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Dichorionic, diamnionic twin pregnancy discordant for anencephaly: Report of two cases and literature review

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Anencephaly is a lethal diagnosis. In the unique situation of a twin pregnancy discordant for anencephaly, early ultrasound diagnosis based on the discrepancy in the appearance of the heads can facilitate management and potentially decrease morbidity and mortality for the unaffected twin. We report two such cases of dichorionic, diamniotic twin pregnancies and provide a review of the literature.

Case report 1

A 20-year-old primagravida at 11 + 1 weeks (by last menstrual period) presented to the emergency room with vaginal bleeding.

Ultrasound evaluation showed a live dichorionic, diamniotic intrauterine twin pregnancy with discrepancy in both crown rump lengths and the appearance of the heads of the twins. Twin A appeared normal, with an average crown rump length (CRL) of 38 mm (estimated gestational age [EGA], 10+5 weeks). Anencephaly was suspected in twin B, based on the average CRL of 31 mm and nonvisualization of the calvarium, which in contrast was clearly visualized in twin A (Fig. 1A). Sonography performed in a dedicated Fetal Diagnostic Center four days later confirmed the initial findings. No fetal abnormalities were identified in twin A on this or subsequent examinations. In fetus B, the structures of the upper head were absent, possibly representing anencephaly or acrania. Genetic testing and selec-

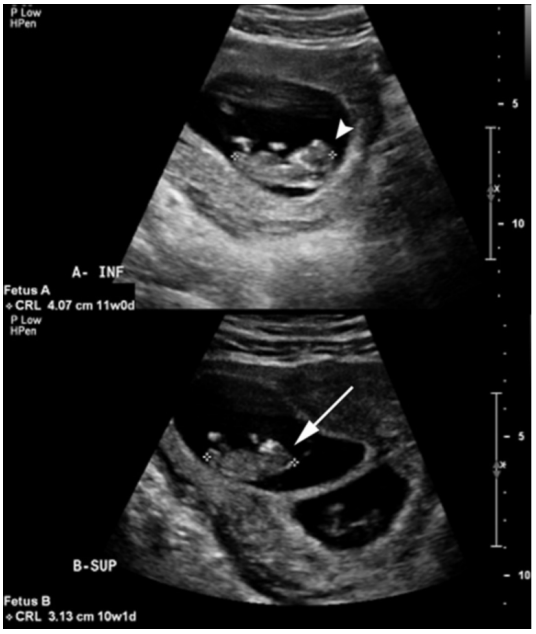

Fig. 1A. Grayscale sonographic findings at 11 + 1 weeks in a dichorionic, diamniotic twin pregnancy discordant for anencephaly that was incidentally diagnosed during an ER visit for vaginal bleeding (Case 1). Longitudinal image (top) through the head of normal twin "A" demonstrates a normal bony calvarium (arrowhead). In contrast, longitudinal image (bottom) through the head of twin "B" demonstrates absence of cranial structures above the level of the orbits (arrow). No bony calvarium is visualized. These findings can be seen in acrania or anencephaly.

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tive fetal reduction options were offered and declined.

At 18 + 0 weeks, anencephaly in twin B was confirmed by the absence of calvarium and brain tissue above the level of the orbits (Fig. 1B). Additional findings included bilateral cleft lip and clubbed feet, which increased the level of concern for an associated chromosomal abnormality.

On subsequent studies, the estimated fetal weight of twin

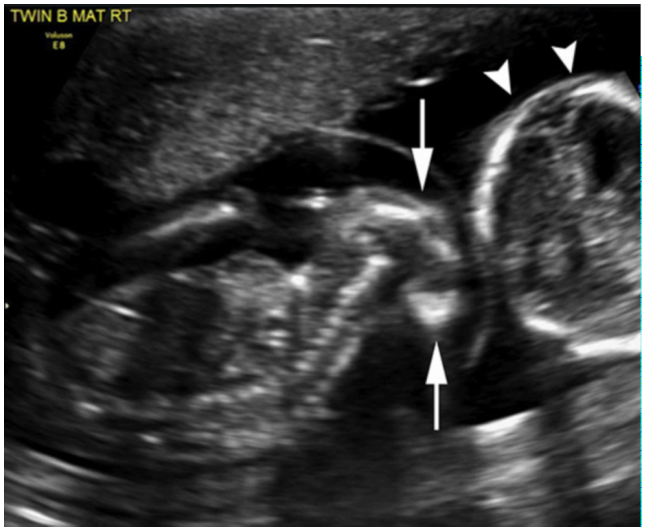

Figure 1B. Grayscale sonographic findings at 11 + 1 weeks in a dichorionic, diamniotic twin pregnancy discordant for anencephaly that was incidentally diagnosed during an ER visit for vaginal bleeding (Case 1). Sonographic grayscale image of twin "B" (Case 1) at 18 + 0 weeks demonstrates absence of structures of the upper head above the level of the brainstem, with no upper skull or brain tissue present (between arrows). Note the presence of normal cranial structures and brain on twin “A” (arrowheads).

A progressively dropped from the 16th percentile (at 24 + 5 weeks) to less than the fifth percentile (on sonography performed at 36 + 6 weeks). Polyhydramnios was noted for twin B at 29 + 5 weeks. Therapeutic amniocentesis was discussed with the couple at 33 + 5 weeks but was declined.

Fetus A was in the breech position on all followup studies from 29 + 5 weeks onward. At the 36 + 6 weeks followup, cesarean delivery was recommended due to prolonged severe fetal growth restriction (FGR) and persistent breech presentation of fetus A. Both twins were delivered via C-section at 37+1 weeks.

At birth, twin A weighed 2180 grams, with a normal physical exam. Twin B was anencephalic and weighed 1340 grams. Additional physical exam findings for twin B included various deformities of the bilateral lower extremities and undescended testes. Apgar scores for twin A were 9 and 9 at 1 and 5 minutes. Apgar scores for twin B were 4 and 4 at 1 and 5 minutes.

Twin B expired 11 hours after birth. Autopsy and genetic analysis of twin B were declined. Twin A was discharged home from the hospital at age 2 days.

Case report 2

A 31-year-old primigravida with twin pregnancy at 11 weeks EGA (based on intrauterine insemination date) presented to the Fetal Diagnostic Center for ultrasound evaluation of chorionicity, viability, and growth in the setting of maternal vaginal bleeding.

Ultrasound showed a live dichorionic, diamniotic intrauterine twin pregnancy. The average CRL of twin A was 35 mm, corresponding to an EGA of 10 weeks 3 days. Acrania of twin A was described, based on absence of cranial vault and presence of exposed brain tissue protruding above the level of the fetal orbits. Twin A was also noted to have an omphalocele. The average CRL of twin B was 50 mm, corresponding to an EGA of 11 weeks 5 days (Fig. 2). No fetal abnormalities were identified in twin B at this exam or subsequent exams. A short and funneled-appearing cervix was noted, measuring 1.7–2.3 cm in length. The patient declined both genetic testing and selective reduction of twin A.

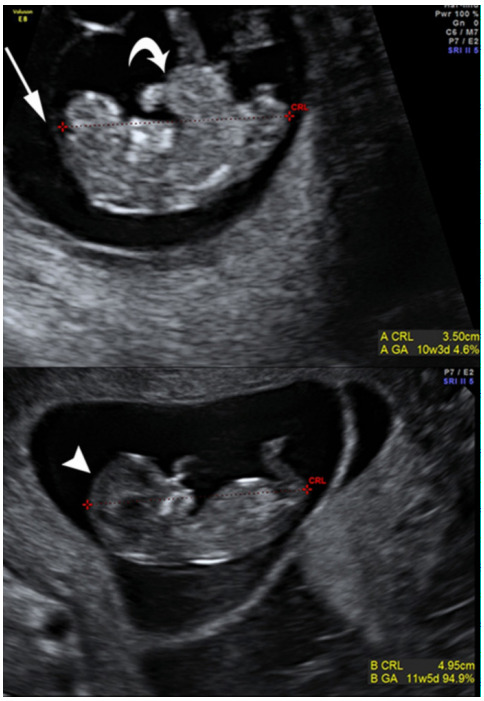

Figure 2. Grayscale sonographic findings at approximately 11 weeks in a dichorionic, diamniotic twin pregnancy discordant for anencephaly (Case 2). Longitudinal image (top) through the head of affected twin "A" demonstrates absence of a normal bony calvarium (straight arrow), which was most suspicious for acrania or anencephaly. In addition, an omphalocele (curved arrow) is noted in twin “A”. In contrast, longitudinal image (bottom) through the head of twin "B" demonstrates normal bony calvarium (arrowhead).

Ultrasound findings were unchanged at the 12 + 1 weeks' followup exam. At 14 + 2 weeks, the demise of twin A was noted. The cervical length remained stable (in the 1.2–1.6 cm range) at serial ultrasound exams between 12 + 1 weeks and 16 + 3 weeks, and light vaginal bleeding persisted. At the followup 18 + 2 weeks' ultrasound exam, twin A was visualized in the endocervical canal. The cervix appeared 2 cm dilated at subsequent vaginal speculum exam, with membranes visible at the level of the external os. After discussion of options, the patient chose expectant management. She passed twin A at home at 19 + 2 weeks. Vaginal spotting persisted, and she opted to continue expectant management.

At 21 + 1 weeks, the patient presented to the hospital with uterine contractions and increased vaginal bleeding, and her cervix was found to be 4 cm dilated. She delivered twin B vaginally. Birthweight was 410 grams. Apgar scores were 1 and 1 at 1 minute and 5 minutes. Neonatal demise occurred 10 minutes after birth.

Discussion

The absence of the cranial vault and cerebral hemispheres, hallmarks of anencephaly, can theoretically be distinguished as early as at 9 weeks' gestation, when the skeletogenous layer around the brain begins to develop (1). The diagnosis of anencephaly can be reliably made at the routine 10- to 14-week sonogram in singleton pregnancies if there is a dedicated search for these findings (2). In early cases of anencephaly, the only finding may be acrania. As development progresses, the preserved mid-brain, brainstem, and facial structures become apparent.

In the cases presented, the first-trimester diagnoses of anencephaly were facilitated by early discordant ultrasound findings in twin gestations. The early diagnosis of anencephaly before 16 weeks' gestational age in the specific situation of a twin may decrease morbidity and mortality to the unaffected, normal twin by providing the option of selective fetal reduction at an optimal gestational age (3). A recent systematic review of pooled case reports of twin pregnancies discordant for anencephaly found that selective feticide resulted in longer gestations and higher birth weights for the unaffected twin (4). A multicenter trial of selective termination with potassium chloride injection of the anencephalic twin showed a risk of miscarriage of 5% to the normal twin when performed before 16 weeks' gestation, compared with a miscarriage rate of 14% when performed after 16 weeks (5).

Polyhydramnios is a common complication of anencephalic gestations and has been reported in up to 57% of dichorionic twin pregnancies discordant for anencephaly; serial ultrasound examinations may help minimize the risk of miscarriage by guiding therapeutic amnioreduction after 24 weeks (6). The benefit of amnioreduction is unclear, however, with one series reporting no miscarriages in 14 cases of twin pregnancies discordant for anencephaly (mean gestational age at delivery of 35.9 weeks) despite a 43% rate of polyhydramnios (6 of 14) (7).

While anencephaly is a lethal defect and could be undiagnosed in early pregnancies that abort spontaneously, the diagnosis remains clinically important in periconceptional planning for the pregnant patient. In the setting of twins discordant for anencephaly, the early discordance of ultrasound findings can facilitate early diagnosis as well as management for the unaffected twin.

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