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Case Report

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Dorsolumbosacral agenesis: Case report and literature review

Abstract

Objetives: To report a case of dorsolumbosacral agenesis and to make a systematic review of the literature focused on prenatal diagnosis.

Materials and methods: We report a case of a 32year old pregnant woman, with a 30 week pregnanacy, without prenatal care, the fetus is diagnosed with dorsolumbosacral agenesis. The mother request voluntary termination of pregnancy. A systematic review of the literature focused on prenatal diagnosis of thos condiction is performed.

Results: We found 50 papers, 6 met the inclusión critiria. Three of them with prenatal diagnosis. In the first case the diagnosis was made at 13 weeks of gestation and termination of preganancy was requested. In the second case corresponded an biamniotic bicorial twin preganancy. One normal feto and one presented dorsolumbosacral agenesis. The diagnosis was made in the second trimester. The pregnancy continued until 34 week of gestation and the affected neonate had perinatal death. The third case, the diagnosis was made at 18 weeks and a male newborn of 2990gr was born at 37 weeks of gestation.

Conclusion: Dorsolumbosacral agenesis is a very severe form of caudal regresión syndrome, with only a few cases reportted in the literature. To the best of our knowladge this is the fourth case reported with prenatal diagnosis.

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Introduction

Dorsolumbosacral agenesis is the most severe form of cranio-caudal regression syndrome.[1] Caudal regression syndrome as a low incidence, reported 1 in 10,000 births[2] and lumbosacral dysgenesis has only a few cases reported in the literature.

Craniocaudal regression syndrome is a clinical syndrome characterized by malformations of the spine, spinal cord, and lower extremities.[2] Dorsolumbosacral agenesis is characterized by agenesis of the sacrum, lumbar vertebrae and one or more thoracic vertebrae.[1,3]

The etiology of craniocaudal regression is not well clarified, it is known that an alteration in gastrulation occurs before 28 days of gestation.[4] Genes such as HLXB9 CYP26A1, F186L and C358R involved in retinoic acid homeostasis, have been studied in the pathophysiology of this condition without conclusive data.[5,2]

The main related risk factor is diabetes, which increases the risk 200 times, but drugs such as lithium, sulfonamides or agents such as radiation and extreme temperatures have been also linked.[5]

The diagnosis can be made prenatally by ultrasound, and there are reports of cases reported from the first trimester,[6] complementary techniques such as magnetic resonance are very useful to confirm the diagnosis.[7,8]

The prognosis will depend on the extent of the neurological lesion and the associated malformations. It is common to find alterations of the musculoskeletal system, as well as gastrointestinal and urinary.[5,2] Intelligence is generally normal, but they require long-term neurological, renal, and orthopedic management. In some cases, after orthopedic treatment, ambulation of patients is achieved.[4]

Dorsolumbosacral agenesis is a pathology that is little described in the literature, which makes prenatal counseling difficult. Our objective is to make a case report and a review of the literature review focused on the prenatal diagnosis of dorsolumbosacral agenesis.

Case report

In August 2019, a 32-year-old woman, migrant from Venezuela, who has livid in Colombia for 1year, without health care insurance. She went to the emergency unit of Hospital La Victoria; third-level hospital in the city of Bogotá.

She had pelvic pain, with a gestation of 28.1 weeks. Medical history: type 2 diabetes mellitus. Diagnosed 3years ago under management with glibenclamide, but she discontinued it 28 months ago, currently without any medical management.

Gynecological history: 4 pregnancies, 2 deliveries, 2 caesarean section, an 1 abortions. Caesarean section for cephalopelvic disproportion 15years ago and iterative caesarean section 9years ago. Early abortion 1year ago. Current pregnancy was planned an desired; primipaternity. No prenatal care.

On physical examination: Heart rate: 80bpm, respiratory rate 20rpm, blood pressure 110/70mmHg, uterine height: 30cm, fetal heart rate 133bpm, rest of the physical examination within normal limits.

Paraclinical tests: Hemogram: hemoglobin 10.4mg/dl, hematocrit 30. Preprandial 153 and postprandial glycemia 213. Creatinine 0.38mg/dl. Hemoclassification O+. Normal transaminases. Positive treponemal test. Partial urine: normal. HIV negative. Hepatitis B: negative.

Obstetric ultrasound: single fetus breech presentation. In the anatomical evaluation: agenesis of the last dorsal vertebra, of all the lumbar vertebrae, of the sacrum and of the coccyx (Figure 1). Heart disease: balanced complete atrioventricular canal and bilateral clubfoot. Polyhydramnios: larger pocket 9.3cm. Posterior placenta with normal characteristics.

She was hopsitalized to reach metabolic control, benzathine penicillin 24-00-00U, and regimen of NPH insulin and crystalline insulin was started, witht a total dose of 0.6U/kg/day. After three days of hospitalization, metabolic control was achieved.

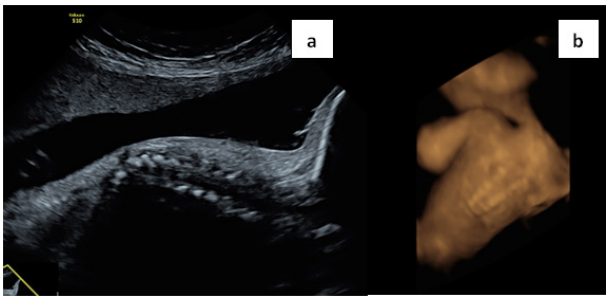

Figure 1 Ultrasound week 30. a) Sagittal section of the column in 2D b) 3D image.

Patient requests voluntary termination of pregnancy, is assessed by social work, psychology and gynecology group for termination of pregnancy. Informed consent is signed. A fetal asystole induction procedure is performed with the application of 10cc of intracardiac potassium chloride. Procedure without any cmoplication. Due to a history of caesarean section and presentation of the pelvis, caesarean section and pomeroy were performed. A female fetus of 1500g, and 28cm height. X-ray is taken (Figure 2). Family does not authorize autopsy.

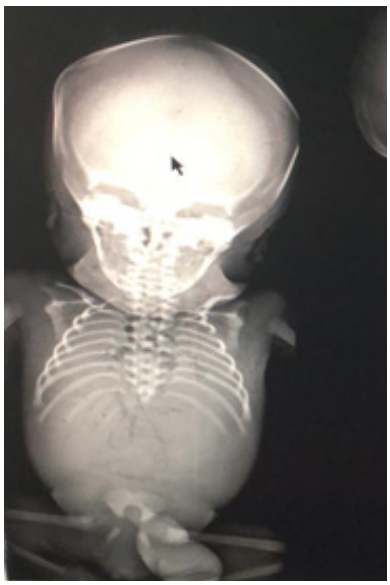

Figure 2 Postmortem radiography. Agenesis of the last thoracic vertebra and lumbosacral vertebrae.

The evolution of the puerperium is satisfactory, glucometry in goals, she was dischraged from hospital, after two days.

Ethical aspects: Authorization was requested from the patient with written informed consent, precautions were taken to guarantee the confidentiality of the information, the anonymity of the patient; the photographic record was taken by the authors.

Materials and methods

In order to answer the question of how many cases of dorsolumbosacral agenesis have been diagnosed prenatally, a systematic review of the literature was carried out, with the following mesh terms: "dorsolumbosacral agenesis", "prenatal diagnosis", "caudal regression syndrome". The following databases were searched: Medline via pubmed, and LILIACS. The search was limited to articles published in English or Spanish. The search was limited to the last 30years. The search was completed in the following databases: OVID, upto date and in academic google. a snowball technique was performed by searching the references of the articles found. The three investigators carried out the search independently and at the end a reconciliation was made between all authors.

Results

50 related articles were found, of these five fulfilled the inclusion criteria. 5 articles on dorsolumbosacral agenesis (Table 1). One of the articles reported two cases for a total of 6 reported cases. In two cases the diagnosis was made prenatally. In both cases, associated malformations were found (atrioventricular canal and myeloschisis).

In the first case, Mihmanli reports a 3-month-old newborn, the last vertebra visualized in the MRI was T9. Patient from a rural area of Turkey, who is diagnosed and an orthopedic management is proposed, but the family did not accept; follow up was not possible.[9]

The second case, Nagy, reports a 37-year-old pregnant woman with type 2 diabetes mellitus. Prenatal diagnosis was made by ultrasound at 21 weeks of gestation. The last vertebra visualized was T5, in addition, ventricular communication and a single umbilical artery were found. The patient requested voluntary termination of pregnancy.[3]

The third case, Loque reports, a 34-year-old pregnant woman, with type 2 diabetes mellitus, diagnosis made by ultrasound at week 18. Agenesis of the last 4 dorsal vertebrae and the lumbosacral spine was documented. A 37-week-old newborn weighing 2990 grams was obtained. With 13-month follow-up in rehabilitation management.[10]

Table 1 Cases of dorsolumbosacral agenesis

Author	Country	Time of diagnosis	Weeks of gestation	Observations
Mihmanli[9]	Turkey	Newborn of 3 months.	Does not apply.	No follow-up. lost after diagnosis.
Nagy[3]	Hungary	Prenatal	21weeks.	Voluntary termination of pregnancy
Luque[10]	Chile	Prenatal	18 weeks.	2900g newborn. 13-month follow-up.
Bosemani[1]	USA	Prenatal	13 weeks	Early perinatal death.
Szumera[11]	Poland	6 years of life	Does not apply.	Stabilization surgery was performed
		8 years of life	Does not apply.	Stabilization surgery was performed

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The fourth case, Bosemani reported a 40-year-old pregnant woman, with a history of thyroid cancer treated surgically, without a history of diabetes. At week 13, a diagnosis of biamniotic bicorial multiple gestation with lumbosacral agenesis of fetus A and dorsal myelomennigocele was made. fetus B normal. In week 21, an MRI was performed in week 21 evidence of agenesis of the last thoracic vertebrae, lumbar vertebrae and sacrum. In addition to thoracic myelomengocele, Chiari II malformation and bilateral clubfoot. Termination of the pregnancy of fetus A is proposed. The patient decides to continue the pregnancy, at week 33 she presents rupture of membranes. The neonate with the malformations dies 6 days after birth, the healthy neonate has a satisfactory evolution.[1]

Szumera,[11] reports two pediatric cases. The first case a 6-year-old girl with agenesis of the last dorsal vertebra, the lumbar vertebrae and the sacrum, the second an 8-year-old boy with agenesis of the last three dorsal vertebrae, lumbar vertebrae and sacrum. The two cases underwent a successful spinal fixation orthopedic procedure for stabilization.

Conclusion

Dorsolumbosacral agenesis is an extreme presentation of the craniocaudal regression syndrome, with a very low incidence. Its diagnosis can be made prenatally by ultrasound, complementary tests such as resonance can be useful.

It is important for the clinical practice to understand this pathology for better prenatal counseling to parents and more informed decision-making.

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Author's contribution

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Conflicts of interest

The author declares that there is no conflict of interest regarding this study.

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