REVIEW ARTICLE

Advances in Ultrasonic Assessment of Acardiac Twin

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ABSTRACT

The development of an acardiac anomaly, one of the most severe human malformations, is a rare complication unique to monochorionic multiple pregnancies. In this condition, the primary malformation is the lack of a well-defined cardiac structure in one twin (the acardiac twin), which is kept alive by its structurally normal co-twin (the pump twin).

The final diagnosis for a twin pregnancy with an acardiac twin may be established after the 9th week, when the cephalic extremity and limbs can be precisely determined.

Direct signs that lead to the diagnosis of an acardiac twin include the absence of cardiac and fetal movements, difficult visualization of the trunk and cephalic poles, thickening of subcutaneous tissue and the presence of umbilical artery reverse flow towards the acardiac fetus mass. The pump fetus may demonstrate hydramnios, cardiomegaly and heart failure.

Once an acardiac anomaly is diagnosed, both the acardiac and pump twins should be assessed to classify the pregnancy according to prognostic factors based on the size and growth of the acardiac twin and the cardiovascular condition of the pump twin. Assessing the extent of heart damage in the pump fetus is a key step in pregnancy management.

There are two available options, either conservative or interventional, for the management of these pregnancies. Serial ultrasound surveillance is important for detecting any worsening of the condition, which may suggest the need for interventions to optimize the pump-twin's chance for survival.

We performed a retrospective analysis of three cases of monochorionic twin pregnancies diagnosed with a twin reversed arterial perfusion (TRAP) sequence during the first and second trimesters. We compared our data with those offered by a review of the literature.

Keywords: Acardiac, Pump twin, TRAP, Monochorionic.

INTRODUCTION

The mythology of the Greeks and Romans is full of monsters of fiction: Giants, cyclops, centaurs, hydras and gorgons. The accounts of travellers reproduced in the natural history of Pline l'Ancien reported the existence in distant countries of men with a dog's head (baboons), a single tall foot (sciapode) or beings whose face is embedded in the chest (or acephala blemmyes). The acardiac fetus may explain some of these monstrous forms and is featured in the work of Hieronymus Bosch.[1]

Monochorionic twin pregnancies are among an increased obstetrical risk pregnancy group. They are based on intertwin discordance, which encompasses fetal size, amniotic fluid volume, fetoplacental hemodynamics and structural defects. In all of these high-risk situations, death of the most affected twin can trigger an exsanguination of the survivor into the dead twin and its placenta, thus threatening the survivor with death or the development of ischemic lesions in various organs, but primarily the brain, in about 20% of cases for either condition.[2]

Acardiac anomaly, one of the most severe human malformations, is a rare complication unique to monochorionic multiple pregnancies. In this condition, the primary malformation is the lack of a well-defined cardiac structure in one twin (the acardiac twin), which is kept alive by its structurally normal co-twin (the pump twin) through a superficial artery-to-artery placental anastomosis. Arterial blood flows in a retrograde fashion from the pump twin towards, rather than away, from the affected twin, which is why an acardiac anomaly is also referred to as a twin reversed arterial perfusion (TRAP) sequence.[3]

This TRAP sequence corresponds to the most extreme manifestation of twin-twin transfusion syndrome.[4]

We performed a retrospective analysis of three cases of monochorionic twin pregnancies diagnosed with a TRAP sequence during the first and the second trimesters. We compared our data with those offered by a review of the literature.

CLINICAL CASES

Case 1

A nulliparous 23-year-old patient with no significant medical history attended our department after 18 weeks of amenorrhea for excessive abdominal enlargement.

An ultrasound performed during the morphologic exam revealed a live fetus and another structure, imprecisely defined, surrounded by a membrane, characteristics that suggested a monozygotic twin pregnancy with a normal live fetus and a second acardiac twin that was acephalus with associated hydramnios. Biometry of the fetus with cardiac activity corresponded with the amenorrhea. The second structure, with a cystic hygroma, demonstrated vascular flow only through the umbilical cord and without evidence of cardiac activity.
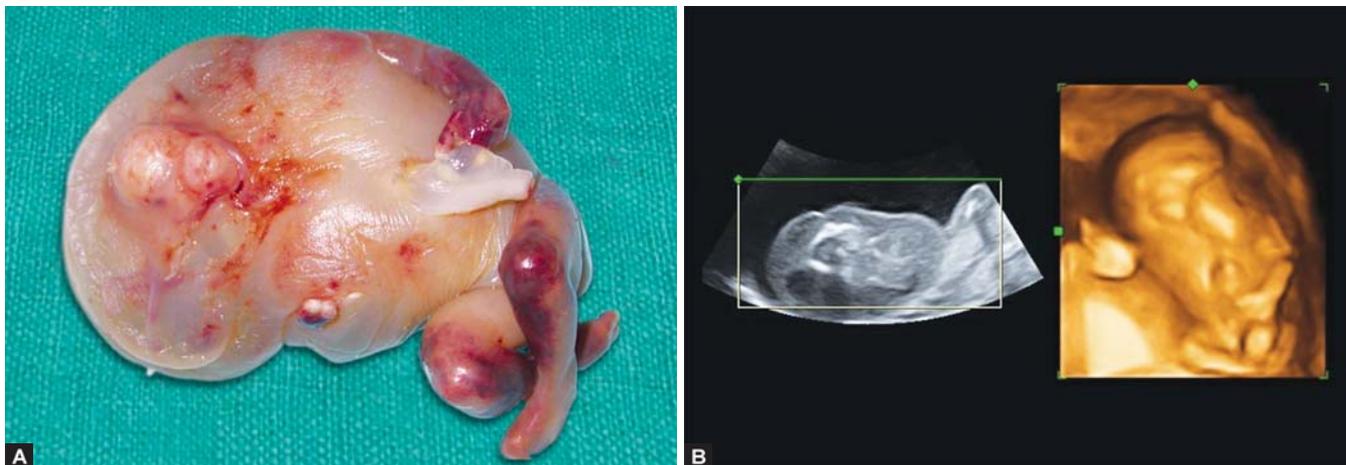
Figs 1A and B: (A) Acardiac fetus at 18 weeks of amenorrhea, (B) ultrasound evaluation of an acardiac fetus with visualization of a cystic hygroma

Fig. 2: Render mode ultrasound to visualize umbilical cords insertions
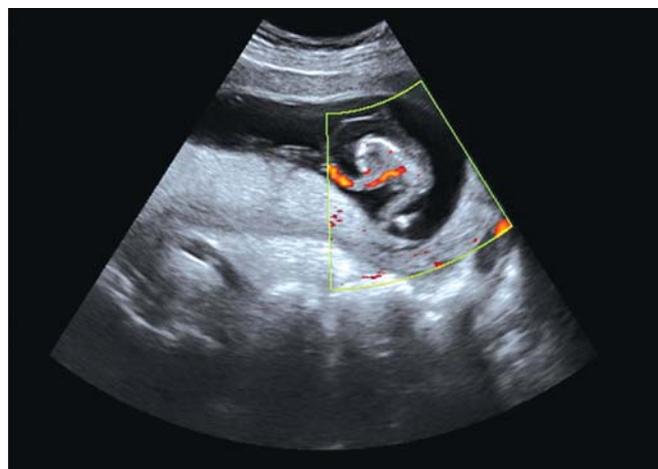

Fig. 3: Visualization of umbilical artery flow with absent cardiac activity

24 hours following the ultrasound examination, the patient had a spontaneous abortion resulting in a fetus with almost normal morphology of approximately 500 gm but with no signs of viability and a dismorphic structure that lacked the upper two thirds of the head, the right upper limb and with all other limbs showing syndactyly (Figs 1A and B). The acardiac fetus weighed about 300 gm; the ratio between the two fetuses was 1.66.

Pathology exam of the malformed fetus confirmed the diagnosis of an acardiac twin (acardius anceps), acephalus and a single umbilical cord artery. No cardiac, pulmonary, tracheal or thymic structures were identified. The genitalia of both fetuses were masculine. The monochorionic placenta had umbilical cord insertions 3 cm apart from each other located eccentrically towards the placental margin (Fig. 2).

Case 2

A 30-year-old primiparous patient with no medical history had a child born at term via vaginal delivery. A routine ultrasound exam performed after 13 weeks of amenorrhea demonstrated a twin monochorionic biamniotic pregnancy with one live fetus corresponding with the amenorrhea.

In the second fetus, we noted the absence of a cephalic pole, initially interpreted as anencephaly. Lack of a fetal heart as well as the presence of flow through the umbilical cord, later established the diagnosis of an acardiac twin (Fig. 3). An early amniocentesis was performed and both structures had a 46XY karyotype. The patient was seen at every 2 weeks for ultrasound examination, fetal echocardiography and Doppler ultrasound.

We want to emphasize two significant aspects: the Doppler exam rendering of an arterioarterial shunt at the placental surface and a major volume difference between the two fetuses (Figs 4A and B).

The patient was readmitted at 30 weeks of amenorrhea for uterine contractions. Ultrasound exam revealed one fetus with biometry corresponding with the amenorrhea and without signs of cardiac failure. The second fetal structure was dysmorphic and without umbilical cord flow (spontaneous obliteration).

Three days after the initiation of tocolytic therapy and the administration of steroids, the patient spontaneously delivered a live 1,200 gm fetus with an Apgar score of 7 at 5 minutes; the child died 18 days after birth.
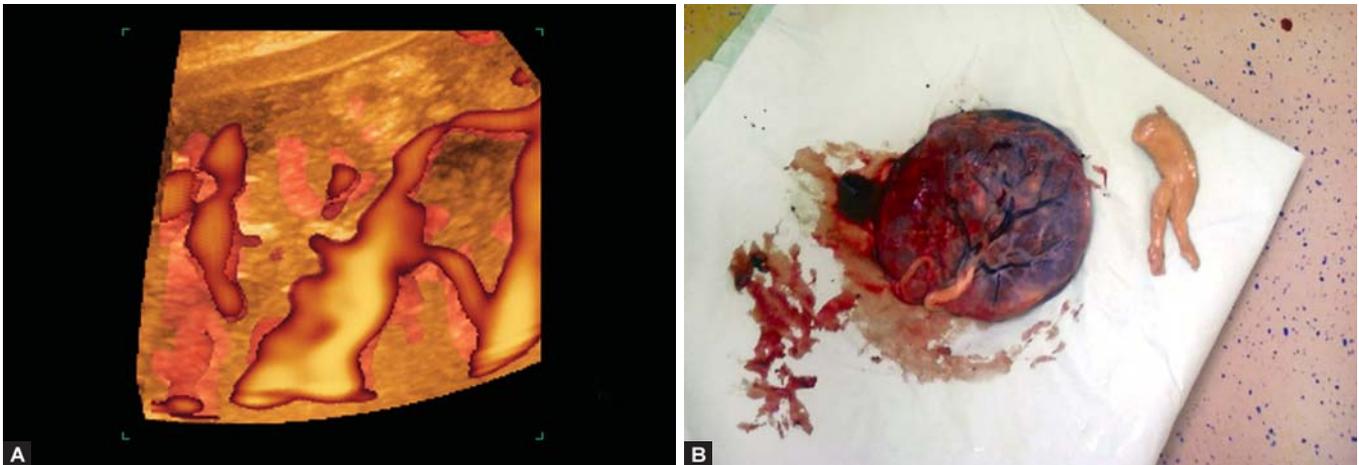
Figs 4A and B: The placental arterioarterial shunt: Ultrasound (A) and macroscopic (B)

Pathologic exam of the malformed fetus confirmed the diagnosis of an acardiac twin with ambiguous genitalia and an absent head, thorax and upper limbs, but with the presence of the pelvis and lower limbs. The fetus had a length of 10 cm and weighed 150 gm.

Case 3

A 21-year-old primiparous patient had a history of an intrapartum fetal death due to an acute hypoxic event. The present pregnancy was monitored beginning at 9 weeks when the diagnosis of a twin pregnancy was established. Ultrasound exam at 11 weeks established the diagnosis of a monozygotic twin pregnancy with one acardiac fetus (Fig. 5). At 14 weeks, an early amniocentesis was performed. Both fetuses had a 46XY karyotype.

The pregnancy demonstrated normal development until 20 weeks of amenorrhea when spontaneous obliteration of the umbilical flow towards the acardiac twin occurred. The second twin had physiological development until 39 weeks. The patient delivered by cesarean section a live fetus weighing 3,800 gm with an Apgar score of 10 at 5 minutes with normal perinatal development. At birth, the second fetus was papyraceous with the appearance of acardius acephalus.

DISCUSSION

The development of an acardiac anomaly is a rare complication of monozygotic multiple pregnancies. It was first described in the 16th century (Benedetti, 1533) and occurs in one out of 35,000 deliveries.[5]

About 1% of all MC twin pregnancies are complicated by a structurally normal twin perfusing an acardiac co-twin, which is often anencephalic, by means of a unique set of vascular connections.[20] Importantly, the acardiac twin does not share the placenta whatsoever.

Umbilical artery flow, which normally runs from the fetus towards the placenta, is instead reversed towards the acardiac twin. At the same time, there are venovenous anastomoses by

Fig. 5: Ultrasound exam at 11 weeks of gestation

which deoxygenated blood from the acardiac twin reaches the normally developed twin through the umbilical vein.[6] The acardiac twin receives all of its blood volume from the so-called pump twin through this reversed arterial connection.[7]

The TRAP sequence is considered to be the key element in the formation of acardiac fetuses, but the temporal sequence is not yet clearly understood.

According to this theory, an anomaly occurs during cardiac embryogenesis. The lack of cardiac activity leads to flow reversal in the umbilical artery. Blood circulates via arterio-arterial anastomoses from the pump twin to the acardiac twin. The acardiac twin, thus, receives partially deoxygenated blood.

The acardiac fetus continues to grow due to the blood flow supplied by the pump fetus. The severity of malformation is directly proportional to the time elapsed from the occurrence of the TRAP sequence to delivery.

The direct effect of the TRAP syndrome on one of the fetuses has been classified according to the degree of tissue resorption as: (A) Acardius anceps (paracephalus) with body and extremities present and a partially developed head and face, (B) acardius acephalus (omphalus) with the pelvis and lower limbs developed and the head, thorax and upper limbs absent,

(C) acardius amorphous with the presence of an amorphous mass, indistinguishable organs and the presence of some axial structures, (D) acardius acormus with only the head present (extremely rare type).[8]

Relatively deoxygenated blood from the pump fetus travels backwards through the anastomoses and reaches the second fetus whose lower body is oxygenated but who presents with secondary atrophy of the heart and other organs located in the upper part of the body.[3,9-11]

Acardiac fetuses may present limb movement due to intact spinal reflexes, which make the diagnosis of fetal death uncertain. Gimenes-Scherer and Davies have described numerous anomalies: The number of affected organs decreases in the craniocaudal direction, the head and upper limbs are most severely affected and the distal segments of the limbs are often missing. The liver is the most frequently affected abdominal organ (89% of cases) due to its location. If the head is absent, malformations of the pelvic organs are less severe and the skeleton has more bones.[12]

The series of cases presented in the literature most often describe a short-umbilical cord with one umbilical artery in the acardiac twin.[13] The three cases presented in this paper were concordant with the above-described characteristics.

Although, the literature most often reports the acardiac twin as being of the female gender, in all cases depicted here, these fetuses were male.

Blood returns to the pump fetus via venovenous anastomoses causing chronic hypoxia, intrauterine growth retardation, cardiomegaly, heart failure and hydramnios.[14]

In our series, only one case had a delivery near term with no signs of chronic intrauterine fetal hypoxia.

Hydramnios was present in the first case of our series and was not accompanied by maternal symptoms that imposed evacuatory amniocentesis.

Although, the literature reports a 90% frequency of fetal chromosomal anomalies in all the cases we followed, the fetal karyotype was normal.

Fusi reported one twin pregnancy case (acardiac fetus and fetal demise in the 24th gestational week) in which the umbilical cord of the acardiac fetus was wrapped around the leg of the pump fetus. The acardiac fetus had thrombi in the lungs, kidneys and liver. Therefore, acardia was thought to be the consequence of a localized vascular event (thrombotic or thromboembolic) of the thoracic artery or abdominal vessels followed by resorption of the affected tissues.[15]

Also, the placenta of the acardiac fetus reported by Steffensen showed villus immaturity (described either in TTTS) with extended villus calcifications that confirmed the absence of circulation and a nonfunctioning placenta in the acardiac twin. Villus capillaries presented nucleated erythrocytes caused by chronic hypoxia in the pump fetus due to blood from the umbilical vein being mixed with blood from the acardiac fetus.[16]

Fetal thrombotic vasculopathy, which further decreases the blood supply in the pump fetus, was also described.

From the few data existing in the literature that reveals the presence of thrombi in the chorial plaque and umbilical vein of the pump fetus, we may conclude that a possible thrombembolic event may be responsible for the TRAP sequence.[17-19]

The final diagnosis for a twin pregnancy with an acardiac twin may be established after the 9th week when the cephalic extremity and limbs can be precisely determined.

The lack of cardiac activity and the presence of flow in the umbilical cord have been cited both in monofetal and multiple pregnancies with triplets.

Diagnosis errors may occur when the acardiac fetus is adherent to the placenta; in that case, the diagnosis will be ascertained only at birth. When the acardiac structure is initially interpreted as intrauterine fetal death, the diagnosis may be amended at following ultrasound exams when one notices that the cellular mass has increased in volume.

Direct signs that lead to the diagnosis of an acardiac twin are the absence of cardiac and fetal movements, difficult visualization of the trunk and cephalic poles, thickening of subcutaneous tissue and the presence of umbilical artery reverse-flow towards the acardiac fetus mass. The pump fetus may demonstrate hydramnios, cardiomegaly and heart failure.

The differential diagnosis must eliminate a possible arrest in evolution of one fetus from a twin pregnancy as well as anencephaly.

Once an acardiac anomaly is diagnosed, both the acardiac and pump twins should be assessed to classify the pregnancy according to prognostic factors based on the size and growth of the acardiac twin and the cardiovascular condition of the pump twin.

There are two available options, either conservative or interventional, for the management of these pregnancies.

The following parameters must be considered:

1. Amnionicity—in cases of diamniotic monochorionic pregnancies, TRAP treatment does not reduce the risk of monoamnionicity. Umbilical cord occlusion and cutting are required in monochorionic or pseudomonochorionic pregnancies in order to avoid death of the pump twin.
2. Assessment of pump twin morphology.
3. Assessment of the pump twin condition (adequate cardiac function).
4. Size of the acardiac twin and cord and their relationship with the pump twin in order to select the most appropriate therapy. Some argue that if the estimated weight of the acardiac twin is below one-quarter of the pump twin's weight, the prognosis is excellent without treatment.[20]

Conservative management is indicated in cases where the acardiac twin is small and when there are no signs of cardiovascular impairment in the pump twin or in cases with spontaneous shunt closure due to tight cord wrapping.

The aim of conservative management is to monitor the pump twin at risk of developing heart failure associated with polyhydramnios and premature birth (cord entanglement was also mentioned).[21]

Follow-up is recommended every 2 weeks for ultrasound examination, fetal echocardiography, NST, biophysical score, Doppler ultrasound and a calculation of the ratio of estimated acardiac/pump fetus weight. In cases of a decreased ratio, expectant management is appropriate, and in cases of an increased ratio, delivery must be induced.[22]

Simpson suggested that inotropic drugs (Digoxin) should be administered when heart failure occurs, while indomethacin and an amniocentesis should be used for reducing the amount of amniotic fluid.[23]

Today, most departments choose active management. The main objective of invasive procedures is to stop the supply of blood from the pump fetus towards the acardiac fetus.

Serial ultrasound surveillance is important for detecting any worsening of the condition, which may suggest the need for interventions to optimize the pump-twin's chance for survival.

Vessel thrombosis under ultrasound guidance using thrombogenic materials, alcohol and sutures soaked in alcohol has a higher risk of possible racanalization of the thrombosed vessels and embolus migration towards the normal fetus resulting in the death of both fetuses.[24]

In 1994, Quintero was the first to report umbilical cord ligation by fetoscopy with a success rate of 70 to 80%, a 10% risk of premature rupture of the membranes and a significant risk of bleeding.[24]

In cases of a large acardiac twin or the rapid growth of an acardiac mass, treatment is indicated. An intrafetal approach used to interrupt the vascular supply to the acardiac twin appears to be superior to cord occlusion techniques.[7]

In 2003, Tan and Sepulveda concluded that elective treatment can include intrafetal ablation, which is simpler, safer and more efficient than cord occlusion.[25]

The first line treatment should be endoscopic coagulation of vessels using a Nd: YAG laser during mid-pregnancy, radiofrequency ablation under ultrasound guidance and thermal coagulation (acardiac 10).

If these methods are not available, however, chemosclerosis of the intrafetal arterial vessel with alcohol should be attempted.[26,27]

In conclusion, the pump fetus must be treated; otherwise it may die in 50 to 75% of cases due to cardiac strain.

Assessing the extent of heart damage in the pump fetus is a key step in pregnancy management.

Other negative prognostic factors include an acardiac twin/pump twin weight ratio > 0.7 and the presence of upper limbs, as was shown in two of our cases.[21,28]

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