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CASE PRESENTATION

Caudal Regression Syndrome with Absence of the Sacrum. A Case Report

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ABSTRACT

Introduction: caudal Regression Syndrome with Absence of the Sacrum is a rare congenital malformation characterized by the partial or total absence of the sacrum, associated with vertebral, genitourinary, and gastrointestinal anomalies. Its diagnosis is based on imaging studies, and treatment requires a multidisciplinary approach, including orthopedic, urological, and gastrointestinal management.
Objective: to present a clinical case of Caudal Regression Syndrome, highlighting its clinical manifestation, diagnosis and management.
Case presentation: a female newborn, the product of dystocic labor at 34,4 weeks, was admitted 50 minutes after birth due to moderate vaginal discharge seven hours prior. The mother had a history of gestational diabetes. Upon admission to the neonatal unit, the neonatal MAMA score was 0. At 20 minutes of life, the neonatal desaturation level was 79 %, with no signs of respiratory distress. The neonatal neonate required free-flow oxygen and was admitted to the neonatal unit for evaluation. Imaging studies showed an absence of the sacrum and lumbosacral vertebral abnormalities.
Conclusions: the entity evaluated is a rare congenital disorder that affects the development of the lower spine, associated with maternal diabetes and environmental factors. Its clinical presentation varies, ranging from skeletal abnormalities to complex visceral malformations. Timely prenatal and postnatal diagnosis is key to optimizing management. Treatment is multidisciplinary and personalized, integrating metabolic control, corrective surgeries, and rehabilitation therapies to improve the patient's quality of life and functionality.

Keywords: Anodontia; Sacrum; Regression; Caudal.

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INTRODUCTION

Caudal Regression Syndrome with Absence of the Sacrum (CRS) is a rare congenital condition that affects the development of the caudal area of the human body, particularly the sacral region. Unlike other congenital anomalies, CRS is characterized by complete or partial absence of the sacrum, a crucial bony structure at the base of the spine. This absence may be accompanied by other malformations in the vertebral column, pelvis, and urinary and gastrointestinal systems.[1,2]

CRS manifests from birth and may present a wide range of symptoms and complications, from mobility problems and spinal deformities to dysfunctions in sphincter control and disorders of the urinary and digestive apparatus. The severity of the condition can vary considerably from one patient to another, depending on the extent of the involvement of the caudal structures and the presence of associated anomalies.[3,4]

Diagnosis of CRS is made through imaging studies such as X-rays, MRIs, and CT scans, which allow visualization of malformations in the spine and pelvis. Treatment of CRS is typically multidisciplinary and includes measures to address potential orthopedic, urological, and gastrointestinal complications.[5] This may involve corrective surgeries, physical therapy, pain management, and other specific interventions based on each patient's individual needs. Given this information, we present the clinical case of a patient diagnosed with Caudal Regression Syndrome.

CLINICAL CASE REPORT

Female newborn, admitted 50 minutes after birth (04/09/2023), born via dystocic delivery at 34.4 weeks of gestation, presenting moderate vaginal fluid leakage seven hours earlier, with a MAMA score of 0.

Current illness history:
• Maternal history: gestational diabetes untreated four years ago.
• Gynecological-obstetric history: G:2 P:0 C:2 A:1 HV:2
• Prenatal history: three inadequate prenatal check-ups, recurrent vaginal infection, threatened preterm labor, rupture of membranes seven hours prior with fetal well-being compromise category II due to fetal bradycardia of 120 bpm.
• Natal history: light meconium-stained amniotic fluid with few clumps, timely clamping of the umbilical cord.
• Postnatal history: at 20 minutes of life, presents desaturation to 79% without signs of respiratory distress with free-flow oxygen supply and admission to neonatology.

Anthropometric data at birth:
• Weight: 2670 g (P: 82,8 %)
• Length: 43.5 cm (P: 30 %)
• Head circumference: 32,5 cm (P: 72,2 %)
• APGAR: eight at one minute, nine at five minutes

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Vital signs:
• Heart rate: 139 bpm
• Respiratory rate: 56 rpm
• Saturation: 93 % room air
• Temperature: 36,3°C
• Glucose: 52 mg/dl
• Blood group: Positive

General physical examination:
Female newborn, active, responsive, afebrile, and hydrated.
• Skin: Turgor preserved.
• Head: Normotensive fontanelles.
• Eyes: Isocoric pupils, normoreactive to light and accommodation.
• Neck: Mobile, symmetrical.
• Lungs: Preserved vesicular murmur. Presents episode of saturation down to 79 % at 20 minutes of life, thus oxygen was administered via nasal cannula, maintaining adequate saturations thereafter.
• Heart: Rhythmic, without murmurs.
• Abdomen: Soft, depressible, with present hydro-aerial sounds. No visceromegaly.
• Inguinal region: Female genitalia. Patent anus.
• Spine: Presence of sacrococcygeal fossa.
• Extremities: Symmetrical, with active movements. Capillary refill: 2 seconds.
• Neurological examination: Within normal parameters.

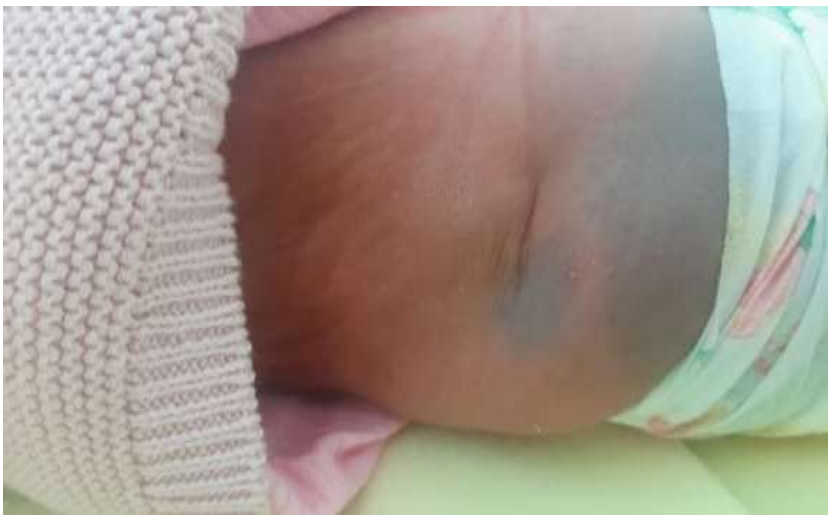

Fig. 1 Image of a baby with sacrococcygeal fossa agenesis.

Analytical data:
• Complete blood count: Leukocytes: 16,82 x103/uL, Hb: 19,1g/dl, Hct: 59,3, Platelets: 243,000.
• Arterial blood gases: pH: 7,40, PCO2: 36, HCO3: 22,2, BE: -2,5, bilirubin: 4,1, Na: 137, K: 5,62, Ca: 1,10, Cl: 106.

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Imaging examination:

• X-ray of the spine, lumbar-dorsal region: agenesis of lumbar vertebrae L4, L5 and sacrococcygeal (Fig. 2).
• MRI of the lumbosacral spine: caudal regression syndrome with total absence of the sacrum.
• Echocardiogram: small muscular interventricular communication, without repercussions, small secundum atrial septal defect, medium repercussion, no signs of pulmonary hypertension (Fig. 3).

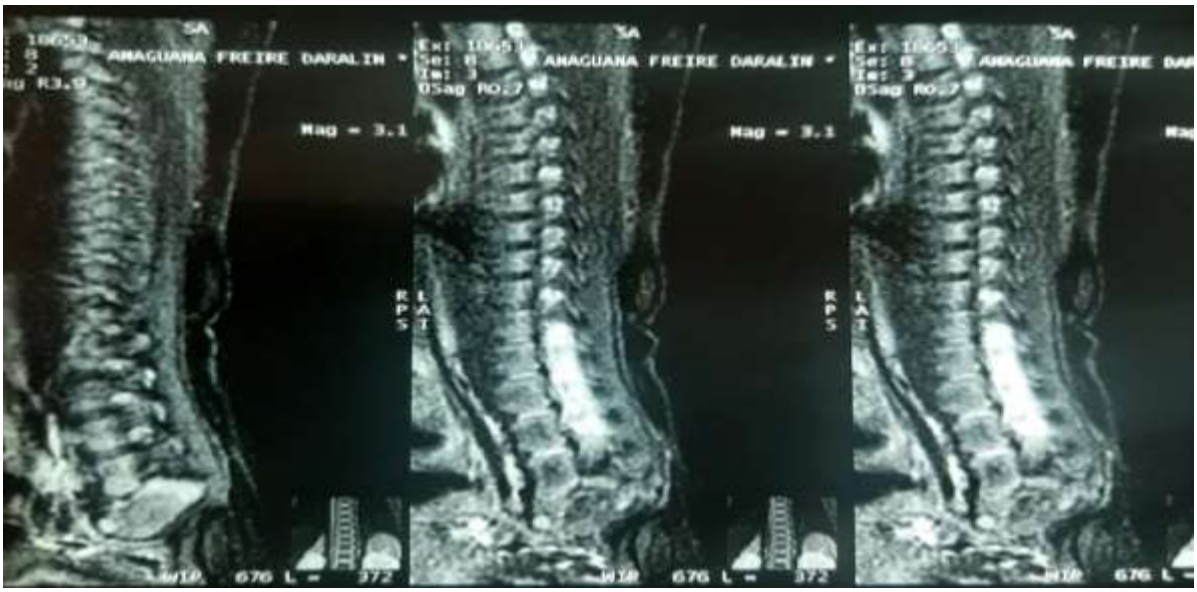

Fig. 2 Image of agenesis of lumbar vertebral bodies L4, L5 and sacrococcyx.

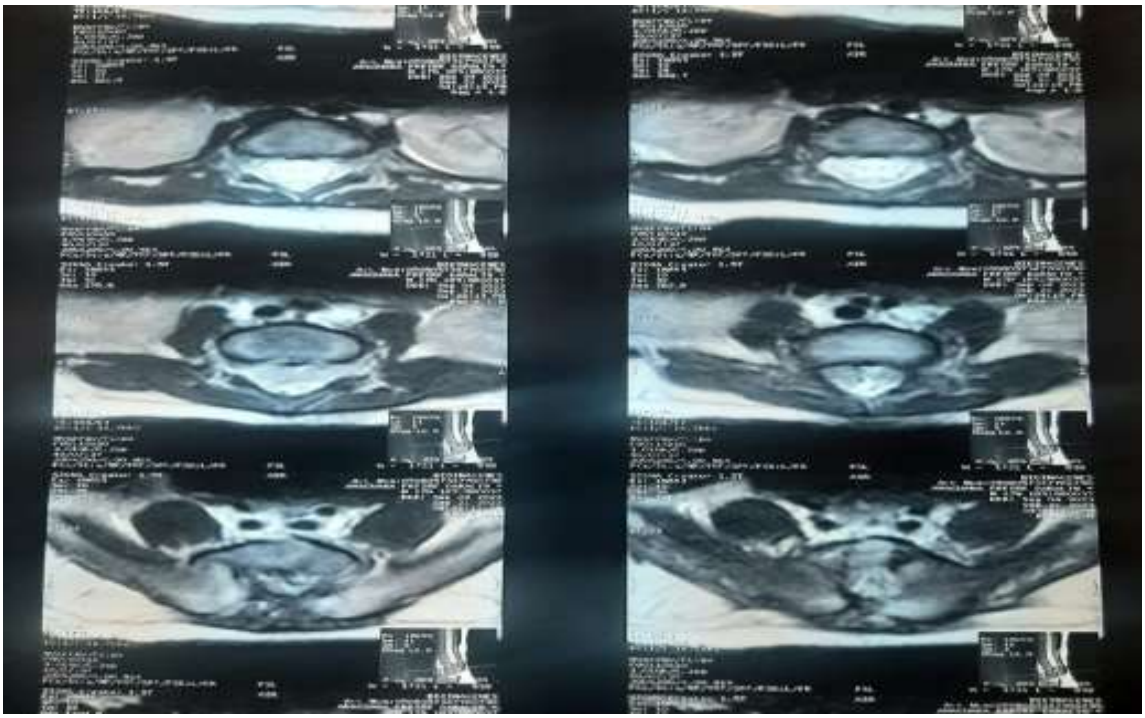

Fig. 3 Image of caudal regression syndrome with total absence of the sacrum.

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Table 1. Problem-based analysis.

ACTIVES	PASIVES
• Prematurity less than 34 weeks • Absence of sacrococcygeal bone • Absence of lumbar vertebrae L4- L5 • Threatened preterm labor • Fetal bradycardia • Cytobacteriological of vaginal secretion positive for gram-negative bacteria • Cardiac alterations	• Gestational diabetes • Maternal history of alcohol, drug, and tobacco use

Table 2. Differential Diagnosis.

Symptoms and signs	Agenesis of sacrum	Spina bifida	Currarino syndrome	Spinal-costal dysostosis	Caudal regression syndrome	VACTERL syndrome
Gestational diabetes	X	X			X	X
Prematurity less than 34 weeks	X	X			X	X
Absence of lumbar vertebrae L4- L5	X			X	X	X
Absence of sacrococcygeal bone	X	X	X	X	X	X
Threatened preterm labor	X	X			X	
Fetal bradycardia	X	X			X	
Cytobacteriological of vaginal secretion positive for gram-negative bacteria	X				X	
Maternal history of alcohol, drug, and tobacco use	X	X			X	X
Cardiac alterations	X				X	X

Source: Authors

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When comparing the compiled clinical data, it can be observed that these coincidences occur in conditions such as Agenesis of the sacrum, Caudal Regression Syndrome, and VACTERL Syndrome. However, symptoms or signs characteristic of Agenesis of the sacrum and Caudal Regression Syndrome present, such as the absence of L4-L5 vertebrae, and with the combination of gestational diabetes, absence of sacrum, and cardiac defects point towards Caudal Regression Syndrome.

Final diagnosis: Caudal regression syndrome with absence of the sacrum.

DISCUSSION

Caudal regression syndrome (CRS) refers to a complex and uncommon disorder characterized by abnormal development at the lower end of the spine due to a neural tube defect.[1]

The etiology of CRS is not clearly defined; however, maternal diabetes is considered to play a crucial role, as several studies have observed single nucleotide polymorphisms in glucose-metabolizing genes such as GLUT1, HK1, and LEP in relation to malformations associated with diabetic embryopathy. Furthermore, it has been suggested that in genetic factors, Hox-mediated gene expression is the target of spinal cord anomalies related to carbon monoxide, especially with an increased risk of congenital vertebral malformations in monozygotic and dizygotic twins. Additionally, vascular hypoperfusion and certain teratogenic drugs have also been proposed as possible causal factors.[7]

This syndrome presents symptoms that can vary widely among individuals and encompasses a spectrum of diseases ranging from mild to severe or potentially life-threatening complications. Affected infants may show anomalies in the sacral and lumbar spine, with symptoms that include:

- Partial or complete agenesis of the sacrum, with possible complications such as narrowing of the hips, underdeveloped gluteal muscles, sacral dimple, and flat buttocks. [8]
- Anomalies in lumbar vertebrae.
- Additional issues in the spinal cord and lower extremities, such as neurological alterations affecting bladder and bowel control, increased urinary frequency, and other severe urological problems.
- Anomalies in the lower extremities such as joint contractures, reduced muscle mass in the legs, clubfoot, and skin folds behind the knees. [9]
- Difficulty or inability to walk without assistance, which may require the use of crutches, braces, walkers, or wheelchairs in severe cases.
- Renal anomalies such as absence or abnormal location of the kidneys, urinary obstruction, neurogenic bladder, and vesicoureteral reflux.
- Anomalies in upper vertebrae, facial features (cleft lip/palate), and conditions like anal atresia or imperforate anus. [10]
- Meningomyelocele, a defect where spinal membranes protrude through a defect in the spinal canal.
- Congenital heart defects and respiratory complications.

It is crucial to highlight that symptoms vary significantly among those affected, and parents should consult with doctors and specialists to address their child's specific case, associated symptoms, and individual prognosis.

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Prenatal ultrasound plays a fundamental role in diagnosing CRS due to its accuracy and non-invasive nature. Through 3D real-time ultrasound, it is possible to visualize the fetal spine in different planes, such as parasagittal, transverse, and coronal. [11] However, various factors can affect the reliability of prenatal ultrasound, such as fetal position, amount of amniotic fluid, and the presence of adipose tissue in the abdominal wall during pregnancy, which may lead to diagnostic errors or overlook important cases.

A study conducted by Zheng et al.,[2] found that the sensitivity, specificity, accuracy, and Youden index of prenatal ultrasound diagnosis of CRS were 92,8 %, 93,5 %, 93,4 %, and 0,86, respectively. However, in two cases, the ultrasound did not detect conus medullaris lesions, and in six cases, it did not identify anal atresia. [11] Reports indicate that prenatal magnetic resonance imaging (MRI) has greater diagnostic accuracy for spinal and cord lesions compared to prenatal ultrasound, underscoring its value as a complement to the latter.

Regarding complications, rates of incidence of hypospadias, toe anomalies, and heart anomalies during prenatal ultrasound were reported as 2/14, 7/14, and 3/14, respectively.[12] Anal atresia is a well-known complication of CRS and is classified into high and low types according to the relative position of the distal rectal pouch and the puborectal muscle. Direct indicators of anal atresia include the absence of the perianal muscle complex and lack of the target sign (a hypoechoic anal sphincter and an echogenic anal mucosa). However, in some fetuses with low atresia, a characteristic sign may be observed, complicating prenatal diagnosis via ultrasound.

Anal atresia is thought to be indirectly identifiable by the dilation of distal intestinal segments and the presence of intraluminal calcified meconium in the second and third trimesters.[13] However, it is challenging to distinguish between normal and abnormally dilated intestines in the third trimester when the rectum and colon contain stool. The presence of atypical direct and indirect signs of anal atresia contributes to the low precision of prenatal ultrasound diagnosis. To some extent, MRI offers greater accuracy in diagnosing anal atresia than ultrasound.[14]

Metabolic control of diabetes during pregnancy is a key preventive measure, highlighting the importance of maintaining appropriate HbA1c levels during gestation. Treatment is determined based on the degree and location of spinal lesions as well as the presence of anomalies in other systems. Generally, orthopedic and neurosurgical interventions are postponed until other critical conditions are resolved. For instance, in cases of children with VACTERL, such as tracheoesophageal fistula or imperforate anus, these are addressed in the first days of life. Urogenital conditions also receive priority attention to preserve renal function, prevent infections, and ensure continence. [7,12]

From a neurosurgical perspective, interventions are divided into three categories. The first includes cases requiring immediate treatment, such as newborns with large lumbosacral dural sac protrusions.[14] The second category encompasses patients who need later attention due to neurological symptoms appearing later or incidental findings such as tethered cord or sacral mass, as seen in Currarino Syndrome. The third category includes patients with symptomatic dural stenosis manifesting in late childhood or early adulthood, presenting with neurological claudication. [8]

Orthopedic management is essential in patients with adequate spinal-pelvic stability and functional hip joints, as they have potential for walking. Spinal-pelvic instability is the most relevant skeletal disorder, affecting the ability to walk and maintain balance in sitting, especially in Pang types IN and IIN. These patients are managed with supportive and comfort measures, with early spinal fusion being an option to prevent contractures and hip dislocations. [15]

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Scoliosis may arise from hemivertebrae or neuromuscular paralysis and rarely responds to the use of orthoses. Hip dislocations, caused by neuromuscular imbalances or acetabular misorientation, often require combined procedures such as open reduction, release, and tendon transposition, as well as pelvic and femoral osteotomies. Proper use of orthoses, serial casts, and soft tissue release in the hip, knee, and feet can significantly improve standing, sitting, and walking ability.[15]

This clinical case highlights the critical importance of thorough neonatal physical evaluation. Even findings that may seem minor, such as a slight depression in the sacral region or cutaneous anomalies in the lumbosacral area, can be early indicators of an underlying complex syndrome.

Early detection and continuous follow-up are essential to prevent long-term complications and improve the quality of life of affected patients. Early identification allows for timely therapeutic interventions and coordination of a multidisciplinary team to address the diverse manifestations of the syndrome.

CONCLUSIONS

Caudal regression syndrome (CRS) represents a complex and uncommon medical challenge, characterized by developmental anomalies at the lower end of the spinal column, primarily associated with neural tube defects. Although its etiology is not completely defined, a relationship with maternal diabetes, genetic factors, vascular hypoperfusion, and exposure to certain teratogenic medications is suggested. Following established guidelines from the Ministry of Public Health (MSP), it was decided to transfer the patient to a tertiary health center, where a multidisciplinary team would assume her evaluation, comprehensive treatment, and continuous follow-up with the aim of optimizing the management of her condition and improving her prognosis.

Author Contributions

EJFB: Conceptualization, Data curation, Formal analysis, Investigation, Project administration, Resources, Supervision, Visualization, Writing - original draft, Writing - review and editing.
SCVG: Data curation, Formal analysis, Methodology, Validation, Visualization, Writing - original draft, Writing - review and editing.
VARP: Data curation, Formal analysis, Investigation, Validation, Writing - original draft, Writing - review and editing.

Requirements for the authors of case report submissions

The authors state that there are no conflicts of interest.

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