A Rare Case of Caudal Regression Syndrome linked to Tethered Cord and Dermal Cysts

Claudia Marcela Restrepo, Erik Muñoz, Juan David Cano, Andres Garay, Andreina Martinez Amado

ABSTRACT

Caudal regression syndrome (CRS) is a rare congenital disorder characterized by agenesis of the vertebral bodies of the lumbosacral spine associated with other malformations of the pelvis and inferior limbs. We present a case of a 18 months boy referred to Central Military Hospital (Bogotá, Colombia) with sacrococcygeal fistula and a permanent hip abduction brace. On physical examination, there was an abnormal palpation of the sacral hiatus and coccyx. His hips were flexed and abducted, but did not have contractures. Neurological examination and psychomotor development were normal. In lumbar MRI, there were found hypoplasia of the sacrum and agenesis of the coccyx with a large subcutaneous and spinal lipoma, tethered spinal cord, and two dermal tracts at the level of L4 and S3 vertebrae. Somatosensory evoked potentials with latency and amplitude within normal ranges. Because of this, operation was not considered.

Keywords: Caudal regression syndrome, Magnetic resonance imaging, Tethered cord, Spinal lipoma.

How to cite this article: Restrepo CM, Muñoz E, Cano JD, Garay A, Amado AM. A Rare Case of Caudal Regression Syndrome linked to tethered Cord and Dermal Cysts. J Spinal Surg 2014;1(3):132-134.

Source of support: Nil

Conflict of interest: None

INTRODUCTION

Caudal regression syndrome (CRS), also known as caudal dysplasia and sacral agenesis syndrome, is a rare congenital disorder characterized by varying degrees of vertebral abnormalities with an incidence of 1 in 7500 normal pregnancies and may affect the lower extremities, the lumbar and coccygeal vertebrae.1 Caudal regression syndrome can be accompanied by multiple congenital anomalies involving multiple systems, such as renal ectopia, imperforate anus, anorectal atresia, tethered-cord, diastematomyelia, lipomyelomeningosel and congenital narrow spinal tract, scoliosis, hip dislocation and contracture, narrow pelvis, syringomyelia, club foot and frog leg.1,2

Although the exact teratogenic mechanism is not known, possible risk factors include hyperglycemia, genetic factors and vascular hypoperfusion.2 In this study, we presented a case of caudal regression syndrome with tethered cord accompanied by dermal cysts with no infection signs. The patient had not showed neurological compromise; for that reason, he was not considered for neurosurgical intervention.

CASE REPORT

We present a case of a 18 months old boy who was brought by his mother to the clinics service of the Central Military Hospital (Bogotá, Colombia). He was product of first gestation of 24-year-old women. There were no history of radiation exposure drug use and serious disease for the mother, for whom the blood glucose had been normal during pregnancy. The mother had a report and ultrasound finding at 30.5 weeks of gestation to be of normal fetal growth, normal amniotic fluid and mild to moderate bilateral pyelocaliectasis. On the first day postpartum, the physical examination revealed a gluteal sacrococcygeal fistula and a paravertebral fistula without signs of inflammation or infection. The patient´s neurological examination and psychomotor development were normal.

Complementary studies were performed when the child was 5 months. There was a renal ultrasound which revealed left renal hydronephrosis and grade IV vesico-ureteral reflux (VUR). On physical examination, there was an abnormal palpation of the sacral hiatus and coccyx. For this finding, a hip plain film was performed and it showed a left acetabular angle of 30º and a permanent hip abduction brace was adopted in order to encourage normal development of the joint (Figs 1A and B). Nevertheless, at this age, he continued to be asymptomatic and only had purulent discharge through both fistulas.

Due to the abnormal physical examination and renal ultrasound findings, a lumbosacral magnetic resonance imaging (MRI), cystogram and a DMSA renal scan (DM-SA-RS) were performed. Cystogram reported a bladder with adequate capacity and posteroinferior sacular dilatation corresponding to a diverticulum with grade II-III left VUR without reporting post-voiding residue.
DMSA-RS demonstrated decreased uptake in left inferior renal pole suggestive of left pyelonephritis. Lumbosacral MRI showed hypoplasia of the sacrum and agenesis of the coccyx with a large subcutaneous and spinal lipoma, tethered spinal cord, and two dermal tracts at the level of L4 and S3 vertebrae (Figs 2A and B).

The child is currently 1 year old and was diagnosed with caudal regression syndrome with tethered spinal cord. To gather information regarding the severity and the extent of tethering, there was need of perform somatosensory evoked potential test which did not demonstrate any abnormality (Fig. 3). At this age, no operation was planned because the patient remained asymptomatic and no other pathological signs were found neither neurological abnormal findings.

**DISCUSSION**

The patient with CRS described in this case report is of interest because of the multiple associated abnormalities connected to the vertebral anomalies in a child born of a nondiabetic mother and the presence of dermal cysts and spinal lipoma which together are very rare presented in literature. Also, the incidence of dermal cyst in all the intraspinal tumors is very uncommon (a incidence of 0, 5-2% from all the intraspinal tumors). It is well known that hyperglycemia is the most common risk factor linked to this kind of congenital dysfunction. Between the causes of this condition there has been some genetical factors; one of them is the 18 p11.2 deletion; however, the exactly cause remains unclear. However, this case shows the need to investigate more deeply into the pathophysiological mechanisms that result in the development of caudal regression.
Regarding the tethered spinal cord and caudal regression syndrome, a study in 1994 reported an incidence of 4 in 19 cases. This means that the tethered cord injury is a common complication of this disease. In this case, this injury is an additional trigger for symptoms in the legs and urinary system.³

In the evaluation and follow-up of a patient born with CRS, there is the need of urologic assessment, neurological evaluation and orthopedics consultations because of the multiple involvements of different systems. The first steps on neurologic assessment consist on plain X-ray films of lumbosacral spine. It is also necessary to perform a MRI of lumbosacral spine in order to visualize the vertebrae compromise and spinal cord defects. There is also important to perform electrophysiological studies in order to assess the integrity of nerves and spinal cord. Surgery is indicated in cases where there is evidence of neurological compromise and delayed psychomotor development.⁴

CONCLUSION

Caudal regression is a rare genetic syndrome appearing in 1:7 RN, without differences by sex characterized by agenesis of the vertebral bodies of the lumbosacral spine associated with other malformations of the pelvis and inferior limbs. The case that we present in this occasion evidence a patient diagnosed with this syndrome at 12 months old, who also has many associated pathologies: left renal hydronephrosis and grade IV VUR, a large subcutaneous and spinal lipoma, tethered cord and two dermal tracts at the level of L4 and S3 vertebral with this in mind draws attention that the patient had no delay in its physical or neurological development. Which demonstrates the ability of the central nervous system to adapt to the malformations that could prevent the path of the nerve endings.

Currently, there is no known clear cause for this syndrome but it is believed to have a genetic component important in addition to being associated with maternal pathologies present during pregnancy, such as diabetes and hypertension.⁵ However is not the case of our patient, because the mother did not provide any of these pathologies during pregnancy and attended controls up to the birth, which represents a call to attention for healthcare professionals, to take into account this type of syndrome even in children of mothers young and healthy.

Finally, it is important to keep in mind that these patients require medical care of multiple specialty to ensure the best possible quality of life as well as keep a strict monitoring throughout his life to prevent possible complications, such as occurs in our patient who continues to assist controls in the Central Military Hospital (Bogotá, Colombia) every 6 months.

REFERENCES