Letter to the Editor

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Caudal regression syndrome in a newborn in distress: A therapeutic challenge

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DEAR SIR

Caudal regression syndrome (CRS) is a rare congenital malformation characterized by varying degrees of sacral and coccygeal vertebrae agenesis, along with shortened femurs and abnormalities in the gastrointestinal, genitourinary, and cardiovascular systems. It affects less than 0.5% of newborns [1]. In this case, a full-term male neonate with neonatal distress and a polymalformative syndrome was admitted to the neonatal intensive care unit. The parents, first cousins in their late twenties, had no comorbidities or family history of congenital deformities. The antenatal period was uncomplicated, except for managed gestational diabetes with insulin, and no exposure to other drugs. Antenatal ultrasound screening was normal.

The infant was delivered via emergency cesarean section due to severe fetal distress. On admission, he exhibited severe respiratory distress with an oxygen saturation of 70% and a rapid respiratory rate of 80 breaths per minute, necessitating immediate intubation and mechanical ventilation. A chest X-ray (Fig. 1) revealed a spontaneous right pneumothorax, which was drained.

Physical examination uncovered prominent facial deformities, including a short neck, low-set ears, left facial paralysis, and convergent strabismus. Additionally, he had a caudal spine deformity with hypoplastic lower extremities, a splayed posture with bilateral hip dislocation, flexed knees, and bilateral congenital talipes equinovarus deformity. Furthermore, the infant presented with an anorectal malformation, evident through visibly flat buttocks and an absent anus (Fig. 2).

Caudal regression syndrome was suspected, prompting a comprehensive diagnostic evaluation, including a full skeletal survey (Fig. 1), echocardiogram, and abdominal ultrasound. The findings revealed sacro-coccyx spine agenesis, hypoplastic iliac bones, thigh abduction with knee flexion, and congenital talipes equinovarus. Additionally, an echocardiogram identified a 3 mm ventricular septal defect with a hypertrophic septum, while abdominal ultrasound revealed a dysplastic right kidney with ipsilateral ureteropelvic junction syndrome. Pelvic ultrasound indicated a sacrum absence, and serum creatinine levels were within the normal range.

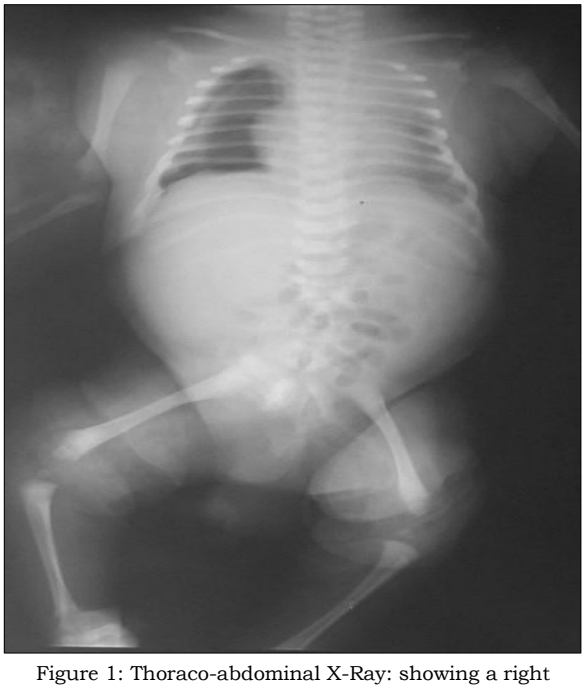

Figure 1: Thoraco-abdominal X-Ray: showing a right pneumothorax and sacro-coccyx spine agenesis with hypoplastic iliac bones.

The diagnosis of caudal regression syndrome was confirmed, and the parents received counseling regarding the long-term prognosis. A sigmoidostomy was performed on day three of life as a permanent treatment for the high anorectal malformation due to the unattainable continence for his condition. Extubation occurred on day ten of life, and breast-feeding was initiated successfully. Consultations with orthopedists, cardiologists, and neurologists were scheduled, with no indications for cardiac or

neurological surgical interventions. Orthopedic treatments aimed at improving pelvic stability were initiated, primarily providing support since complete recovery was not achievable.

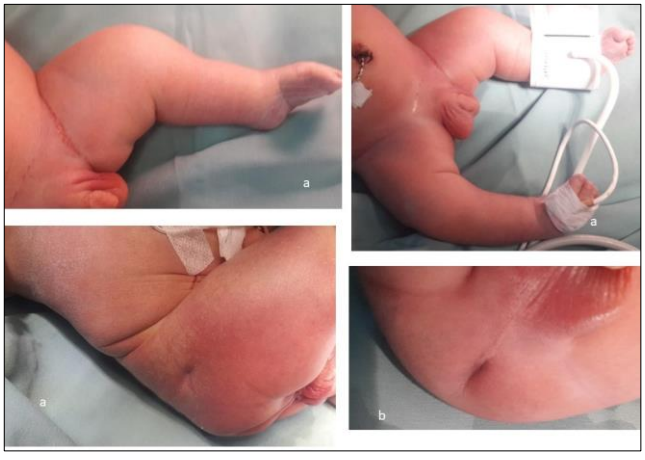

Figure 2: Clinical photographs revealing a caudal spine deformity with hypoplastic lower extremities and splayed posture with bilateral hip dislocation, flexed knees, and bilateral congenital talipes equinovarus deformity (a) and an anorectal malformation (b).

The infant undergoes regular check-ups with a multidisciplinary team, and a long-term follow-up plan is in place.

Caudal regression syndrome is an early embryological developmental failure affecting 1 in every 60,000 births [2] and 1 in every 350 births in diabetic mothers. The impact of hyperglycemia is linked to DNA structure malformation [3]. While other environmental factors have been identified, they were not present in this case. Although its male predilection is not firmly established in the literature, it is strongly suggested [3].

Antenatal diagnosis is possible and can aid in perinatal care planning, but its accuracy varies with the severity of the syndrome [4]. Postnatally, diagnosis relies on clinical and radiological examinations. The characteristic "frog-like" position, hip abduction deformity, and irreducible knee flexion are observed in affected infants [5]. Visceral malformations often accompany the syndrome, necessitating a comprehensive radiological workup to rule out issues in various systems.

Our patient had multiple associated anomalies requiring a multidisciplinary approach and intricate treatment planning. Initial concerns focused on pulmonary distress and anorectal malformation, requiring immediate attention. The remaining malformations call for a step-by-step treatment of each system, but consensus on treatment is lacking due to the complexity and poor prognosis of the syndrome [6].

The next area of focus is orthopedics; however, with both lumbar and complete sacral agenesis and bilaterally dysplastic hips, the functional prognosis is poor. Treatment emphasizes rehabilitation and stabilization to enhance the patient's quality of life through stable sitting and ambulation [3]. Neurological deficits are usually irreversible, limiting potential interventions.

Long-term follow-up is crucial to prevent complications like growth impairment and neurogenic bladder. Prognosis is determined by the severity of associated abnormalities and the timing of interventions, contingent upon the cooperation of the patient's family.

In summary, caudal regression syndrome is a rare condition posing therapeutic challenges. Given its poor prognosis, prenatal diagnosis and managing diabetes and other implicated etiologies should be the primary therapeutic concern.

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