

Caudal Regression Syndrome in an infant of a diabetic mother: A 14-year follow-up case report

Abstract

Caudal Regression Syndrome (CRS) is a developmental anomaly affecting the caudal spine, spinal cord, and associated organ systems. CRS is strongly associated with poor maternal glycemic control in early pregnancy. We report a male neonate born to a mother with poorly controlled type 2 diabetes. Postnatal examination and imaging confirmed CRS with an associated horseshoe kidney. The infant was referred to a multidisciplinary team and is now 14 years old. The patient has had extensive management of orthopedic, urinary and bowel complications. Despite these setbacks, he successfully attends school and has a positive outlook on life.

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Introduction

Caudal Regression Syndrome (CRS) is a rare congenital condition characterized by abnormal development of the caudal spine and spinal cord, ranging from isolated partial sacral agenesis to complete lumbosacral agenesis [1]. The overall incidence is estimated at 1-2.5 per 100,000 live births [2]. The condition is most commonly classified using the four-type Renshaw system, based on the extent and symmetry of sacral and lumbar vertebral absence [1].

CRS is thought to result from disruption of caudal mesoderm development between the third and seventh weeks of gestation, a critical period for formation of the lower spine, spinal cord, hindgut, and genitourinary structures [3].

Maternal diabetes is the best-established risk factor for CRS. It is identified in approximately 16-22% of reported cases and the incidence of CRS rises from 1-2.5 per 100,000 live births in the general population to approximately 1 in 350 live births among infants of diabetic mothers - roughly a 200-fold increase in risk [2,4]. However, maternal diabetes does not account for all cases: CRS has also been described in normoglycemic pregnancies, where genetic predisposition, vascular hypoperfusion of the caudal embryo, and sporadic occurrence have been proposed as alternative mechanisms [5].

Most published reports of CRS describe the antenatal, neonatal, or early childhood presentation, with comparatively little long-term outcome data available once children reach adolescence. We report a case of severe CRS in an infant of a diabetic mother, with prospective follow-up extending to 14 years of age, to describe both the index presentation and the real-world functional, medical, and psychosocial trajectory of this condition over time.

Case presentation

A male neonate was born at 40 weeks' gestation to a 33-year-old gravida 3 para 2 mother with type 2 diabetes mellitus on insulin. Glycemic control during pregnancy was poor (HbA1c 11.8%). She booked for antenatal care at 14 weeks' gestation. Antenatal ultrasound first raised concern for sacral agenesis at 18 weeks' gestation and demonstrated an associated horseshoe kidney, with the right kidney enlarged and crossing the midline and the smaller left kidney fused to it.

The infant was delivered by emergency caesarean section for fetal distress. Apgar scores were 8 at 1 minute and 9 at 5 minutes; no active resuscitation was required. Birth weight was 2480 g.

On postnatal examination, the infant had flattening of the gluteal region, an abnormal gluteal cleft, overlapping toes bilaterally, and reduced spontaneous movement and tone in the lower limbs, with no upper limb abnormalities (Figure 1a and 1b). Plain radiography of the spine confirmed partial sacral agenesis involving the lower sacral segments (Figure 2a and 2b). Renal ultrasound confirmed the horseshoe kidney seen antenatally.



Figure 1: (a) Anterior view showing reduced spontaneous lower-limb movement and abnormal positioning (genitalia redacted for publication); (b) posterior view showing flattening of the gluteal region and an abnormal gluteal cleft.



Figure 2: (a) Anteroposterior and (b) lateral plain radiographs of the spine demonstrating partial sacral agenesis.

The infant was assessed by a multidisciplinary team comprising neonatology, orthopedics, and urology, and was discharged with plans for ongoing multidisciplinary follow-up.

Follow-up at 14 Years: This patient has remained under multidisciplinary follow-up, including the renal, orthopedic, spinal defect, and stoma clinics, and is now 14 years old.

From an orthopedic perspective, at age 11 he underwent bilateral through-knee amputation for severe, progressive lower-limb contractures to facilitate wheelchair use. He is fully independent in his wheelchair, propelling himself without assistance and reporting no difficulties with day-to-day mobility (Figure 3).

He has urinary incontinence and wears nappies and is followed at the stoma clinic for management of neurogenic bowel dysfunction, consistent with neurogenic bladder and bowel dysfunction secondary to his underlying spinal anomaly.

He attends a specialized educational and therapeutic school for learners with cerebral palsy, physical disabilities, neurodiversity, and other specific learning or medical needs, and lives on site as a boarding pupil during term time. He is passing his classes and attends school consistently.

Socially, he is described by his family as living a normal teenage life and integrating well with his peers despite his physical disability. From a psychological perspective, he is reported to be coping well: he has not displayed prolonged low mood and has not required input from a counsellor or psychologist. The family receives a disability grant in support of his care.

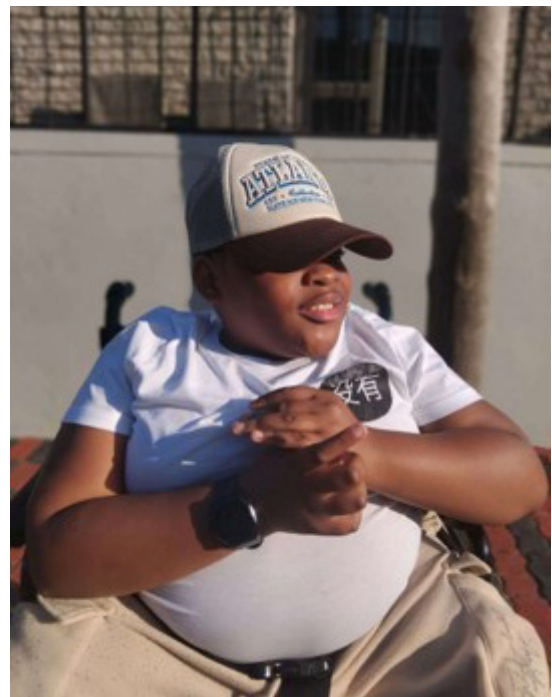


Figure 3: The patient at 14 years of age. Written consent for use of this photograph, including his face, was obtained from the patient and his family.

Discussion

CRS is well described in the antenatal and neonatal period, but published follow-up rarely extends beyond early childhood. This report is therefore notable for 14 years of prospective follow-up, allowing the trajectory of this index case to be considered directly against the limited long-term outcome data available in the literature.

Antenatal detection in this case was early, with sacral agenesis first suspected at 18 weeks' gestation and confirmed on serial scans. This is consistent with literature describing improving antenatal ultrasound and fetal MRI detection of CRS and illustrates the value of antenatal surveillance in pregnancies complicated by poor glycemic control [6,7].

The birth weight of 2480 g in this case is unexpectedly low for a term infant of a diabetic mother, in whom macrosomia is a more typical finding. This may reflect intrauterine growth restriction secondary to severe, sustained maternal hyperglycemia - a recognized but less commonly reported complication of poorly controlled diabetes in pregnancy.

The horseshoe kidney identified in this index case reflects the well-documented overlap between caudal regression and genitourinary malformation, which share a common

embryological origin in the caudal mesoderm [7,8]. The persistence of urinary incontinence and neurogenic bowel dysfunction requiring stoma clinic follow-up at 14 years in this patient is concordant with the largest available long-term cohort data: Cho reported ongoing bladder dysfunction requiring lifelong urological surveillance in their cohort of sacral agenesis patients followed from infancy [9]. Similarly, Diallo et al highlights chronic bowel dysfunction as an under-recognized long-term complication of CRS and Thompson et al. found that 70% of patients with sacral agenesis had impaired or absent bladder control on long-term follow-up [10,11]. This reinforces the recommendation that bladder and bowel surveillance - including stoma care where required - should be lifelong rather than confined to early childhood.

The orthopedic course in this patient - progression to bilateral through-knee amputation at age 11 - is a more severe outcome than is typically emphasized in the literature, which more often describes conservative bracing or limited surgical correction of contractures [12]. The decision to proceed to amputation rather than prolonged attempts at limb salvage appears to have resulted in a functionally independent wheelchair user, which is a useful real-world data point for clinicians counselling families about this pathway.

Cognitively and educationally, this patient is performing well at a specialized school for physically disabled learners, passing his classes consistently as a boarding pupil. His academic progress in an appropriately adapted educational setting illustrates that suitable school placement is both achievable and effective for children with severe CRS. This is a domain largely absent from the existing CRS literature, which focuses heavily on motor, urological, and radiological outcomes; this case therefore adds outcome data in an area where the literature currently offers very little long-term comparison.

From a psychosocial perspective, this patient appears to be coping well at 14 years without formal psychological input, which is notable given that the literature consistently identifies psychosocial adaptation as a major and ongoing concern in CRS, particularly given the chronic impact of the condition on mobility and independence. Thomson et al. found that quality-of-life scores differed between younger and older patients with sacral agenesis, which they interpreted as reflecting a long-term adaptive process; the apparent good adjustment seen in this adolescent may represent a similar adaptive trajectory [11]. However, the absence of low mood reported by the family should not be taken to mean that this patient has no unmet psychological needs, and we would recommend that psychological screening remain part of his ongoing multidisciplinary follow-up rather than being guided solely by the absence of overt distress.

Finally, this index case was managed in a public-sector, resource-constrained setting in South Africa, with the family supported in part by a government disability grant of R2,400 per month (approximately \$130 USD). Much of the published long-term outcome literature on CRS originates from single tertiary centers in high-income settings [9,11], this case illustrates that comparable long-term functional and educational outcomes are achievable in a lower-resource multidisciplinary setting, provided follow-up is sustained.

Conclusion

This case report describes a child with severe caudal regression syndrome born to a mother with poorly controlled type 2 diabetes mellitus (HbA1c 11.8%), who has been followed prospectively for 14 years - one of the longest documented follow-up periods for this condition in the published literature. Despite the resource-constrained setting in which he has been managed, this patient demonstrates satisfactory physical functioning and psychosocial adjustment at 14 years of age.

Declarations

Acknowledgements: We thank the patient and his family for their consent to the publication of this report and accompanying clinical photographs.

Consent: Written assent from the patient and consent from his family were secured for the use of clinical photographs in this report.

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