Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Caudal Regression Syndrome: A Case Report

M. Raboua[1*], M. Mekouar[1], H. C. Ahmanna[1], I. Zouita[1], D. Basraoui[1], H. Jalal[1]

[1]Radiology Department Mother and Child Hospital-CHU Mohammed VI University Cadi Ayad Marrakech, Morocco

DOI: 10.36347/sjmcr.2024.v12i05.071 | **Received:** 16.04.2024 | **Accepted:** 20.05.2024 | **Published:** 23.05.2024

***Corresponding author:** M. Raboua
Radiology Department Mother and Child Hospital-CHU Mohammed VI University Cadi Ayad Marrakech, Morocco

Abstract **Case Report**

Caudal regression syndrome is a rare malformation syndrome that involves varying degrees of agenesis of the sacral and coccygeal vertebrae, along with shortening of the lower limbs, and gastrointestinal, genitourinary, and cardiovascular anomalies. Its precise cause remains unknown, although its association with maternal diabetes is well established. We present a rare case of this syndrome in a one-month-old newborn whose mother has known diabetes and who also presents with anorectal malformation syndrome.

Keywords: Caudal regression syndrome, sacrococcygeal and lumbar vertebral agenesis.

INTRODUCTION

Caudal regression syndrome is a rare malformation syndrome that involves varying degrees of agenesis of the sacral and coccygeal vertebrae, along with femur shortening, and gastrointestinal, genitourinary, and cardiovascular anomalies. Its incidence is 1 to 5 cases per 100,000 births. Its precise cause has not yet been identified, but its association with maternal diabetes is well established [1]. We present a rare case of this syndrome in a one-month-old newborn whose mother has known diabetes and who also presents with anorectal malformation syndrome.

PATIENT AND OBSERVATION

This is a one-month-old male infant, born from a poorly monitored pregnancy, admitted with anorectal malformation. There is no history of consanguinity or malformations in the siblings. However, there is a history of type II diabetes in the mother, which is poorly controlled. Clinical examination reveals an infant in good general condition, with malformations of the lower half of the body including thigh hypotrophy with hip abduction and irreducible knee flexion, resulting in a "frog-like" appearance of the lower body associated with a single orifice anorectal malformation. A standard X-ray of the pelvis and lower limbs (Figure 1A) revealed a funnel-shaped pelvis with femur shortening, thigh abduction, and knee flexion, resulting in the "frog-like" position of the lower limbs. A lateral X-ray of the dorsolumbosacral spine (Figure 1) showed sacrococcygeal and lumbar vertebral agenesis without other abnormalities of the spine segment above the agenesis. Given the results of the clinical examination and standard X-ray, the diagnosis of caudal regression syndrome was made. An abdominal ultrasound, performed as part of the malformation assessment, revealed a sigmoid kidney without other associated malformations. A spinal MRI was performed, showing lumbar and sacrococcygeal agenesis resembling a shield, high insertion of the dorsal medullary cord at the level of D11 which is blind at its distal part with an underlying oblong cystic formation below the dorsal medullary cord with T1 hyposignal, T2 hypersignal, without detectable osseous wall defect.

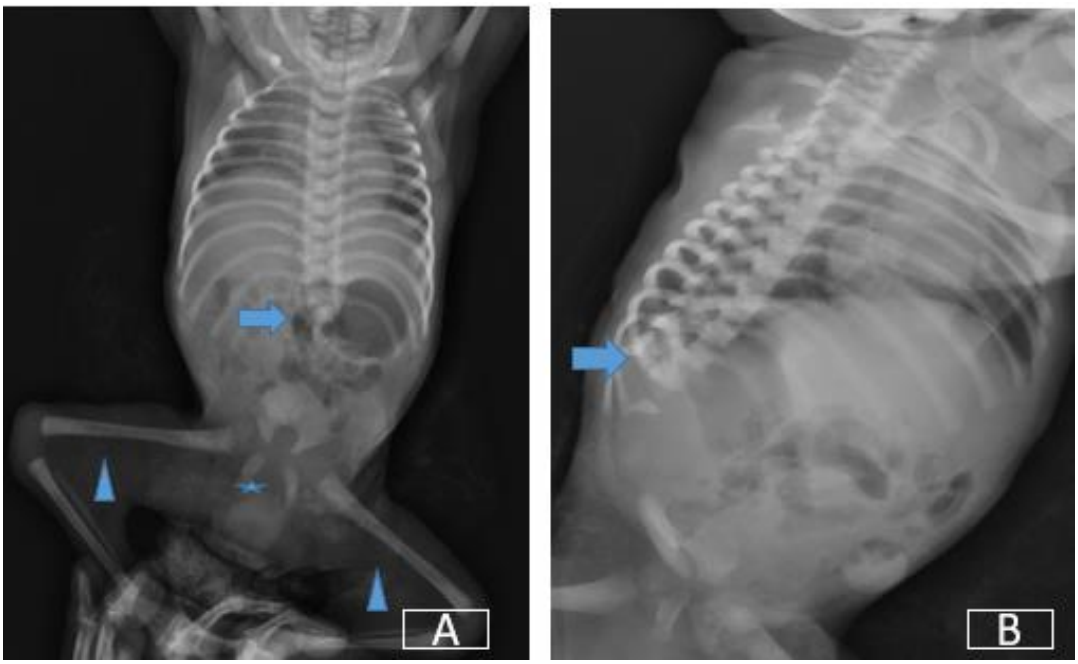

Figure 1: Standard X-ray of the pelvis and lower limbs (A) and lateral X-ray of the dorsolumbosacral spine (B): revealed a funnel-shaped pelvis (asterix) with femur shortening, thigh abduction, and knee flexion, resulting in the "frog-like" position of the lower limbs (arrow head) with sacrococcygeal and lumbar vertebral agenesis (arrow)

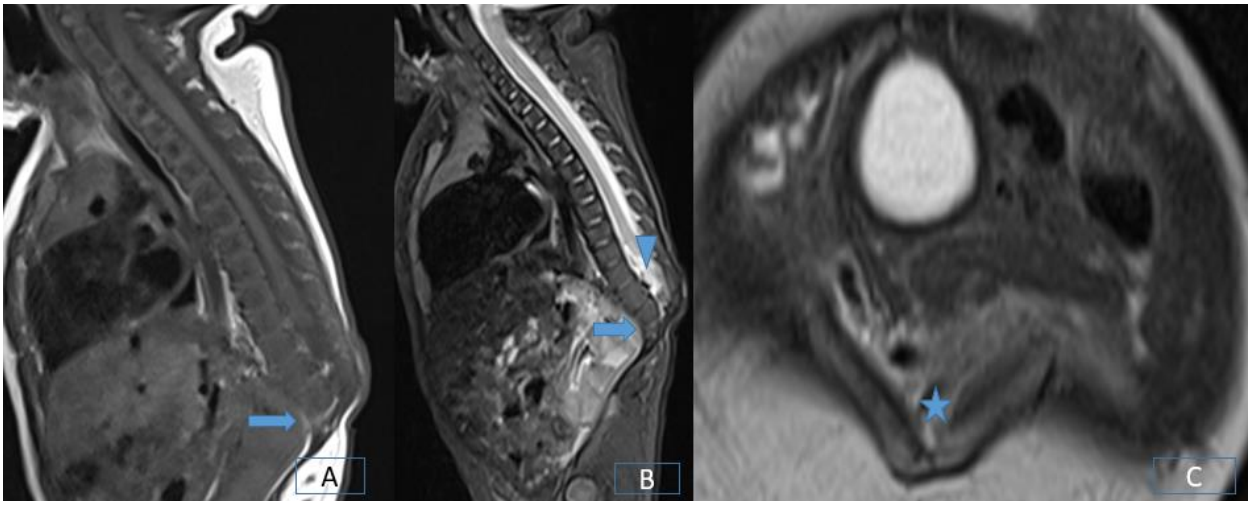

Figure 2: Sagittal MRI sections: T1 (A) and T2 (B), and axial T2 (C): showing lumbar (arrow) and sacrococcygeal (asterix) agenesis resembling a shield, high insertion of the dorsal medullary cord at the level of D11 (arrow head) which is blind at its distal part with an underlying oblong cystic formation below the dorsal medullary cord with T1 hyposignal, T2 hypersignal, without detectable osseous wall defect

DISCUSSION

Caudal regression syndrome is a rare malformation syndrome that involves varying degrees of agenesis of the sacral and coccygeal vertebrae, along with femur shortening, and gastrointestinal, genitourinary, and cardiovascular anomalies. The etiopathogenesis of this anomaly is not straightforward; however, its association with maternal diabetes is well established. Prenatal diagnosis relies on morphological ultrasound by determining the cranio-caudal length and femoral length. Imaging is crucial for diagnosis but also for therapeutic decision-making [2]. Sacrococcygeal agenesis is often associated with agenesis of the lumbar vertebrae as well as anomalies of the medullary cone better explored by MRI [3]. Gastrointestinal anomalies such as anal imperforation, anorectal atresia, esophageal or duodenal atresia should be sought. Urological anomalies such as agenesis, dysplasia, or ectopy are prone to complications, particularly recurrent urinary infections that can sometimes affect functional and vital prognosis [4]. In our case, this syndrome was associated with a common anorectal malformation.

CONCLUSION

Caudal regression syndrome encompasses a range of orthopedic, gastrointestinal, genitourinary, and

neurological congenital malformations. Its close association with maternal diabetes is well established, highlighting the importance of early prenatal diagnosis for appropriate management.

REFERENCES

1. Van der Knaap, M. S., & Valk, J. (1988). Classification of congenital abnormalities of the CNS. *American journal of neuroradiology*, *9*(2), 315-326.
2. González-Quintero, V. H., Tolaymat, L., Martin, D., Romaguera, R. L., Rodríguez, M. M., & Izquierdo, L. A. (2002). Sonographic diagnosis of caudal regression in the first trimester of pregnancy. *Journal of ultrasound in medicine*, *21*(10), 1175-1178.
3. Sharma, P., Kumar, S., & Jaiswal, A. (2013). Clinico-radiologic findings in Group II caudal regression syndrome. *Journal of Clinical Imaging Science*, *3*, 26.
4. Barkovich, A. J., Raghavan, N., Chuang, S., & Peck, W. W. (1989). The wedge-shaped cord terminus: a radiographic sign of caudal regression. *American journal of neuroradiology*, *10*(6), 1223-1231.
5. Belkacem, S., Chellaoui, M., & Dafiri, R. (2010). Le syndrome de Currarino. *Feuillets de radiologie*, *50*(3), 115-120.