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PARTICULARITIES OF A FETUS WITH SIRENOMELIA AND CONCEPTUAL SEPARATION FROM CAUDAL REGRESSION SYNDROME

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ABSTRACT

Sirenomelia is a very rare congenital anomaly, defined by partial or complete fusion of the lower limbs. Additional malformations may also occur, the phenotype being variable. Over time, sirenomelia has been considered the last stage of the evolution of caudal regression syndrome. However, the different characteristics in the context of the two syndromes, such as the single umbilical artery or, less often, two vessels, renal agenesis and the imperforate anus characteristic of sirenomelia as opposed to caudal regression syndrome where all three umbilical vessels are identified, along with sacral agenesis, led to the separation of the two entities. Sirenomelia is a multifactorial disease, with genetic heterogeneity, most cases being sporadic. Its pathogenesis is incompletely deciphered.

Thus, we present a case of sirenomelia, diagnosed after abortion at the end of the first trimester. The fetal autopsy gave us details of skeletal and visceral abnormalities. The sex of the aborted could not be determined due to the incomplete development of the genital tract.

To our knowledge, this is the first case reported in Constanta County. We hope that this presentation will be useful in trying to understand the causes of this condition.

Keywords: sirenomelia, caudal regression syndrome, renal agenesis

Introduction

Sirenomelia is a very rare congenital anomaly, characterized by partial or complete fusion of the lower limbs. Additional malformations may also occur, including genitourinary abnormalities, gastrointestinal abnormalities (imperforate anus), lumbar spine abnormalities (spina bifida), sacro-coccygeal agenesis, heart malformations, and agenesis or hypoplasia of one or both kidneys. Sirenomelia is usually fatal in the newborn period.

This condition is found in about one in 100,000 live births, 150 times more common in monozygotic twins than dizygotes and singletons (1), without racial predilection, and the ratio between male and female is 3: 1 (2).

Sirenomelia is a multifactorial disease, with genetic heterogeneity, most cases being sporadic. Genetic studies in guinea pigs are an important step towards understanding the pathogenesis. Sirenomelia occurs in mice that do not have Cyp26a1, an enzyme that degrades retinoic acid (RA) and causes a decrease in bone morphogenetic protein (Bmp), signaling in the caudal embryonic region. The phenotypes of these mutant mice suggest that human sirenomelia is associated with an excess of RA signaling and a deficit in Bmp, signaling in the caudal body. The environmental triggers are not well known either.

Although the causes remain unknown, two pathogenic theories have been developed:

The first theory, known as vascular theft theory (3), is based on the aberrant vascular model of individuals with sirenomelia. Invariably, fetuses have a single umbilical artery, which originates immediately below the celiac branch. Then, the aorta narrows aberrantly and has a considerably smaller number of branches for vascularization of the kidneys, intestines and genitals. Thus, the umbilical artery redistributes blood flow to the placenta, depriving the circulation in the lower half of the body.

The second theory predicts the impairment of organogenesis due to the action of teratogenic factors during gastrulation (4). Maternal diabetes mellitus has been associated with caudal regression syndrome and sirenomelia (5). Smoking, retinoid administration, and prenatal cocaine exposure have also been identified in association with sirenomelia (6).

Case presentation

Non-viable conception product, gestational age 12-16 weeks, 15 cm long, from a primiparous pregnant woman 23 years old, with low socio-economic status and unsupervised pregnancy, no known personal pathological history, no heredocolaterale antecedent of congenital anomalies, with no history of exposure to teratogenic factors, including drugs or radiation.

Figure 1: Sirenomeliac fetus with normal development of the cephalic extremity and upper limbs, but with a herniated abdominal contents and a single lower limb.

After fetal extraction, the normal development of the cephalic extremity and upper limbs is noted, but with a herniated abdominal contents and a single lower limb (Figure 1).

Following the autopsy of the chest, the heart and lungs have normal dimensions, but, on the section, the heart is unicameral.

In the abdomen, the stomach is absent, and the liver, the spleen, and the intestine are herniated (Figure 2). Anal canal atresia is also observed.

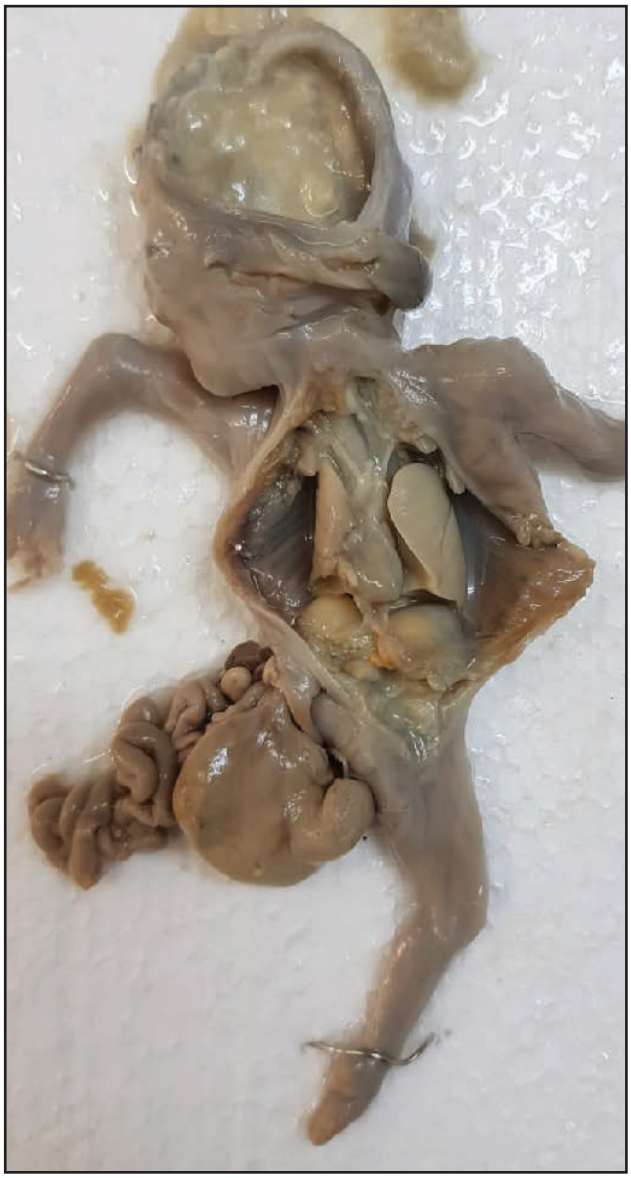

Figure 2: Sirenomeliac fetus autopsy with normal dimensions of heart and lungs. The stomach is absent and the liver, spleen, and intestine are herniated.

The fetus has right renal and bladder agenesis. The genitals are incompletely developed and therefore could not be established the sex of the aborted. The single lower limb is

fully developed. After the incision, a femur and a single tibia were identified.

Discussions

Sirenomelia is a very rare multisystemic malformation with phenotypic variability. Usually, it is a condition incompatible with life due to visceral abnormalities. There are some exceptional cases of survival due to the operations of reconstruction of the pelvic organs, respectively of separation of the lower limbs and the existence of a functional kidney.

From a clinical point of view, the spectrum varies from mild forms, where the fusion of the lower limbs is superficial, at the level of the skin, with the preservation of vascularity and bones for both limbs, and up to the most severe form, in which only a bone fragment can be identified.

The fetus has a characteristic Potter-type facies with flattened nose, hypertelorism, prominent epicanthal folds, low-set abnormal ears, retrognathia (7) and wrinkled skin.

The constellation of visceral malformations is also variable. Usually, the renal and urethral dysplasias were observed, but bilateral renal agenesis was also frequently reported. Urinary tract malformations are associated with various genital abnormalities, agenesis or hypoplasia of the external genitalia, usually without affecting the development of the gonads.

Referring to the gastrointestinal tract, there are tracheo-esophageal fistulas, intestinal atresia and imperforate anus.

Over time, sirenomelia has been considered the last stage of the evolution of caudal regression syndrome. However, the different characteristics in the context of the two syndromes, such as the single umbilical artery, or, less frequently, two vessels, renal agenesis and imperforate anus characteristic of sirenomelia as opposed to caudal regression syndrome where all three umbilical vessels are identified, along with sacral agenesis, have led to the separation of the two entities.

Both conditions are closely related to the diagnosis of mothers with type II diabetes, about 25% of them give birth to children with caudal regression syndrome and only 2% of them have children with sirenomelia. Of course, some teratogenic environmental factors have been identified, such as the administration of retinoids during pregnancy or cocaine use, but there are also situations, as in our case, where the already known environmental factors cannot be identified. That is why it is very important to describe new cases with the intention to identify new causes.

Conclusions

Sirenomelia is a very rare congenital anomaly. To our knowledge, this is the first case reported in Constanta County. We hope that this presentation will be useful in trying to understand the causes of this condition.

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