

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

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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

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small soft tissue projections near the umbilical cord insertion could not be characterized. A well-formed femur of 61 mm (corresponding to 31 weeks) was seen continuous with specks of ossified structures representing the distal limb bones [Figure 1B]. These structures were surrounded by the thick edematous amorphous soft tissue mass [Figure 1C and D]. There were some specks of linear calcification within the central region of the amorphous fetal mass which appeared like vertebral remnants. The central region of the torso also showed echogenic shadow inside ill-defined tubular structures. Doppler insonation of the umbilical vessels of this fetal mass showed reversed flow directions. All these features were diagnostic of acardius myelancephalus.

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

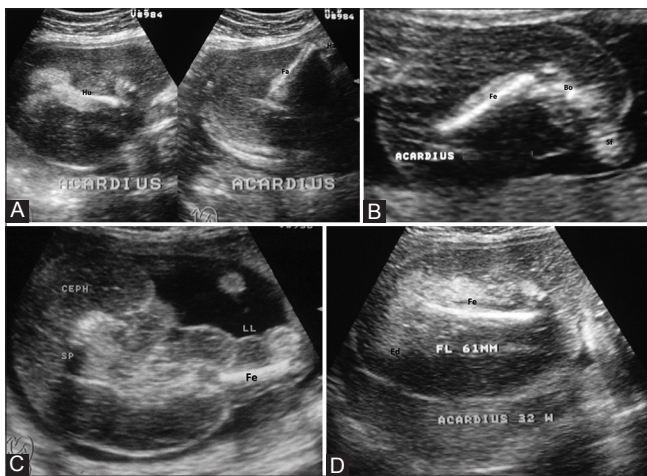


Figure 1 (A-D): (A) The antenatal USG of the acardius myelancephalus: The left humerus (Hu) and forearm bones (Fa) are distally connected to the malformed carpals and metacarpals (Ha), surrounded by edematous soft tissue. Left upper limb girdle bones or right upper limb bones are not seen. (B) The lower limb bone along its whole length. The long bone at the caudal region near the umbilical cord insertion is the femur (Fe), connected to specks of bones (Bo) representing the rudimentary leg bones. A small soft tissue protrusion (Sf) near it could not be characterized. (C) An oblique section through the torso: The cephalic end (CEPH) is indistinguishable and specks of bone represent the vertebral remnants (SP). The femur (Fe) is surrounded by the amorphous soft tissue (LL). (D) The full-length view of the femur. The normally calcified femur (Fe) corresponds to 31 weeks growth within the soft tissue mass (Ed)

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.^[1,2] 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%.^[3,4] The increased cardiac load on the normal pump twin leads to high perinatal mortality.^[5] 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.^[6] The weight of an acardius may be calculated using the formula: Weight in grams = $(1.2 \times \text{longest length}^2) - (1.7 \times \text{longest length})$, which can also be used for predicting the outcome.^[7] As this condition is sporadic, karyotyping is not usually indicated.

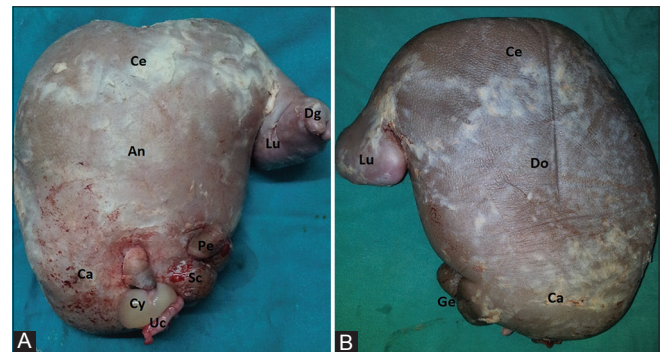


Figure 2 (A and B): (A) The external appearance of the aborted acardius myelancephalus. The ventral surface (An) shows normal skin and the cephalic (Cr) and caudal (Ca) ends are made out by the positions of the left upper limb ((Lu), digits (Dg), umbilical cord (Uc), and the genitalia (SC -edematous scrotal sacs, Pe-edematous penis). A cyst (Cy) is seen closer to the cord insertion. (B) Dorsal surface of the acardius. The dorsal skin surface (Do) is normal. The left upper limb (Lu) and the genitalia (Ge) are partially visible from behind (Ce-cephalic end, Ca-caudal end)

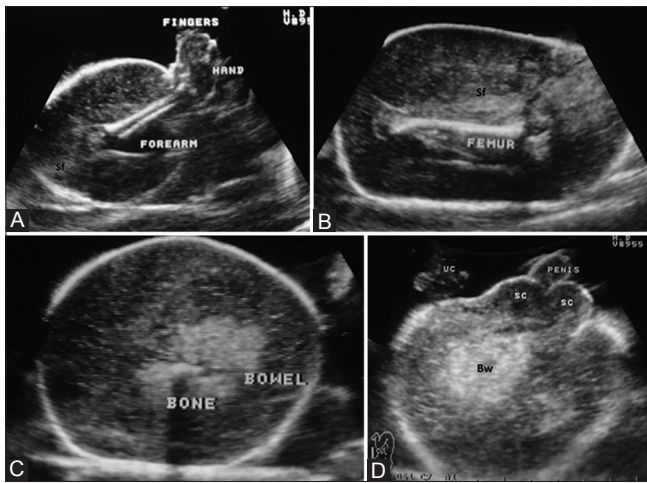


Figure 3 (A-D): (A) Echopsy of the aborted acardius myelancephalus after immersing it in water. The section at upper torso shows the left forearm bones surrounded by soft tissue (Sf) and the distal deformed hand and rudimentary fingers (thumb and index). (B) Echopsy showing femoral details. The full length of the femur is seen embedded in the soft tissue. (C) Echopsy showing the bowel loops. An axial section shows the cross-sectional view of the femur (bone) and part of more echogenic bowel loops (bowel). (D) The details of the genitalia by echopsy. A slightly oblique axial section at the level of the genitalia shows the more echogenic bowel loops (Bw), edematous empty scrotal sacs (SC), and penis. Part of the umbilical cord (UC) is also seen

Among the various subtypes of acardiac fetuses (acardius acephalus, acardius anceps, 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.^[8,9] The usefulness of USG as an alternative non-invasive autopsy tool was suggested as early as 1989.^[10] 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).^[11] Virtual non-invasive fetal autopsy using conventional radiography, computed tomography (CT), or magnetic resonance imaging (MRI) was suggested by different

authors.^[12,13]

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.

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