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Caudal Regression Syndrome

Síndrome de Regresión Caudal

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ABSTRACT

Background: Caudal Regression Syndrome is an infrequent congenital malformation of the vertebral spine's lower segments characterized by a truncated medullar cone and aplasia or hypoplasia of the sacrum. Specifically, sacral agenesis

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represents an incidence of 0.01 to 0.05 of every newborn. Among the main risk factors are elevated glycosylated hemoglobin, hormonal imbalance, and lipid protein metabolic disorder. A correlation has been established between anomalous pregnancies and congenital malformations in newborns, with a prevalence of 5 to 10% in children of diabetic mothers, with an incidence of 1 per 350 cases, representing a 200% rise over the general population. Objective: To report an extremely uncommon epidemiologically rare case of Caudal Regression Syndrome, characterized by caudal musculoskeletal involvement. **Clinical case:** a 27-year-old female in her 34th week of gestation during her second pregnancy presented to the high specialty, high-risk pregnancy consultation with a background of glycemia of 205mg/dl, glycosylated hemoglobin percentage (%HbA1c) of 10.1%, and estimated average glucose levels of 243mg/dl. The patient arrived at the emergency room during active labor with premature membrane rupture, both deciding factors for pregnancy termination. The outcome was a sole female live product with a vertebral spine interrupted approximately by T10, non-palpable iliac crests, sacrum, and coccyx, and shortening of hypotrophied inferior extremities. **Conclusion:** successful prevention in the early diagnosis of malformations during pregnancy requires strict prenatal control for any pregnant woman with metabolic risk factors. The first prenatal consultation presents an opportunity to perform diabetic screening. If diagnosed, follow-up procedures such as ultrasounds at a second level of medical care should be performed for the timely detection of congenital malformations associated with high levels of glycemia.

Keywords: Caudal Regression Syndrome, Caudal Agenesis, Sacral Regression.

RESUMEN

Antecedentes: el Síndrome de Regresión Caudal es una malformación congénita poco frecuente de los segmentos inferiores de la columna caracterizada por un cono medular truncado y aplasia o hipoplasia del sacro. Concretamente en la agenesia sacra se presenta una incidencia entre 0.01 y 0.05 por cada 1000 nacidos vivos. Dentro de los factores de riesgo, principalmente están los niveles elevados de

hemoglobina glicosilada, desbalances hormonales y desorden metabólico lipídico y proteico, se ha relacionado embarazos anómalos con malformaciones congénitas en bebés en un 5 al 10 %, en hijos de madres diabéticas, con una incidencia de 1 caso por cada 350, con un aumento de 200% sobre la población general**. Objetivo:** reportar un caso extremadamente infrecuente epidemiológicamente de Síndrome de Regresión Caudal, caracterizado por el compromiso musculoesquelético caudal. **Caso clínico:** femenina de 27 años de edad, secundigesta con embarazo de 34 semanas de gestación atendida en consulta de especialidad por embarazo de alto riesgo, con antecedente de glicemia de 205 mg/dl, porcentaje de hemoglobina glicosilada (%HbA1c) 10.1%, glucosa promedio estimada 243 mg/dl. Paciente que ingresa al Servicio de Urgencias con trabajo de parto en fase activa y ruptura prematura de membrana, por lo que se decide interrupción del embarazo. Se obtiene producto femenino único vivo, con columna vertebral interrumpida en T10 aproximadamente, no se palpan crestas iliacas, sacro o coxis, acortamiento de extremidades inferiores, hipotróficas. **Conclusión:** para lograr una prevención exitosa en el diagnóstico temprano de malformaciones durante el embarazo es necesario un control prenatal estricto ante toda embarazada con factores de riesgos metabólicos, en la primera consulta prenatal es la oportunidad para realizar el cribado de diabetes, si se diagnostica se deberán seguirse realizando los ultrasonidos en un segundo nivel de atención médica para la detección oportuna de malformaciones congénitas asociada a la elevación de la glucemia.

Palabras clave: Síndrome de regresión caudal, agenesia caudal, regresión sacra.

INTRODUCTION

The term “Caudal Regression Syndrome (CRS)” first coined by Bernard Duhamel, 1961, and later on by Dr. Renshaw, 1978, naming it Caudal Regression Secuency or Caudal Agenesis (1). Different terms have been associated, amongst which: caudal dysplasia, dysgenesis or sacral regression, congenital sacral agenesis, sacral defect with anterior meningocele, sacrococcygeal dysgenesis and caudal dysplasia sequence, can be found thus, it can be defined as An infrecuent congenital malformation of the vertebral spine’s lower segments characterized by a truncated

medular cone and aplasia or hipoplasia of the sacrum, with variable involvement of the gastrointestinal, genitourinary, skeletal and nervous systems (2-3). The neural tube defects vary between 1-10 of each 1000 newborn, concretely sacral agenesis manifests within 0.01 and 0.05 of every newborn; in Latin America, in Costa Rica specifically an incidence between 2010 and 2014 of 0.07 for every 1000 newborn (4). Multiple factors have been linked to the manifestation of this syndrome, highlighting: Diabetes Mellitus, being the most relevant factor, and the most coorelated to the aforementioned syndrome, numerous reports indicate the coorelation between gestational diabetes and the presence of fetal anomalies (5).

The mechanism of the malformation is yet to be found, but situations that can be associated have been identified, amongst which stand out:

-High levels of glycosylated hemoglobin (6) as well as hormonal imbalance and lipid protein metabolic disorder (7), which have been corelated to anomalous pregnancies leading to congenital malformations in newborns in up to 5 to 10%, of children of diabetic mothers (8), with an incidence of 1 case per 350 cases, and a rise of 200% over the general population (9).

The predisposing factors of Caudal Regression Syndrome are:

- Hematic depletion: In these patients, an aberrant vessel derived from the yolk artery has been observed, which redirects blood from the abdominal aorta to the placenta through the umbilical cord. This vessel finally acts as a single umbilical artery that steals the flow that should reach the caudal end of the fetus, conditioning ischemia and hypoperfusion (3).

-Genetic: 2 main genes become relevant:

●Homebox gene (hlxb9): Isolated finding of dominant autosomical inheritance in chromosome 7q36 as part of the Currain Syndrome. (10)

●TBX4 gene: Associated with mono-allelic variants with skeletal and lower extremity defects (6). They have also become associated with acinar dysplasias, pulmonary hypoplasias and cardiac hypoplasias, with predominantly right heart failure. Monoallelic variants in the TBX4 gene have been described as being associated with skeletal defects of the pelvis and lower extremities. It was found in a fetus with a new multiple malformation syndrome associated with sacrococcygeal agenesis, bilateral

lower limb aplasia, hypoplastic left hypoplasia, bilateral pulmonary hypoplasia, hydroureteronephrosis and non-immune hydrops fetalis, which presents a nonsense homozygous variant in the TBX4 gene, that said, biallelic variants in the TBX4 gene are associated with a severe syndromic phenotype of sacrococcygeal agenesis and lower limb reduction defects (11).

Other studies have described microdeletion of chromosome 22q13.31 as a very rare condition. A patient has been described who showed neurodevelopmental disorders, dysmorphic features and multiple congenital anomalies in which the analysis of the Priferic Nervous Systena matrix revealed a de novo interstitial microdeletion of 3.15 Mb in the 22q13.31 region encompassing 21 RefSeq genes and seven non-coding microRNAs.(12) Xing et al, in 2024, mentioned the role of the Vangl1 and Vangl2 genes in vertebral development and discovered that their deletion causes vertebral abnormalities that resemble congenital vertebral malformations in humans (13).

-Enviromental factors that have been linked to alterations in the development of the notochord such as: lithium salts, trypan blue, sulfurs, cisteins and retinoic acid (6).

CRS alterations range from partial or complete agenesis at the sacrococcygeal level with associations of urogenital and gastrointestinal malformations, respiratory diseases and a possible cardiac alteration such as ventricular septal defect (5).

There have been different classifications, finding antecedents since 1924, highlighting those of Foix and Hill menad, of these, Renshaw in 1978, based his classification based on radiological findings, describing 4 types:

•Type I: unilateral partial or total agenesis of the sacrum.

•Type II: bilateral and symmetrical partial agenesis of the sacrum.

•Type III: total agenesis of the sacrum associated with variable anomalies in the last lumbar vertebrae and iliac bones.

•Type IV: total agenesis of the sacrum with anomalies of the lumbar and iliac half fused behind the last vertebrae if present (14).

In order to detail the malformations and associated characteristics, Pang, 1993, modified Renshow's classification, grouping them into 5 types with a spectrum ranging from subtotal coccygeal agenesis to total sacral agenesis with the abscense of some lumbar vertebra (10). Guille, 2002, and collaborators created a new classification with correlation to the vertebral deformity and the walking potential of those affected, with the presence of 3 groups, and 3 types according to the separation or fusion of the iliac bones, those being group 1-A. those who will have greater walking potential, after group 2 there is no walking capacity present (4). Subsequently, using the 2006 classification as a guideline, Balouglu published a new classification in which he proposed 2 groups: Group A with the presence of myelomeningocele and Group B: without the presence of said association (6).

In order to make a timely diagnosis, each case should be separately analyzed, as well as disposing of specific hospital infrastructure, such as both image and ultrasonographic studies capable of making prenatal or postnatal diagnoses. Between 60 to 90% of the alterations found are associated with: spinal cord alterations, anorectal alterations, genitourinary alterations, abdominal wall defects and lower extremity defects (6).

Performing a prenatal ultrasounography in the first trimester of pregnancy, proves difficult, due to the incomplete ossification of the sacrum making it difficult to make a timely diagnosi (8). The first signs that allow one to suspect a caudal regression syndrome in this first trimester is the decrease in craniocaudal length (15).

In the second and third trimester, at this point an ossification of the sacrum has already occurred, finding manifestations of it or even its absence. Reassessment by a maternal-fetal specialist is recommended after 22 weeks (16). Using ultrasound, it is possible to identify characteristic patterns of the syndrome such as immobile lower extremities, in a "frog" or "Buddha" position, and appreciable muscular-cutaneous hypotrophy (15). The use of nuclear magnetic resonance (NMR) can be used since it has greater sensitivity and specificity, improving the characterization of associated anomalies, it is the confirmatory imaging study for the identification of the spinal cord that is usually found dysplasia, high terminal cone (17). In cases in which,

due to some circumstances, the prenatal diagnosis could not be made, the diagnosis will have to be made as briefly as possible with the support of the physical examination in which deformity of the lumbar region, narrow hips may be found, gluteal hypoplasia, imperforate anus, flattening of the Intergluteal fold, isolated or complex lower extremity deformities, as well as the use of cabinet studies such as radiographs in lumbosacral projection, and nuclear magnetic resonance imaging with which it can be evaluated. associated anomalies within the spinal canal or spinal and vertebral features (6). The prognosis of fetuses with caudal regression syndrome depends on the severity of the lesion and the coexistence of other anomalies (1). In addition to the degree of vertebral anomaly, an important factor is the associated malformations, with those related presenting a worse prognosis. with the genitourinary system such as bladder exstrophy or renal agenesis (6), gastrointestinal anomalies such as duodenal atresia, imperforate anus, intestinal malrotation, incontinence and encopresis (10).

Signs of neurological deficit such as inability to mobilize the lower extremities considerably obscure the prognosis. This will depend on the severity of vertebral malformations and the presence of associated anomalies, being different for each case. It is important to distinguish the type of sacral agenesis, since each one is more often associated with specific deficits, mainly related to urological and cardiac malformations. Early neonatal death in severe forms occurs due to complications; Infants who survive usually have normal cognitive function (6).

The purpose and justification of reporting this case is due to a globaly low incidence and therefore being very rare and infrequent in the “Dr. Julián A. Manzur Ocaña of the Social Security Institute of the State of Tabasco, Mexico as well. In addintion, the case raises awareness on the importance of carrying out prenatal consultations with purposeful search through ultrasonographic studies from week 22 of gestation, facilitating early detection of possible malformations. Bibliographic research over the last five years was carried out through Scopus, Web of Science and Clarivate among others, to support the topic described, making comparisons and finding the similarities described by other authors worldwide.

Clinical Case Report

Female of 27 years of age, coursing though her second pregnancy on her 34 week of gestation (WG) taking as reference her last menstrual period (LMP) and 32 WG confirmed by obstetric ultrasound; as part of her clinical history the patient refers that the birth control was done monthly during a span of six occasions by a private physician with diverse USG's. She attended her prenatal consultation for the first time at the "Dr. Juan Puig Palacios of the Social Security Institute of the State of Tabasco (ISSET) at 28.5 WG, with obstetric ultrasounds performed by private means, which does not report any abnormality. The personal pathological history includes grade II obesity, gestational diabetes without nutritional or pharmacological control, a history of vaginal and urinary infections, and gestational hypertension.

After proper care and evaluation in the Family Medicine Unit, the patient is referred to the "Dr. Julián A. Manzur Ocaña" from the Social Security Institute of the State of Tabasco (ISSET), to the high-risk Obstetrics consultation, "She is attended by a specialist doctor and reports a pregnancy of 34 weeks by the date of the last menstrual period (LMP), blood pressure of 150/90, blood glucose level of 205 mg/dl, urea 21.4 mg/dl, creatinine 0.5 mg/dl, uric acid 3.4 mg/dl, cholesterol 119 mg/dl, triglycerides 113 mg/dl, glycosylated hemoglobin percentage (%HbA1c) 10.1%, estimated average glucose 243 mg/dl. An obstetric ultrasound and appointment for results are requested.

In the institutional obstetric ultrasound, a pregnancy of 32.4 weeks of gestational age is reported in a breech presentation, with no reported structural abnormalities. 72 hours after the ultrasound, she was admitted to the hospital with transvaginal bleeding, active labor, and premature rupture of membranes of less than 12 hours duration, with umbilical cord collapse, so the decision was made to interrupt the pregnancy. An emergency cesarean section was performed, with the fetus in a transverse presentation and shoulder impaction, making extraction difficult and causing uterine artery tears. A single live female newborn (NB) was obtained, starting resuscitation maneuvers. Secondary apnea was observed, requiring assisted mechanical ventilation to secure the airway, and the newborn was

transferred to the Neonatal Intensive Care Unit (NICU) due to type 1 respiratory distress, with the neonate being hyporeactive and presenting generalized cyanosis.

Upon examination, primitive reflexes are absent, newborn shows short neck, rhythmic heart beat of good intensity, grade II/V murmur present, soft abdomen with hepatomegaly of 2 cm, interrupted spine at approximately T10, iliac crests, sacrum or coccyx not palpable, shortening of the lower extremities, hypotrophic, fracture of the upper tibial epiphysis on the right leg, genitals according to age and gender, with the following anthropometric measurements: weight: 3050 g, height: 39 cm, head circumference: 31 cm, chest circumference: 33 cm, abdominal circumference: 32 cm, foot: 6 cm, APGAR: 3-5/8, SILVERMAN: 00; Vital signs: heart rate: 146 beats per minute, respiratory rate: 0, temperature: 36.9°C, Capurro: 34 weeks of gestational age. An echocardiogram was performed, reporting obstructive hypertrophic cardiomyopathy, dysplastic pulmonary valve with dome-shaped opening causing mild pulmonary stenosis, and ventricular septal defect. (figure 1 and 2)

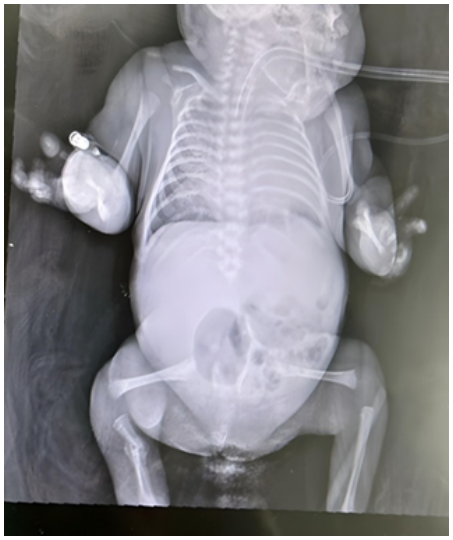

Figure 1. A portable anteroposterior (ap) full-body x-ray reveals interruption of the continuity of the vertebral bodies at the level of the lumbar vertebrae. The pelvic bones are not clearly defined, sacral aplasia and a fracture of the right tibia in the lower extremity are observed. Source: Photograph of the newborn from the clinical archive of the Department of Radiology at ISSET.

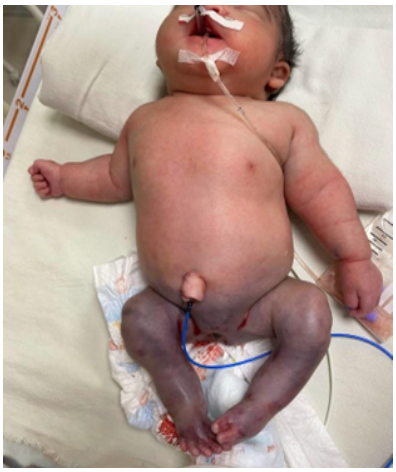

Figure 2. Physical examination reveals hypoplastic lower segment with significant reduction in circulation and edema of the lower limbs. Source: Photograph of the newborn from the clinical archive of the Neonatal Intensive Care Unit of ISSET.

At 72 hours of age and stay in the Neonatal Intensive Care Unit (NICU), newborn begins to exhibit Kramer III jaundice, associated with moderate edema in the lower limbs, as well as bruising from the hypogastrium, with the presence of equinovarus foot, and absence of pedal pulses with capillary refill of 3 to 4 seconds.

On the fourth day of life, borderline levels of hyperbilirubinemia are observed, warranting phototherapy initiation; subsequently, signs of pulmonary congestion develop, prompting the initiation of diuretic therapy.

Due to improvement in her respiratory pattern without desaturations and with the following ventilatory parameters: Maximum Inspiratory Pressure (MIP) 11, Positive End-Expiratory Pressure (PEEP) 5, Fraction of Inspired Oxygen (FiO2) 30, it is decided to initiate Continuous Positive Airway Pressure (CPAP).

On the 6th day, she presents respiratory pattern deterioration requiring reintubation. A thoracic X-ray reveals complete right lung atelectasis. She is then evaluated by the pediatric neurology service, and a neuro-axis tomography reports agenesis of L2, L3, L4, L5, and caudal regression of the sacrum (Figures 3 and 4)

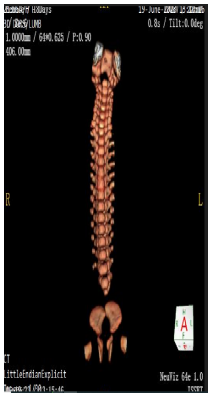

Figure 3. Spine ct scan: presence of cervical, thoracic, and lumbar vertebrae. Disruption at l3, l4-l5, sacrum not observed, iliac crests with a distance of 13 mm. Source: Clinical file of the newborn from the Department of Imaging of ISSET.

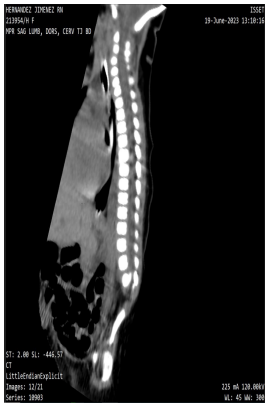

Figure 4. Spine ct scan: intervertebral discs with homogeneous density, preserved spinal canal up to l3, no identification of the spinal cord. Source: Clinical file of the newborn from the Department of Imaging of ISSET.

On the 8th day, leukocytosis is observed with a count of 14,000/µL, as well as a decrease in hemoglobin levels to 11.3 g/dL, elevated D-dimer greater than 10 mg/L, and negative blood culture.

At 15 days of age, there is still evidence of leukocytosis (27,180/µL), prompting a second bronchial culture, establishing a diagnosis of late-onset neonatal sepsis and initiating antibiotic therapy management.

On the 19th day, metabolic, digestive, and hemodynamic deterioration is observed, leading to fasting. Additionally, renal deterioration is evident with hypertension and progressive decrease in urine output. Bradycardia persists along with pulmonary patterns of wheezing and crackles. Due to ongoing hematologic and hemodynamic abnormalities, a packed red blood cell transfusion is administered.

At 28 days of age, the decision is made to extubate for initiation of management with continuous positive airway pressure (CPAP). Forty-eight hours later (30 days of age), there is good evolution in the pulmonary pattern, so the decision is made to progress to nasal prongs. However, she is re-intubated due to reassessment showing poor secretion management.

On the 32nd day of life, there is persistent digestive and hemodynamic deterioration, with intolerance to oral intake, severe abdominal distension, absence of bowel movements, and intestinal loop dilatation on abdominal X-ray. Criteria for Systemic Inflammatory Response Syndrome (SIRS) and septic shock are established. Twenty-four hours later (33 days), positive culture results reveal moderate growth of ¨*Stenotrophomonas maltophilia*¨ sensitive to trimethoprim/sulfamethoxazole.

At 36 days of age, upon examination of the patient, there is pallor of the skin and mucous membranes, with decreased level of alertness, no spontaneous eye opening, and presence of respiratory distress, which worsens with sudden bradycardia and desaturation. Resuscitation maneuvers are initiated, including adrenaline administration, but there is no recovery of heart rate. Resuscitation efforts are discontinued, and the diagnosis of death is made.

DISCUSSION

This is the first documented case, in at least, the last three decades, of Caudal Regression Syndrome at the “Dr. Julián A. Manzur Ocaña” from ISSET; This makes it an epidemiologically relevant issue for the institution, coupled with the importance of highlighting the conscientious performance of prenatal control, making clear the unavoidable practice of screening for prenatal consultation mainly in women with metabolic alterations, as well as the performance of ultrasonographic studies.

Always with purposeful search of possible malformations in pregnant women with risk factors; Worldwide, CRS has an incidence between 0.01-0.05% per 1000 newborns described by Méndez-Salazar and collaborators in 2020 (4).

Gestational diabetes mellitus is the most common metabolic complication in pregnancy. Worldwide, it has a prevalence that varies between 1 – 14%, with Latin America being the most susceptible. This pathology is of high impact, among the most related risk factors for the appearance of gestational diabetes were advanced age, obesity and pathological history, which increases the probability of developing gestational diabetes 1.2 times. The risk factor most recorded in confirmed cases of caudal regression syndrome is gestational diabetes, a pathological entity that conditions the possibility of congenital malformations as a result of hyperglycemia in the first trimester of pregnancy, the prevalence of gestational diabetes is 4% (18). Account 11% in diabetic pregnant the cardiac malformation is the most common anomaly for which the caudal regression associated with diabetes (19-20).

In the case presented, the patient went for the first time to her corresponding Family Medicine Unit until 28.5 SDG, with a history of gestational diabetes without nutritional or pharmacological control; Taking into account the prevalence in 2022 of diagnosed and undiagnosed diabetes, which was 12.6%, and with a prevalence of total diabetes of 18.3%, (21) these are alarming and suggestive data for first-level health personnel inboth public or private institutions, to carry out adequate prenatal control, with the corresponding screenings for risk factors, especially gestational diabetes, and timely detection of any malformation of the product, and if there is even minimal suspicion, the early reference to second or third level healthcare.

The diagnosis of CRS can be made through obstetric Ultrasonography from the second trimester of gestation (16), performed by a maternal-fetal subspecialist. If the necessary infrastructure is not available, the study must be subrogated or the patient referred to a third level healthcare center. To confirm the structural damage of the newborn, Nuclear Magnetic Resonance (MRI) is the study with the greatest sensitivity and specificity since it details up to what level the structure of the vertebral column and its respective spinal cord is interrupted (6). Cardiac anomalies represent 50% of the cases, the most common being cardiomyopathy (22), coinciding with the

case presented, ultrasound revealed asymmetric septal hypertrophic cardiomyopathy, mild tricuspid insufficiency and grade IV/VI mesocardial systolic murmur.

This malformation is associated with hyperglycemia, which in a disorganized manner incites the closure of the neural tube, reduction of mitosis and premature maturation (these findings have been found in experimental models). Clinically, various malformations can be observed, ranging from an absence of the coccyx to agenesis of the lumbar and sacral region, to the formation of a cyst in the spinal cord (syringomyelia) (9).

CRS is associated with visceral malformations in different systems such as the gastrointestinal, genital, urinary, neurological and cardiac systems. Among the musculoskeletal malformations, narrow hips, flattening of the intergluteal fold or gluteal hypopasia may occur, accompanied by various malformations such as flexion contractures, muscle atrophy and femoral hypoplasia (3). In this clinical case, the product presented an interrupted spine at T10, shortening of the lower extremities, hypotrophic, with agenesis of the pelvis, fracture of the tibia and right fibula. The CT findings reported were disruption of the spine from Incomplete L3 that obstructs the spinal canal, fused iliac crest, pubic bones separated by 13 mm from the intervertebral discs, spinal canal preserved up to L3 without being able to identify the spinal cord by this means. The case presented in this report is so interesting to evaluate and comment on its evolution, since it constitutes a case of CRS that involves the lumbosacral region, according to world epidemiological statistics it has a low incidence rate. In the literature there are several cases of CRS in which vertebral agenesis extends to the sacral and lumbosacral level, similar to what was reported in this study; although the rarest are still dorso-lumbosacral agenesis (3).

The prognosis of fetuses diagnosed with CRS is directly related to the severity of the injury and the existence of other anomalies at the same time; Prenatal diagnosis at an early stage will give parents the opportunity to make informed decisions about neonatal complications (23). Multidisciplinary medical support (pediatrics, neonatology, cardiology, orthopedics, imaging and neurology) is essential for the care and treatment of this type of syndrome if the baby is born alive.

CONCLUSIONS

To achieve successful prevention in the early diagnosis of malformations during pregnancy, strict prenatal control is necessary for any pregnant woman with metabolic risk factors. The first prenatal consultation is an opportunity to perform diabetic screening. If diagnosed, follow-up procedures should be performed, such as ultrasounds at a second level of medical care for the timely detection of congenital malformations associated with high levels of glycemia. Emphasis should be placed on the first level of care on the education of the patient and immediate family member about the possible complications of diabetes in pregnancy for adequate metabolic control and their prevention. In the majority of published cases, detection ocurrs during the second trimester of pregnancy, so it becomes key to perform structural ultrasounds (from week 18 to 20) with a focus on checking the integrity of the lumbar spine, especially in patients with Diabetes Mellitus.

ACKNOWLEDGMENTS

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ETHICAL CONSIDERATIONS

Consent was requested from the mother for the publication of the images.

CONFLICT OF INTEREST

There is no conflict of interest to carry out this study.

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