis or kyphosis often necessitates surgery to improve posture and enable effective rehabilitation (27, 28, 31). Corrective surgery may also enhance motor function and bowel and bladder control (29). Urinary incontinence and recurrent urinary tract infections (UTIs) are common but whether spinal cord release improves these remains uncertain (3).

Phillips et al. noted that type I and II deformities had excellent ambulatory potential and recommended surgery for lower limb deformities, such as open reduction for hip dislocation or osteotomy for knee flexion contracture (25). In contrast, managing spinopelvic instability in type III and IV cases is more controversial and technically demanding, with high complication rates (1, 10, 11). Some suggest that lumbopelvic fusion may further limit sitting in patients with stiff hips (1, 10, 11).

Spinopelvic fusion and instrumentation carry high complication risks, including skin breakdown, surgical site infection, and pseudoarthrosis (2, 27, 28). Ferland et al. reported high fusion rates using vascularized rib grafts (32).

Effective management of SA requires a multidisciplinary approach – including pediatricians, neurosurgeons, orthopedists, urologists, and physical therapists –with careful preoperative planning. The main limitation of this study is the small number of patients and its short term.

Conclusion

SA is a congenital spinal disorder associated with a wide range of orthopedic, neurological, and visceral comorbidities. The functional and financial impacts of this disease are debilitating to patients and their families. Lumbopelvic instability significantly reduces patients' quality of life; however, surgical stabilization can improve function, sitting ability, and cosmesis.

Conflict of Interest

The authors declare no conflict of interest in this study.

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