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Case Report

Sirenomelia -Case Report

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Abstract

Although electrical injuries are not a frequent reason for admission to emergency clinics, they are very important in terms of morbidity and mortality. It has important effects on morbidity and mortality as a result of both traumatic and systemic complications. It may occur with different clinics depending on the duration and form of the injury. Clinical findings may not be correlated. In this case report, we discussed the clinical and imaging findings of hemoptysis after electrical injury in a young male patient.

Keywords: sirenomelia; caudal dysgenesis; caudal regression; congenital anomaly

Introduction

Sirenomelia consists of a rare congenital anomaly, which can be considered as the most severe form of caudal dysgenesis and is defined by a fusion of the normally paired bilateral lower limbs into a single median limb (in a mermaid) due to a primary developmental defect that affects several primordial structures of the midline, with varied etiology and not yet fully understood. Abnormalities of embryological development occur around the third to fourth week of intrauterine life and are found alone or associated with genitourinary, gastrointestinal, cardiovascular, respiratory or neurological disorders, including renal, ureteral and bladder agenesis, imperforate anus and absence of external genitalia, considered serious malformations that can lead to death in the perinatal period. Sirenomelia is fatal in most cases due to characteristic pulmonary hypoplasia and bilateral renal agenesis. Its incidence varies between 1-5 per 100,000 live births, with a higher frequency in monozygotic twins, with a sex ratio of 3 boys to 1 girl. Maternal diabetes is the maternal disease known to be associated with sirenomelia, but other risk factors are being associated, such as folic acid deficiency, vitamin A deficiency, maternal obesity, exposure to radiation, use of medications during pregnancy (anticonvulsants, oral contraceptives, nalidixo acid) and use of cocaine during pregnancy. Prenatal diagnosis is based on a morphological ultrasound study that generally identifies changes such as oligohydramnios, renal agenesis, a single lower limb, a single umbilical artery, absence of bladder, undetermined external genitalia, anorectal atresia or lumbosacral agenesis. Considering the exposed pathology, we report a case of sirenomelia associated with other malformations observed in the Neonatology department of the Hospital Materno Infantil do HC-FAMEMA in Marília/SP.

Case Description:

A.C.S.S., 33 years old, white, second pregnancy with a previous cesarean section 2 years and 6 months ago. Having obesity (BMI 35) and previous DM2, she was continuously using NPH insulin (18-0-8) and regular insulin (0-5-4). She had gestational hypothyroidism, requiring the use of Levothyroxine 25mcg (1-0-0) during pregnancy. She denied addictions, use of illicit drugs, smoking, alcoholism. She had adequate prenatal care, negative serology. A morphological ultrasound was performed, which showed absolute oligoamnios and unilateral renal hypoplasia, without other changes. She was admitted to our service due to the hypothesis of renal malformation and oligoamnioabsolute. USG was performed in our service, which showed ILA 31, and she was admitted to hospital and the decision was made to perform a cesarean section. A live fetus was obtained, aged 36 weeks, with the presence of caudal regression syndrome (sirenomelia), undefined sex and imperforate anus. Newborn of indeterminate sex, absence of external genitalia, born by cesarean section, immediate cord clamping, received in heated sterile fields, taken to a heated crib, positioned, mouth and nostrils aspirated, checked for absent heart rate, absent breathing and generalized cyanosis. PPV was started with FiO2 30%, with an improvement in heart rate, but still lower than 100 bpm. A new PPV cycle was performed with FiO2 50%, with no response. OTI was performed in the delivery room, evolving with HR>100 bpm, cyanosis of the extremities, O2 Sat 75%. Referred to the neonatal ICU. Apgar 2/7. PN 2135 g. Height 44 cm. Head circumference 33 cm, chest circumference 31 cm, abdominal circumference 27 cm. In the neonatal ICU, a chest x-ray was performed which showed pulmonary hypoplasia, an abdominal ultrasound was performed which showed bilateral renal agenesis and a transthoracic echocardiogram which showed pulmonary hypertension in the newborn. X-ray of the lower limbs showed that there was no numerical impairment of the bones, just fusion of soft tissues. Karyotype 46 XX, autosomal chromosomes and no apparent abnormalities were collected.

After 6 hours in the neonatal ICU, the newborn died, neonatal resuscitation maneuvers were performed for 20 minutes without success. Placenta sent to pathology: Discoid placenta measuring 14.0 x 13.0 x 4.0 cm and weighing 290 g. The fetal surface is covered with soft, shiny gray membranes. The umbilical cord has an eccentric insertion 4.0 cm from the placental margin, measures 48.0 cm in length by 1.2 cm in maximum external diameter and the sections show two vessels. The maternal surface shows poorly individualized, reddish-brown cotyledons. In the sections we see red and spongy tissue with a yellow and firm area, measuring 0.8 cm in the largest dimension. Free membranes are grayish and opaque. (AA) Diagnosis: Placenta compatible with the 3rd trimester of pregnancy, perivillous fibrin deposits, acute chorioamnion and single umbilical artery.

Discussion

Duhamelem 1961 defined the mermaid syndrome anomaly as the most severe form of caudal regression syndrome. Abnormalities of embryological development associated with sirenomelia can occur around the fourth week of gestation, with changes in the mid-posterior and caudal mesodermal axis of the blastoma. At this gestational age, the kidneys are located in the pelvis while the gonads are intra-abdominal, so any damage to the caudal end of the embryo affects the development of the internal genital organs (except the gonads), the terminal intestine, the bladder, the kidneys and the bones pelvic limbs. From an etiological-physiopathogenic point of view, two hypotheses have been put forward to explain the abnormal development of the lower limbs. One related to a primary error in blastogenesis, due to a primary defect during the final phase of gastrulation where failures of the caudal somites and the caudal bud occur; and the other, related to a vascular event based on the abnormal development of the umbilical vessels that results in the presence of a single aberrant umbilical artery causing an insufficient blood supply to supply the caudal portion of the embryo. Studies suspect an autosomal dominant neomutational genetic cause with male predisposition, since in all statistics boys are more prevalent than girls. It is, therefore, a multisystemic condition that affects newborn survival. The main malformations associated with sirenomelia are renal agenesis (66.6%), colon atresia (blind pouch -100%), anal imperforation (97.8%), rudimentary or absent uterus (55.1%), pelvis-sacral anomalies (100%), single umbilical artery (92.1%). Associated renal agenesis can be unilateral or bilateral, with the prognosis of individuals with unilateral renal agenesis (URA) depending on the function of the kidney present. Sirenomelia associated with bilateral renal agenesis (BRA) is incompatible with life because the prolonged absence of amniotic fluid results in pulmonary hypoplasia, leading to severe respiratory failure at birth, which is the most frequent cause of neonatal death. Risk factors maternal disorders frequently associated with spinal dysraphism are: Folic acid deficiency - certainly associated with myelomeningocele; Gestational diabetes – with a pathophysiological mechanism that has not yet been clarified, but maternal diabetes is related to an increased incidence of caudal regression syndrome; Maternal obesity – studies show a greater likelihood of developing spina bifida in the fetus; Medications – exposure of the fetus to anticonvulsants is associated with an increased risk of spinal dysraphism, and the effects of these medications may be due to changes in folic acid metabolism. According to Duhamel's classification, the syndrome can be grouped into different forms due to the degree of severity, according to the anatomy and the alteration found in the lower limbs. In 1987, Stocker and Heifetz classified sirenomelia according to the agenesis of the bones of the lower limbs: Type I - no numerical impairment of the bones, only fusion of soft tissues; Type II - presence of a single fused fibula; Type III - absence of fibula; Type IV - presence of partially fused femurs and single fibula; Type V - with partially fused femurs and absent fibula; type VI - with single femur and tibia; in Type VII - a single femur is present, with no tibia, fibula and feet being observed. Diagnosis is made by ultrasound, preferably in the first trimester of pregnancy where the fetus is surrounded by a sufficient amount of amniotic fluid that comes almost entirely from the amnioblasts of the amniotic membrane making it easier to visualize the changes present, as the oligoamnios present in the next trimesters of pregnancy can make it difficult to identify associated malformations. The presence of oligoamnios, intrauterine growth retardation, difficulty in visualizing the lower limbs is suggestive, being easier to identify when there is only the presence of a femur and a tibia. Doppler ultrasound is useful because it allows the identification of a single umbilical artery through the visualization of a large artery that emerges from the abdominal aorta and runs centrally towards the umbilical cord, corresponding to the persistence of a vitelline artery - this being a finding that can be considered pathognomonic for prenatal diagnosis of sirenomelia. Other possible changes visualized on ultrasound are intrauterine growth delay, hypomotility, renal anomalies, and other uncommon malformations such as cyclopia and anencephaly.

Conclusion

Sirenomelia is a rare syndrome, and in its etiopathogenesis, several risk factors are considered, such as maternal age, affecting pregnant women under 35 years of age, maternal pathologies, multiple pregnancies, single umbilical artery, oligohydramnios, among others. Due to its low incidence and associated with the lack of knowledge of its etiological factors, without any definitive conclusion defined, we present a case report aiming to contribute to data from the medical literature. The reported case was diagnosed as Sirenomelia type I in addition to visceral malformations typical of the pathology, such as the absence of bladder, bilateral renal agenesis, imperforate anus, undefined external genitalia and the presence of a single umbilical artery. In this case, the presence of pulmonary hypoplasia resulting from bilateral renal agenesis was observed, a condition that is most frequently associated with early mortality in newborns with sirenomelia.

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