Fetal Acardia Syndrome in monozygotic twin pregnancy: Case report

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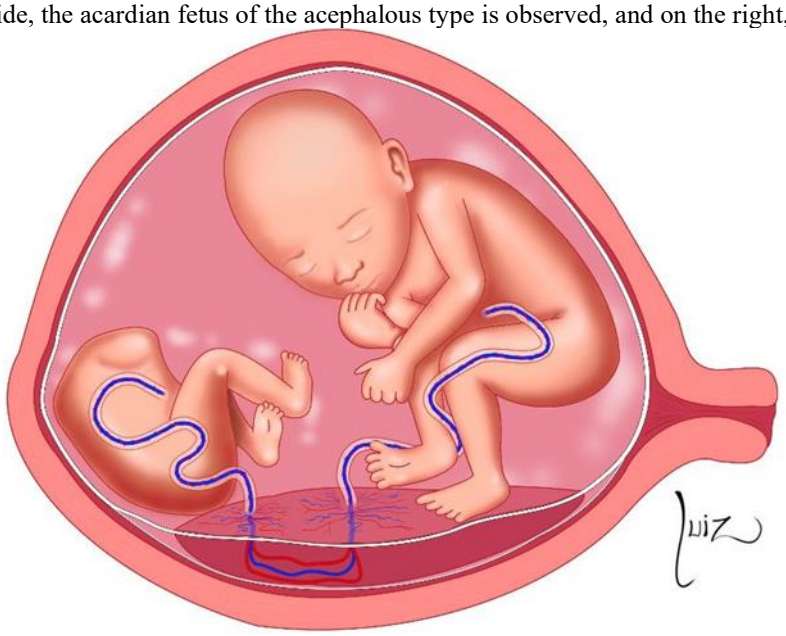
ABSTRACT
Fetal acardia is a rare complication that affects monochorionic/diaminiotic twin pregnancies and is characterized by the presence of a fetus with no heart (acardiac fetus) or a rudimentary heart (hemicardiac fetus). The objective of the present article is to present a case of fetal acardia with diagnosis confirmed by anatomopathological examination in which the "pump fetus" survived the pregnancy without specific treatment and without intercurrences.

Keywords: Diamniotic, Twin pregnancy, Monochorionic, Fetal Acardia Syndrome.

1 INTRODUCTION

Fetal acardia is a rare complication affecting monochorionic/diaminiotic twin pregnancies. It is characterized by the presence of a fetus that does not have a heart (acardian fetus) or in which there is a rudimentary heart (hemicardial fetus) (Figure 1). In this syndrome, the acardiac/hemicardial twin receives, through arterioarterial and venovenous anastomoses, deoxygenated blood pumped by the heart of the healthy fetus ("pump" fetus), which can bring a series of hemodynamic consequences to the viable fetus.1-3 years

Figure 1: Illustrative representation of monochorionic and diaminiotic twin pregnancies associated with fetal acardia syndrome. On the left side, the acardian fetus of the acephalous type is observed, and on the right, the "pump" fetus.



Source: personal archive.

The syndrome was first described by Benedetti in 1533. Its epidemiology, encompassing risk factors, is poorly understood, although the number of case reports has increased. Based on the few existing data in the literature, its incidence is estimated between 1:35,000 and 1:48,000 pregnancies, especially in monozygotic pregnancies (1% of these) and may occur rarely in dizygotic pregnancies.Ps. 2:4-7

Van Gemert et al., based on ultrasound examinations and assisted reproductive technologies, demonstrated an incidence of 2.6% in monozygotic twin pregnancies and 1:9,000 to 1:11,000 in pregnancies in general.8

The sequence of reverse arterial perfusion (TRAP sequence), also known as TRAP syndrome, is pointed out as one of the main risk factors and presumed pathophysiological mechanism for the occurrence of fetal acardia.9,11 This name is explained by the dynamics of the blood flow of one of the twins, which is exactly the opposite of that observed in normal fetal circulation. In it, the blood coming from the "pump" fetus, with a lower oxygen content, reaches the other twin (fetus that will develop acardia) through two umbilical arteries from anastomoses that develop over the placenta. This blood irrigates only the lower portions of the recipient twin and then returns to the placenta, from where it returns to the "pump" fetus and restarts its cycle.9.10

These pathophysiological changes have two catastrophic consequences for the recipient fetus: only the lower portions of its body will develop and it will become unviable. Added to this, the "pump" fetus, under increased cardiac demand, can develop cardiomegaly and severe congestive heart failure, which can culminate in its death.10-12 months

In view of the above, it is evident that fetal acardia syndrome is a rare complex phenomenon, whose comprehension, in its most diverse aspects, still requires further studies. Thus, the objective of the present article is to present a case of fetal acardia with a diagnosis based on anatomopathological examination in which the "pump fetus" survived pregnancy without specific treatment and without intercurrences.

2 CASE REPORT

This is a 28-year-old woman, gestational age (GA) of 37 weeks and 6 days, in her eighth pregnancy, with a history of 1 vaginal delivery, 2 cesarean sections, 1 stillbirth and 4 abortions. She is brown, smoker, non-alcoholic, and was under high-risk prenatal care (HRAP) for alloimmunization for the Rh factor. Throughout her pregnancy, she had an episode of urinary tract infection, which was treated with cure. She received heparin throughout her pregnancy due to a previous thromboembolic event.

According to the probable date of delivery, the patient was admitted to the hospital, where she received care, for cesarean section and bilateral salpingotripsy. Surgical antibiotic prophylaxis was performed and surgery was performed under spinal anesthesia. During delivery, a live cephalic newborn, APGAR 8/8, was extracted, which was assisted by a pediatrician in the delivery room, without the need for neonatal resuscitation. Subsequently, a single placenta was manually extracted, which was apparently complete and had translucent membranes and an umbilical cord composed of three vessels. A second amniotic sac containing amorphous material was removed from it, presumably the fetal deceased who was referred for anatomopathological evaluation.

This evaluation, in turn, showed a monochorionic and diamniotic twin placenta with a third trimester pattern, measuring 19.0 x 16.0 cm in area and 2.5 cm in thickness. The plancentary fetal surface presented a prominent chorionic vascular web, with apparent arterioarterial and venovenous anastomoses between the fetal circulations, findings compatible with those found in the TRAP sequence.

An intact amniotic sac measuring 8.0 cm in diameter was identified in the chorionic plaque. On opening, an acardial malformed fetus was observed, consisting only of the lower limbs (Figure 2a). Adjacent to these structures, a solid-cystic formation measuring 9.0 x 8.0 x 6.0 cm was observed (Figure 2b), containing organoid structures, including intestinal segments. A rudimentary cephalic portion was identified, with structures of the oropharynx and airways wrapped in skin, as well as salivary gland, lymph nodes and ectatic lymphatic vessels with cystic hygroma pattern. Nerve tissue surrounded by meninges was also observed.

Figure 2: Amorphous material removed together with the placenta. A - Integrated amniotic pouch containing lower limbs (arrow); b- solid-cystic structure containing intestinal segment (two arrows) and rudimentary cephalic portion (3 arrows).

In the chorionic plate, the umbilical cord of the acardial fetus, which measured 4.0 x 0.9 cm and was composed of three vessels, was observed (Figure 3). The insertion of the umbilical cord of the normal fetus, which measured 3.5 x 1.3 cm and also contained three vessels, was also identified. On the cuts, the placental disc exhibited Spongy, brown-winey appearance, without infarctions or other lesions worth noting. In view of all these findings, the diagnosis of monochorionic twin placenta was established and diamniotic, with pattern from third quarter gestational, associated with an acardian malformed fetus, probably associated with TRAP sequence.

Figure 3: Photo showing the three vessels found in one of the umbilical cords.

Source: Authored by the authors.

3 DISCUSSION

Data from the literature show that the incidence/prevalence of twin pregnancies increases significantly with advancing age, reaching its maximum peak in an age group that is around 40 years of age. It is speculated that, at this stage of female reproductive life, follicle-stimulating hormone (FSH) would have maximum activity in stimulating the growth of ovarian follicles, favoring the maturation of multiple follicles.13

Added to this is the advent of assisted reproduction techniques, with ovarian hyperstimulation and in vitro fertilization, which are increasingly widespread.13-15 In the present case, although the patient did not present the factors in question, she was multiparous, which is also associated with an increased risk of twins.

Fetal acardia in monochorionic twin pregnancies has placental teratoma as the differential diagnosis. It is essential that the anatomopathological examination be thorough so that the basic morphological characteristics of fetal acardia are observed, since these do not make up the structure of the teratomas. Among them, the presence of a cord stands out Umbilical vascularized and exclusive to the amorphous mass, as well as a degree of organization of the axial axis of the acardiac fetus. It may present the development of the cranial and caudal poles, as well as the growth of some internal organs and the central skeleton (spine).16.17

In the present case, the amorphous mass found next to the placenta had an exclusive and complete umbilical cord, surrounded by Wharton's jelly and covered by an amniotic membrane, consisting of 2 arteries and a vein. In addition, macroscopy showed lower limbs and some rudimentary internal organs, corroborating the established diagnosis.

In 1960, Napolitani and Schreiber pointed out characteristics of fetuses with fetal acardia syndrome and, based on this, established a classification based on morphology.18-20 This classification subdivides fetuses into categories (see below), but does not provide information related to prognosis or therapeutic management:

Acephalous – Without formation of the cephalic pole and thoracic structures, but in which it is possible to distinguish the lower limbs. It is the most common presentation (60-75% of cases).

Amorphous – Represented by fetuses that present as an indistinguishable tissue mass. It accounts for about 20% of cases.

The body and extremities of fetuses in this category are formed, but the skull and face have deformities. They account for about 10% of cases.

Achormic – This group includes those fetuses in which only the cephalic structure is detected, inserted close to the umbilical cord. This is the rarest form, accounting for about 5% of cases.

In the present case, it was not possible to include the acardic fetus in any of these morphological categories, with overlapping of traits from more than one group. This fact reinforces the need for further studies that allow a better understanding of the processes involved.

In the prenatal diagnosis of fetal acardia syndrome, ultrasound methods can detect the TRET sequence. The findings, especially between the eighteenth and twenty-fourth weeks of gestational age, include the presence of a fetus with severe malformations, including the absence of the fetus cephalic pole and functioning heart. In addition, Doppler velocimetry may be useful in identifying reverse arterial perfusion in the acardian fetus and in characterizing abnormal vascular communications present in the placenta and umbilical cord. 20.21 AM

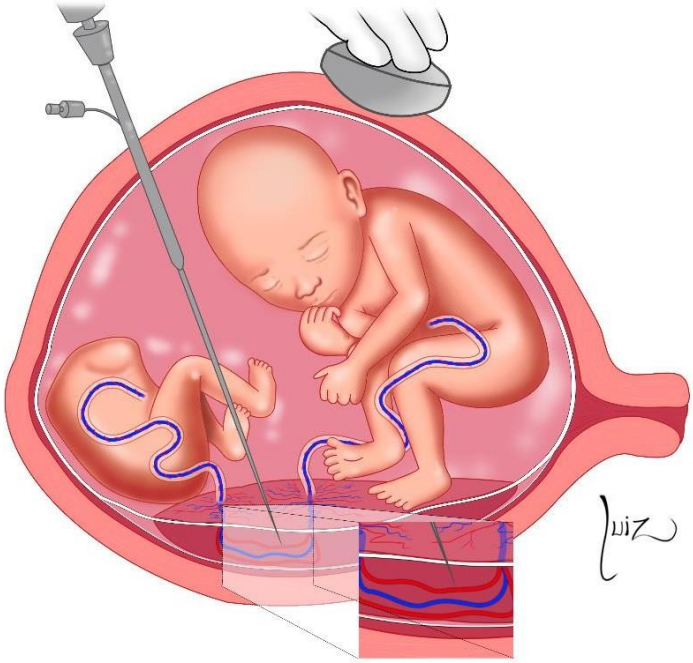
In the present case, based on the data obtained from the evaluation of the patient's medical records, it is assumed that the prenatal diagnosis of the syndrome has not been defined. In this scenario, early diagnosis can be essential to ensure the development of the donor fetus, indicating antenatal intervention. The survival of the "pump" fetus is closely related to factors such as the presence or absence of hemodynamic complications. In this sense, Wong et al 22,23, based on ultrasound findings related to the size of the acardial fetus and signs of hemodynamic involvement of the "pump" fetus, proposed a classification that can help determine the most severe cases of the syndrome and indicate intervention.

According to this classification, pregnancies in which the acardian fetus is considered small when compared to the "pump" twin and in which the latter does not present hemodynamic disorders, are considered to have a better prognosis and patients can have conservative management. In the present case, such characteristics were present, which may explain the fact that the donor fetus was born without cardiovascular or any other complications.

The management of fetal acardia syndrome is based on the detection of the TRAP sequence and on actions aimed at maximizing the chances of survival of the "pump" fetus, to the detriment of the acardian fetus. Among the most widely used methods in medical practice and with good clinical outcomes are ultrasound-guided laser coagulation (Figure 4) and radiofrequency ablation of intrafetal vessels.23rd

However, it is not yet clear what the correct time is for the intervention. Recent studies show that the first trimester would be the best time and the period with the best results.23.24

Figure 4: Illustrative representation of the ultrasound-guided laser coagulation procedure.



Source: personal archive.

Therefore, the importance of prenatal care is highlighted, not only for the early diagnosis of this condition, but also for a better characterization of the syndrome, allowing the institution, in an appropriate time, to perform an intervention aimed at ensuring the survival of the viable fetus, as pointed out by Szekely et al 25 and Davenport et al 26.

4 FINAL THOUGHTS

In view of the above, it is concluded that fetal acardia syndrome is a rare phenomenon, which still requires further studies and research in order to expand its understanding in its most diverse aspects. In addition, it is evident that adequate prenatal care is extremely important for early diagnosis and maintenance of the viable fetus.

Finally, anatomopathological diagnosis is always indicated for diagnostic confirmation, with placental teratoma as the main differential diagnosis. The clinicopathological correlation is fundamental, and it is necessary to complete the request for anatomopathological examination with the main clinical and imaging data.

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