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**Case Report**

# Prenatal diagnosis of caudal regression syndrome in a non-diabetic mother: a rare case report

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## ABSTRACT

Caudal regression syndrome (CRS) is a rare congenital malformation characterized by partial or complete agenesis of the sacrum and variable involvement of lumbar spine and lower limbs. It is classically associated with maternal diabetes, though sporadic cases in non-diabetic mothers have been reported. We report a case of a 25-year-old gravida 2 para 1 live 1 woman with previous lower segment caesarean section (LSCS) three years back, non-diabetic, who was diagnosed prenatally at 20 weeks of gestation with CRS on level II ultrasonography. There was a significant family history of type 2 diabetes mellitus in the patient's mother, along with a history of three intrauterine fetal deaths (IUFDs). The fetus showed abrupt termination of the spine at the lumbosacral level with an absent sacrum and hypoplastic lower limbs. This case highlights that CRS can occur even in the absence of maternal diabetes, emphasizing the importance of detailed anomaly scanning and consideration of genetic and familial factors.

**Keywords:** Caudal regression syndrome, Prenatal diagnosis, Non-diabetic mother, Fetal anomaly, Ultrasonography

## INTRODUCTION

Caudal regression syndrome (CRS) is a rare congenital malformation with an estimated incidence of approximately 1-2 per 100,000 live births.[1] CRS also referred to as caudal dysgenesis syndrome (CDS), results from abnormal development of the caudal region of the embryo. The condition exhibits a broad phenotypic spectrum and may affect the gastrointestinal, genitourinary, musculoskeletal, and neural systems.[2] CRS is a rare condition, and its exact pathogenesis remains uncertain. Proposed etiological factors include maternal diabetes, genetic susceptibility, and impaired vascular perfusion; however, a definitive cause has not been established. Advances in prenatal imaging facilitate early detection, while meticulous postnatal evaluation is necessary to delineate the full extent of anomalies. The degree of organ involvement correlates with disease severity. Abnormalities are primarily structural, and varying combinations of respiratory, cardiac, gastrointestinal, genitourinary, orthopedic, and neurological impairments may occur. Optimal care requires coordinated input from multiple specialties. As the underlying defect cannot be reversed, management is largely supportive and aimed at improving function and quality of life.[3]

In 1993, Pang described a classification system that divides the condition into five categories based on anatomical severity. Type I involves either a partial or complete absence of the sacrum. Type II is characterized by complete sacral agenesis accompanied by varying degrees of lumbar vertebral deficiency, with preservation of the connection between the ilia and the lower vertebral column. Type III represents a complete absence of the lumbosacral spine, where the terminal vertebral plate articulates with the iliac bones through an amphiarthrodial joint. Type IV is marked by failure of normal separation of the caudal soft tissues. Type V, the most severe form, is

characterized by fusion of the lower limbs with a single central femur and tibia, a condition known as sirenomelia (mermaid syndrome).[4]

Prenatal diagnosis is usually achieved during the second-trimester anomaly scan, allowing early counseling and decision-making.

## CASE REPORT

A 25-year-old woman, gravida 2 para 1 live 1, presented for routine antenatal care at our tertiary care center. She had a history of one previous LSCS done three years back for non-reassuring fetal heart rate. The patient was non-diabetic, normotensive, and had no history of drug intake, fever, or infections during early pregnancy.

There was a significant family history of type 2 diabetes mellitus in the patient's mother. Additionally, the patient's mother had a suspicious history of three IUFDs, though medical records were unavailable. There was no history of consanguinity.

First-trimester screening was unremarkable. A detailed level II anomaly scan performed at 20 weeks of gestation revealed: Abrupt termination of the fetal spine at the lumbosacral level, absence of sacral vertebrae, hypoplastic and malpositioned lower limbs, normal cranial anatomy and upper limbs and no obvious abdominal wall or cardiac anomalies

Amniotic fluid volume was within normal limits. Based on these findings, a diagnosis of CRS was made.

The patient and her family were counseled regarding the diagnosis, prognosis, and possible postnatal complications including neurogenic bladder, bowel dysfunction, and impaired mobility. After informed counseling, the couple opted for medical termination of pregnancy as per the medical termination of pregnancy (MTP) Act.

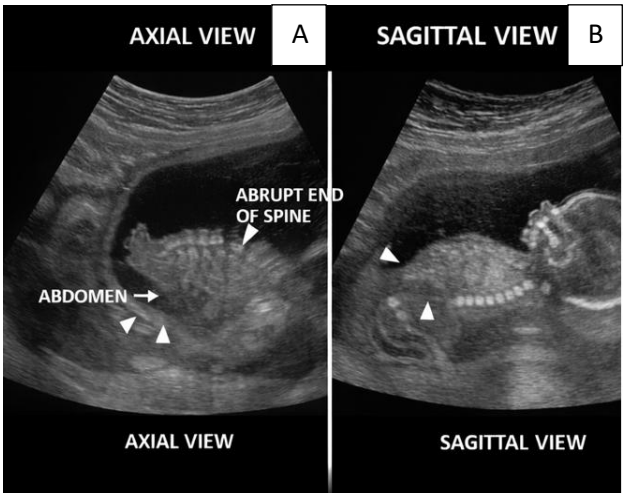


**Figure 1 (A and B): Ultrasound of a 20-week fetus with CRS.**

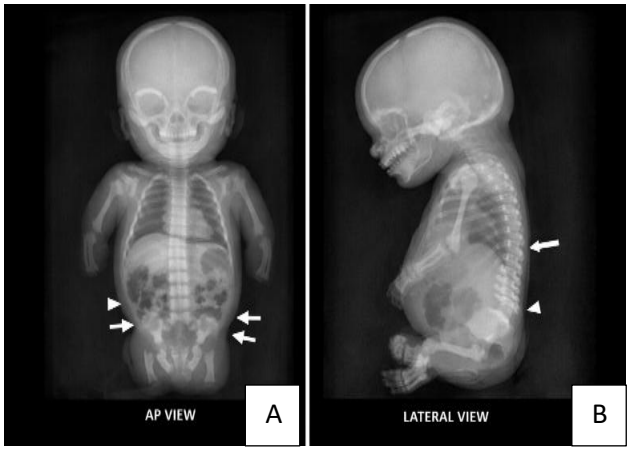


**Figure 2 (A and B): X-ray of a 20-week abortus with CRS.**

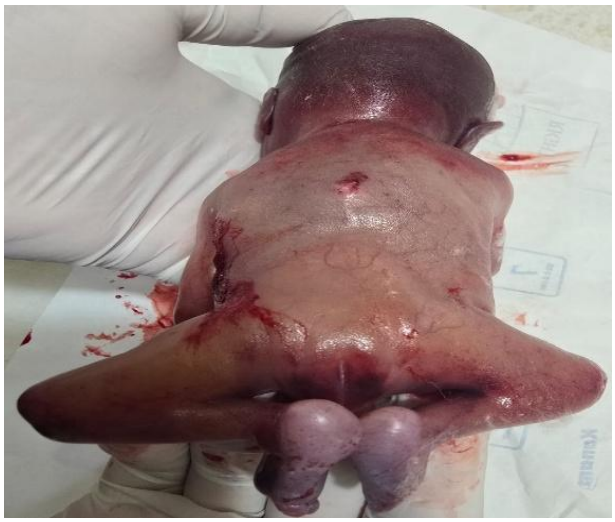

**Figure 3: Photograph of abortus showing regression of sacral spine.**

## DISCUSSION

CRS, also known as caudal dysgenesis or caudal dysplasia sequence, comprises a diverse group of congenital anomalies caused by abnormal development of the lower spine, spinal cord, and surrounding mesoderm-derived tissues. The condition often involves multiple organ systems, commonly affecting the genitourinary, gastrointestinal, musculoskeletal, and neurological systems. Recent literature continues to cite a population incidence in the range of ~1-2 per 100,000 live births, underscoring its rarity and the likelihood that many clinicians encounter only isolated cases during practice.[1]

Although maternal pregestational diabetes remains the most consistently reported association, CRS is increasingly recognized in pregnancies without overt diabetes, suggesting a multifactorial pathobiology with contributions from genetic susceptibility and environmental/teratogenic influences.[5] In the present case, the mother was non-diabetic, but a family history of type

2 diabetes was present-an observation that supports the concept that CRS can occur outside classical maternal diabetes and may reflect shared metabolic/genetic vulnerability rather than a single deterministic cause.[5,6] Several mechanisms have been suggested to explain the development of CRS. These include abnormalities in secondary neurulation and impaired formation of the caudal mesoderm, disturbances in key embryonic signaling pathways-particularly those related to retinoic acid regulation-and reduced blood supply to the caudal part of the embryo due to a "vascular steal" phenomenon. Despite these proposed explanations, a single definitive mechanism capable of accounting for the entire spectrum of CRS has not yet been established.[7]

***Prenatal diagnosis and phenotypic spectrum***

A key strength of this report is antenatal detection at mid-gestation on targeted ultrasound. Prenatal sonographic suspicion is typically raised by abnormal lumbosacral anatomy, shortened crown-rump length for gestation, abnormal lower-limb posture/movement, and associated findings such as oligohydramnios when severe renal anomalies coexist. When ultrasound visualization is limited or when delineation of the spinal cord and soft-tissue anomalies is required, fetal MRI provides complementary information and improves anatomic characterization and counseling accuracy.[6] This is clinically relevant because CRS spans mild sacral hypoplasia with near-normal limb function to severe lumbosacral agenesis and, at the extreme end, sirenomelia.[7,8]

***Prognosis, survival, and counselling***

Outcome in CRS is primarily determined by (1) the level/extent of spinal and sacral involvement and (2) the presence of life-limiting associated anomalies, particularly severe renal/urinary tract malformations, major cardiac defects, and pulmonary hypoplasia secondary to early oligohydramnios.[3,7] In general, isolated or milder forms can be compatible with long-term survival, while severe CRS and sirenomelia carry a markedly worse prognosis; for example, published sirenomelia cases commonly report death shortly after birth due to associated visceral anomalies, illustrating the lethality of the most severe caudal dysgenesis phenotypes.[9]

Importantly, even among survivors, long-term morbidity is substantial and centers on mobility and continence. In a recent patient-reported outcome study of radiologically confirmed sacral agenesis, ~70% had impaired/absent bladder control, and ~80% required walking aids, emphasizing that neuro-urologic dysfunction and orthopedic disability often dominate long-term care needs and quality-of-life discussions.[10] These contemporary data are useful for counseling families following antenatal diagnosis, particularly when termination decisions, delivery planning, and anticipatory postnatal care are being considered.

***Management implications for the present case***

Because CRS is a structural developmental disorder, management is supportive and individualized, requiring a multidisciplinary approach (maternal–fetal medicine, neonatology, pediatric urology, orthopedics, neurosurgery, rehabilitation, and, when indicated, pediatric surgery).[3,7] In our case, early prenatal recognition allowed timely counseling regarding spectrum severity, likely multisystem involvement, and prognosis. Postnatal evaluation (or fetal assessment where feasible) should focus on defining the anatomic extent of spinal/sacral defects, screening for renal/urinary tract and anorectal anomalies, and documenting limb/hip deformities-elements that guide both immediate neonatal priorities and long-term functional planning.[7,10]

## CONCLUSION

CRS can occur in pregnancies without maternal diabetes. A high index of suspicion, meticulous anomaly scanning, and detailed family history are crucial for early diagnosis. This case underscores the need to consider CRS even in non-diabetic pregnancies and highlights the role of prenatal ultrasonography in detecting rare congenital anomalies.

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