Fetal echopsy (ultrasonographic autopsy) of an acardius myelancephalus and its correlation with antenatal ultrasonographic findings

Balakumar Karippaliyil, Milind Karippaliyil, Misha Karippaliyil
Department of Ultrasonography, Balku’s Scan, PVS Hospital, Calicut, Kerala, India

Correspondence: Dr. Karippaliyil Balakumar, Balku’s Scan, PVS Hospital, Calicut - 2, Kerala - 673 002, India. E-mail: balakumar_k@dataone.in

Abstract

Aberrant transplacental arteriovenous shunts between the placental and cord vessels of monozygotic monoamniotic twins or triplets result in the formation of an acardius. The prenatal diagnosis of this condition has been reported occasionally in the literature. A subtype categorized as acardius myelancephalus was diagnosed at 32 weeks of gestation by ultrasonography (USG). The pregnancy was aborted because of poor prognostic predictors and the acardius was subjected to ultrasonographic autopsy (fetal echopsy). The antenatal USG features were correlated with echopsy findings for confirmation of the antenatal findings and for a better visual perception of the prenatal diagnostic features. The echopsy revealed more precise details. Fetal echopsy avoids the medicolegal problems concerned with parental consent for classical invasive fetal autopsy.

Key words: Acardius myelancephalus; fetal echopsy; fetal ultrasonographic autopsy; TRAP sequence; twin pregnancy; ultrasonography

Introduction

Multiple pregnancies have more maternal and fetal complications. One such severe manifestation, twin-to-twin transfusion syndrome (TTTS) leads to twin reversed arterial perfusion sequence (TRAP sequence) resulting in an acardius. It is very rarely reported in triplet and even in quintuplet pregnancies. A rare subtype of acardius, termed acardius myelancephalus, was diagnosed at 32 weeks of gestation and therapeutically aborted because of progressive hydrops of the “pump” co-twin. The amorphous fetus was subjected to echopsy to correlate the findings with antenatal USG features. The fetal echopsy revealed more definite and conclusive structural details without the help of actual invasive autopsy, establishing its role for academic purposes. There are no reports on echopsy of acardius myelancephalus till date, to the best of our knowledge.

Case Report

A primigravida scanned at 9 weeks of gestation was reported to have monochorionic monoamniotic twins. She was referred at 32 weeks of gestation without follow-up USG scans to assess the fetal growth. The scanning was done with a high-density (HD) convex probe of 2.5-5 MHz (Aloka Prosound 5000 machine). The USG showed a normal fetus with normal activities. The co-twin was represented by an amorphous soft tissue mass with no cephalic or caudal structures, though connected by the three-vessel umbilical cord. The normal intrathoracic or intra-abdominal structural details were not defined within the abnormal fetal mass. A deformed rudimentary upper limb showed only the radius and ulna connected to rudimentary carpals, metacarpals, and two digits [Figure 1A]. Three
This pregnancy was aborted since the normally appearing “pump twin” developed progressive hydrops. The acardius myelancephalus was subjected to echopsy after immersing in a trough of water. This simple technique provided clear and finer details of this fetus with a 2.5-5 MHz HD convex probe. The fetus was represented by an amorphous mass covered with normal skin [Figure 2A]. There was total absence of cephalo-caudal differentiation [Figure 2B]. The left upper limb represented by the radius and the ulna was connected distally to the deformed hand with rudimentary carpals and metacarpals connected to thumb and index finger. Other digits were absent [Figure 3A]. The left humerus was not defined and there was no representation of the limb girdles or right upper limb. The cervical vertebrae were absent. Few small calcified shadows arranged in a linear fashion appeared like rudimentary thoracic vertebrae by their position. The single largest bone within the lower torso near the umbilical cord insertion represented one femur connected distally with few calcified shadows showing no characteristic features. This femur was surrounded by the amorphous soft tissue mass [Figure 3B]. The head, heart, and intra-abdominal visceral structures were absent, but for a central echogenic shadow within the tubular structures showing features of a bowel loop segment [Figure 3C]. The soft tissue projections near the umbilical cord represented the edematous empty scrotal sacs and penis [Figure 3D].

Discussion

Acardius results from TRAP sequence which is a severe form of TTTS. It has an incidence of 1 in 35,000 pregnancies and 0.3-1% of monozygotic twins. This is due to abnormal transplacental arterio-arterial and arterio-venous shunts. The reversed perfusion through the umbilical arteries and more hypoxic flow of “twice” used blood through the vein (usually they have single umbilical artery) of an acardiac twin causes poor oxygenation of the upper body organs. The larger normal pump twin within the polyhydramniotic sac is prone to high output cardiac failure and hydrops, with a mortality of 50-70%. The increased cardiac load on the normal pump twin leads to high perinatal mortality. Serial scanning is indicated for earlier detection of these changes with the help of 2D/3D scans supplemented with Doppler studies of the umbilical arteries for prognostication. The weight of an acardius may be calculated using the formula: Weight in grams = (1.2 × longest length)-(1.7 × longest length), which can also be used for predicting the outcome. As this condition is sporadic, karyotyping is not usually indicated.
Among the various subtypes of acardiac fetuses (acardius acephalus, acardius aniceps, acardius amorphus, acardius myelancephalus, and acardius acormus), acardius amorphus is rare. The fetus reported here with features of acardius amorphus and presence of rudimentary limb was antenatally diagnosed as acardius myelancephalus. This pregnancy was terminated since the “pump” twin developed severe degree of progressive hydrops and the “recipient” had all lethal features of an acardius. However, the major findings as well as the finer details were confirmed by fetal echopsy.

The invasive fetal autopsy is unavoidable for both cytogenetic evaluation and a better understanding of the structural abnormalities. In our present social setup, a classical fetal autopsy invites many legal formalities and serious litigations later. The rate of fetal autopsy is also getting reduced these days. The usefulness of USG as an alternative non-invasive autopsy tool was suggested as early as 1989. Later, a group of authors proposed a reliable USG-guided needle autopsy instead of the classical adult autopsy to overcome a similar situation and termed it as ultrasonographic autopsy (echopsy). Virtual non-invasive fetal autopsy using conventional radiography, computed tomography (CT), or magnetic resonance imaging (MRI) was suggested by different authors.

We illustrate the same non-invasive principle applied to fetal autopsy using USG (and thus termed fetal echopsy) where a morphological confirmation of the antenatally diagnosed anomalies is possible. This will avoid the procedural obstacles for getting the parental consent for a fetal autopsy and the litigations following it, thus improving the visual perception and confirmation of an anomaly and enhancing the research methodology.

References