Rare Discordant Genetic and Structural Anomaly in Monochorionic Twins – A Challenging Approach

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Section 1 – Quiz

A 34-year-old primigravida was referred to our outpatient clinic at 13 weeks of gestation due to a twin pregnancy with discordant fetal malformation, detected in the first-trimester ultrasound. The first-trimester ultrasound revealed a diamniotic monochorionic pregnancy [Figure 1]. Twin A had a crown-rump length (CRL) of 65.45 mm (gestational age of 12 weeks^{+6}) [Figure 2], nuchal translucency (NT) of 1.8 mm (<95th centile), presence of nasal bone, and normal ductus venosus and tricuspid flow. The remaining fetal anatomy assessment revealed no structural anomalies and normal amniotic fluid. Twin B had a CRL of 54.09 mm (gestational age of 12 weeks^{+0}), which corresponds to 17% CRL discordance [Figure 3], and a NT of 7.1 mm (>99th centile), which corresponds to 75% NT discordance, with septated structures, compatible with suspected nuchal cystic hygroma [Figures 4 and 5]. It also showed absent nasal bone, poorly filled bladder, and diminished amniotic fluid, and bilateral renal agenesis was suspected. Evaluation of the heart, situs, four-chamber view, and tricuspid flow was not possible due to technical difficulty. Ductus venosus flow was normal. The placenta was anterior, and both umbilical cords had three vessels.

Combined first-trimester screening revealed an increased average risk for trisomy 21 (1:102) and a low risk for trisomies 18 (1:6384) and 13 (1:6331), pregnancy-associated plasma protein-A of 0.63 MoM, and free beta-human chorionic gonadotropin of 1.44 MoM.

In a subsequent ultrasound, at 16 weeks gestation, the following anomalies were identified in twin B: occipital encephalocele, hydrocephalus, bilateral renal dysplasia,

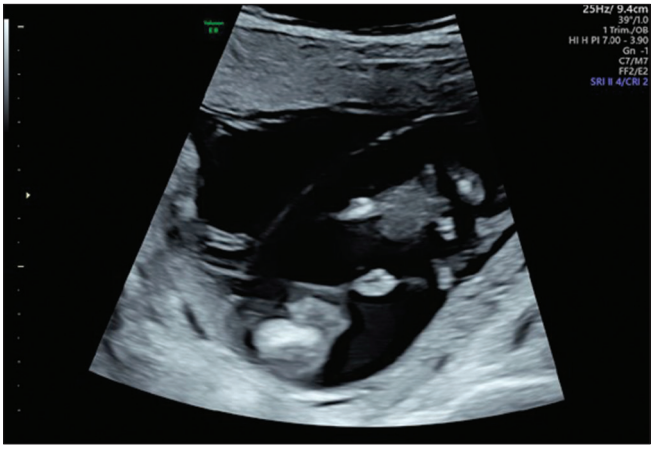

Figure 1: First-trimester ultrasound: chorionicity determination at 12 weeks of gestation, where a T-sign and a single placental mass may be observed, compatible with monochorionic diamniotic twin pregnancy

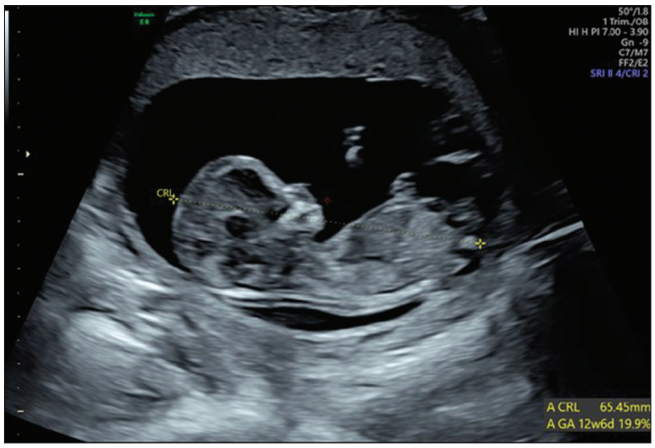

Figure 2: First-trimester ultrasound of twin A: mid-sagittal plane showing crown-rump length (CRL) measurement of 65.45mm

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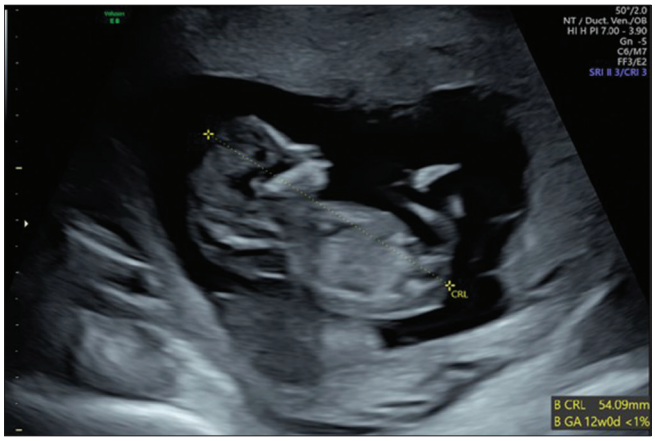

Figure 3: First-trimester ultrasound of twin B: mid-sagittal plane showing crown-rump length (CRL) measurement of 54.09mm, revealing a 17% discordance of the twins' CRL

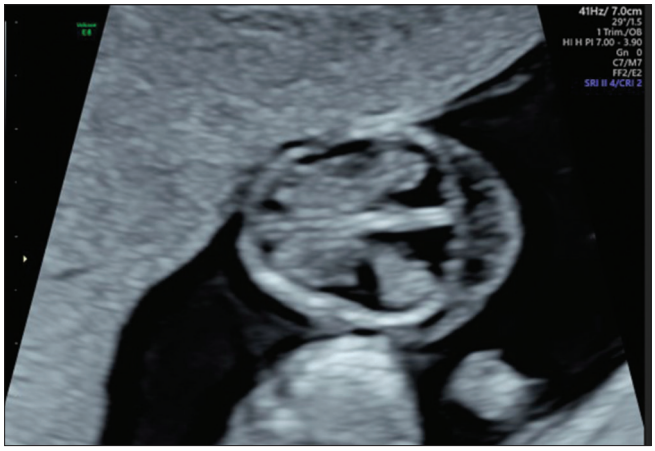

Figure 5: First-trimester ultrasound: nuchal translucency evaluation (axial plane) of twin B, which was above the 99th centile and showed septated internal structures, suggesting cystic hygroma

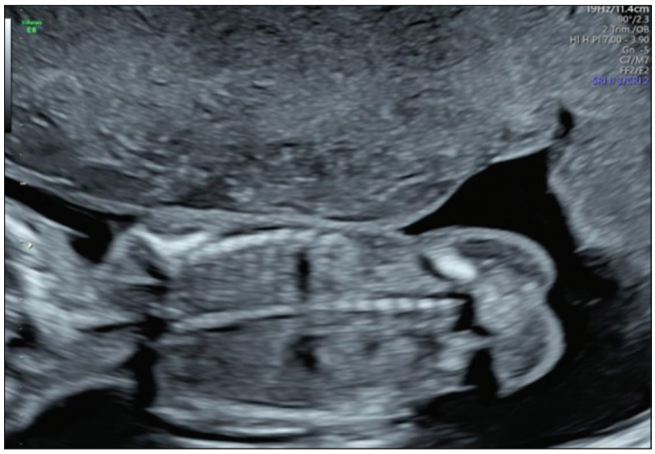

Figure 7: Second-trimester ultrasound of twin B (coronal plane), showing bilateral renal dysplasia

bladder, and stomach bubble were poorly filled, and oligohydramnios [Figures 6 and 7]. Twin A had a normal basic anatomy assessment, namely, the skull, brain, face,

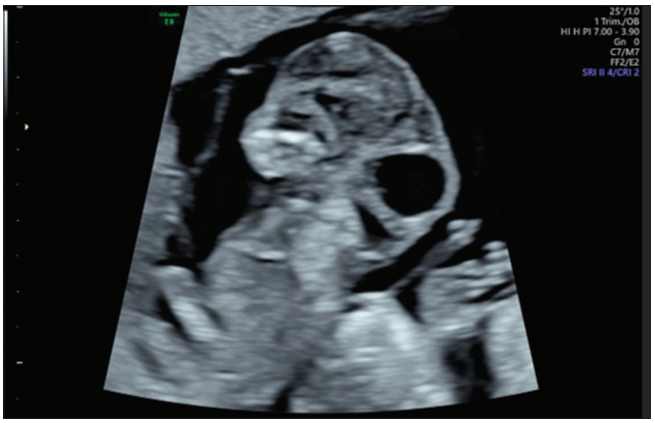

Figure 4: First-trimester ultrasound: nuchal translucency evaluation (mid-sagittal plane) of twin B, which was above the 99th centile and showed septated internal structures, suggesting cystic hygroma

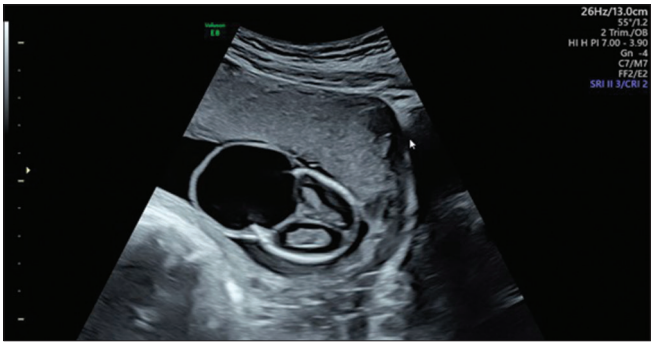

Figure 6: Second-trimester ultrasound of twin B (axial plane), showing occipital encephalocele and hydrocephalus

heart, abdomen, kidneys and bladder, spine, and limbs. Both fetuses had normal female genitalia. Fetal growth assessment was concordant with gestational age for twin A; however, there was a growth discordance of 24%. Twin A amniotic fluid was normal.

Invasive testing was offered, the couple agreed, and amniocentesis was performed at 17 weeks, sampling both amniotic sacs individually. The couple also agreed on the genetic evaluation of both parents.

After a discussion of the probable diagnosis and the likely prognosis for both the affected and the normal twin with fetal medicine experts, the couple decided on conservative management and refused selective termination of the affected twin.

At 20 weeks, the cardiac screening assessment of twin A was normal, and dextrocardia and interventricular communication were detected in twin B [Figure 8]. Twin B hydrocephalus and occipital encephalocele were progressively increasing in severity [Figures 9 and 10], and bilateral postaxial polydactyly was identified [Figures 11 and 12]. At this scan, growth discordance was 26%. Twin A amniotic fluid was normal, and twin B had oligohydramnios. The umbilical artery, ductus venosus, and middle cerebral artery peak systolic velocity flow were normal for both fetuses. Cervical assessment revealed a normal cervical length.

At 23 weeks, ventriculomegaly was identified in twin B, with a posterior atrium measuring 17 mm, and encephalocele progressed in severity on subsequent scans [Figures 13 and 14]. No additional sonographic anomalies were identified, and sequential fetal Doppler evaluations remained normal.

At 27 weeks, twin A was in cephalic presentation and had an estimated fetal weight (EFW) of 1249 g (87th centile). Fetal Doppler and biophysical profile were normal. Twin B was in breech presentation, had an EFW of 801 g (6th centile), with a growth discordance of 36%, and had oligohydramnios. Fetal Doppler was normal.

At 28 weeks of gestation, the patient was admitted to the hospital due to preterm prelabor rupture of membranes. The cervical length was 10 mm, with cervical funneling. Fetal well-being assessment was normal for both fetuses. Fetal lung maturation and tocolysis were administered after excluding the signs of maternal infection. Prophylactic antibiotic therapy was started, and after a 2-week latency period, during which fetal well-being was assessed through daily cardiotocography and weekly biophysical profile, the patient had an eutocic delivery at 30 weeks of gestation. The first newborn was twin A, with a birth weight of 1670 g and an Apgar score of 6 and 8 on the 1st and 5th min, respectively. The second newborn was twin B, with a birth weight of 1200 g and an Apgar score of 1 and 1 on the 1st and 5th min, respectively. She died moments after birth. A fetal autopsy was refused by the couple. At 6, 12, and 18 months of age, the surviving infant presented normal psychomotor and overall development.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent form. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published, and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

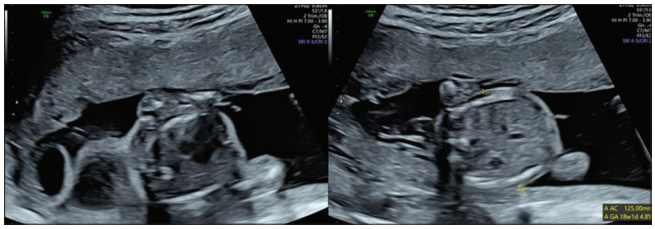

Figure 8: Second-trimester ultrasound showing dextrocardia in twin B

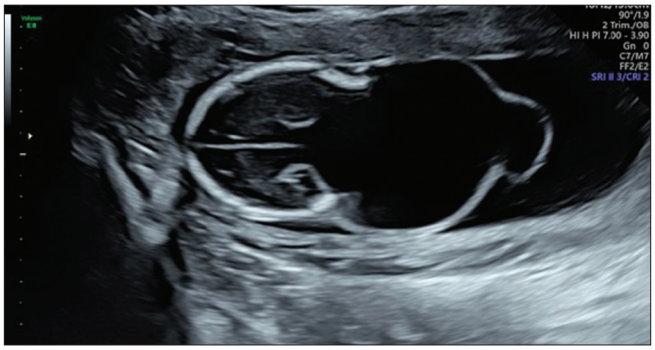

Figure 9: Second-trimester ultrasound of twin B (axial plane), showing progressively severe encephalocele at 18 weeks of gestation

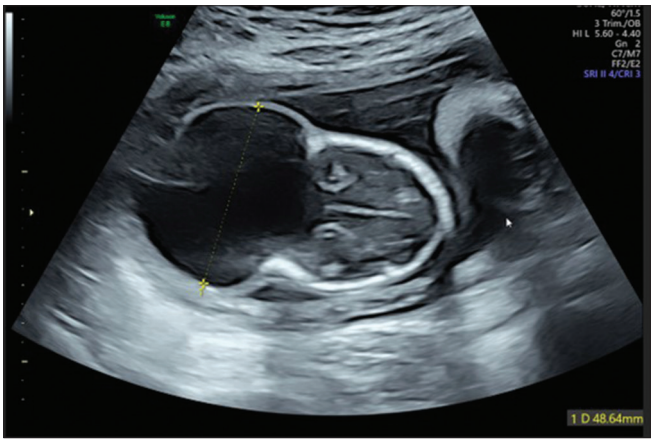

Figure 10: Second-trimester ultrasound of twin B (axial plane), showing progressively severe encephalocele at 19 weeks of gestation

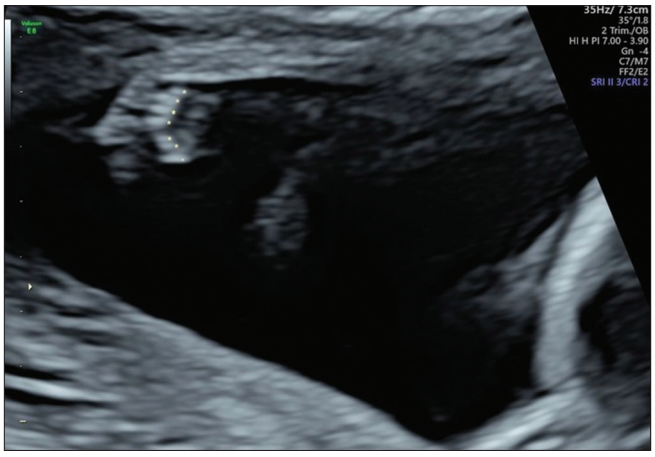

Figure 11: Second-trimester ultrasound of twin B, showing foot postaxial polydactyly

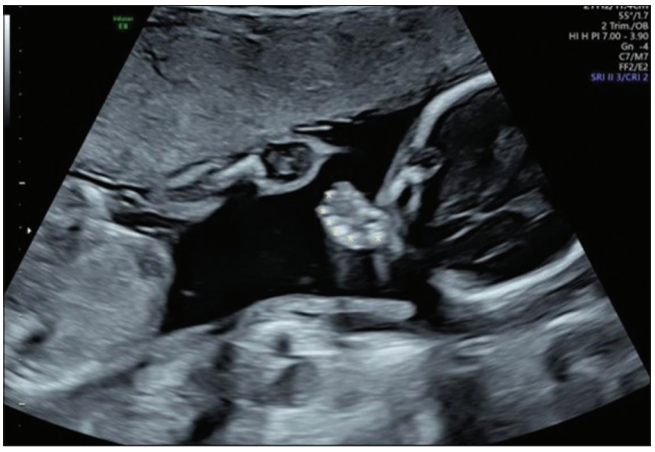

Figure 12: Second-trimester ultrasound of twin B, showing hand postaxial polydactyly

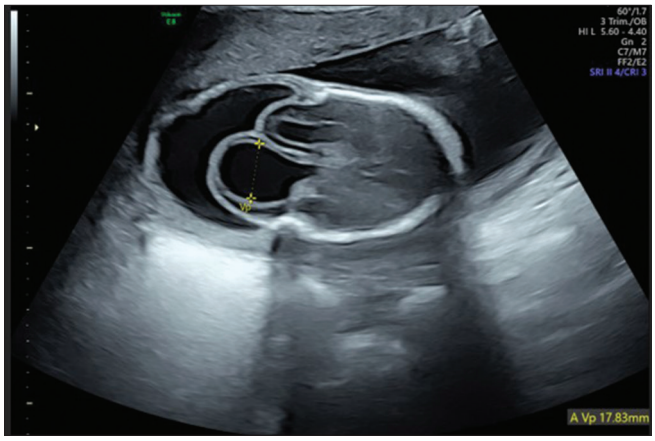

Figure 13: Second-trimester ultrasound of twin B (axial plane), showing posterior atrium measuring 17.83 mm at 24 weeks, compatible with ventriculomegaly

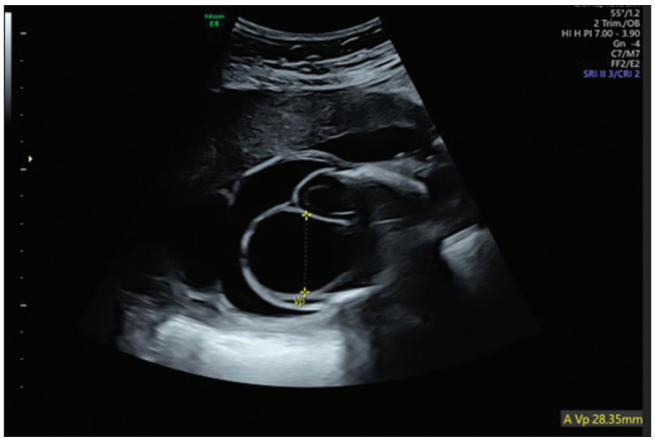

Figure 14: Second-trimester ultrasound of twin B (axial plane), showing posterior atrium measuring 28.35 mm at 25 weeks of gestation, suggesting progressively severe encephalocele

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Conflicts of interest

There are no conflicts of interest.