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Prenatal Ultrasonographic Diagnosis of Ectopia Cordis, Membrane-Covered Omphalocele, Posterior Encephalocele, and Unilateral Microphthalmia in a 26-Week Fetus: A Case Report

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ABSTRACT

Multiple congenital fetal anomalies involving the anterior thoracoabdominal wall, cranial vault, and ocular structures are rare and are associated with high perinatal mortality and a poor prognosis. Prenatal ultrasonography plays a pivotal role in the early detection of these anomalies, facilitating accurate diagnosis, parental counselling, multidisciplinary management, and informed decision-making, particularly in resource-limited settings. We report the prenatal ultrasonographic diagnosis of multiple severe congenital anomalies in a 32-year-old gravida 4, para 3 woman who underwent a routine second-trimester fetal anomaly scan at 26 weeks' gestation. Two-dimensional ultrasonography demonstrated a live singleton fetus with ectopia cordis, a membrane-covered liver-containing omphalocele, posterior encephalocele, and unilateral microphthalmia. Because the cranial and anterior abdominal wall defects distorted the fetal anatomy, gestational age was determined using the femur length, which measured 45.3 mm, corresponding to 26 weeks' gestation. Spectral Doppler confirmed fetal cardiac activity with a heart rate of 128 beats per minute. The combination of ectopia cordis and omphalocele raised suspicion for an incomplete form of pentalogy of Cantrell, although definitive confirmation was not possible because fetal echocardiography, fetal magnetic resonance imaging, prenatal genetic testing, and postnatal pathological examination were unavailable. Following multidisciplinary counselling regarding the severe congenital anomalies and poor fetal prognosis, the parents elected medical termination of the pregnancy. This case highlights the diagnostic value of meticulous prenatal ultrasonography in identifying complex multisystem congenital anomalies, the importance of selecting unaffected biometric parameters when conventional measurements are unreliable, and the critical role of early diagnosis in facilitating appropriate counselling and individualized pregnancy management in resource-constrained healthcare settings.

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KEYWORDS:
Prenatal ultrasonography, ectopia cordis, omphalocele, encephalocele, microphthalmia, pentalogy of Cantrell.

INTRODUCTION

Congenital anomalies remain an important cause of fetal loss, neonatal mortality, and long-term childhood disability worldwide. The World Health Organization estimates that approximately 240,000 newborns die within the first 28 days of life each year because of congenital disorders, with a disproportionate burden occurring in low- and middle-income countries. Early prenatal identification of major structural abnormalities is therefore essential for accurate diagnosis, prognostic assessment, parental counselling, and appropriate pregnancy management (1).

Prenatal ultrasonography is the primary imaging modality for fetal anatomical assessment because it is safe, non-invasive, widely available, and relatively affordable. The International Society of Ultrasound in Obstetrics and Gynecology recommends systematic assessment of the fetal head, face, spine, thorax, heart, abdominal wall, abdominal organs,

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kidneys, bladder, extremities, placenta, umbilical cord, and amniotic fluid during the routine mid-trimester examination. Detection of one major abnormality should prompt a careful search for additional anomalies and, where available, referral for detailed fetal cardiac assessment and prenatal genetic evaluation (2–4).

Ectopia cordis is an exceptionally rare congenital malformation in which the heart lies partially or completely outside the thoracic cavity because of defective formation and fusion of the ventral body wall. It is frequently accompanied by intracardiac defects and abnormalities of the sternum, diaphragm, pericardium, or anterior abdominal wall (5). Omphalocele is a midline abdominal wall defect in which abdominal contents herniate through the umbilical ring while remaining enclosed within a membranous sac. Its prognosis is strongly influenced by the size and contents of the sac and, more importantly, by associated cardiac, chromosomal, and syndromic abnormalities (6).

The coexistence of ectopia cordis and omphalocele raises suspicion for pentalogy of Cantrell, a spectrum of ventral midline developmental defects classically involving the supraumbilical abdominal wall, lower sternum, anterior diaphragm, diaphragmatic pericardium, and heart (7–10). Additional central nervous system or ocular abnormalities may indicate a broader disturbance of early embryogenesis and should widen the differential diagnosis to include chromosomal disorders and other complex malformation syndromes.

We report the prenatal ultrasonographic diagnosis of ectopia cordis, a membrane-covered liver-containing omphalocele, posterior encephalocele, and unilateral microphthalmia in a live singleton fetus at 26 weeks' gestation. Reports of this particular combination remain scarce. The case highlights the value of a systematic fetal survey, the need to use unaffected biometric parameters when structural abnormalities distort conventional measurements, and the contribution of multidisciplinary counselling to pregnancy management in a resource-constrained setting.

CASE REPORT

A 32-year-old gravida 4, para 3 woman was referred to the Department of Radiology, Federal Teaching Hospital Birnin Kebbi, for a routine second-trimester fetal anomaly scan at 26 weeks' gestation. The pregnancy had been uneventful, and she reported no abdominal pain, vaginal bleeding, leakage of liquor, fever, or reduction in fetal movements before presentation.

Her previous three pregnancies had resulted in uncomplicated term vaginal deliveries of healthy infants. There was no history of recurrent miscarriage, stillbirth, neonatal death, or congenital anomalies. The patient had no history of hypertension, diabetes mellitus, epilepsy, thyroid disease, chronic renal disease, autoimmune disease, or other significant medical illness. She also denied exposure to ionizing radiation, recognized teratogenic medications, alcohol, cigarette smoking, or recreational drugs during the pregnancy. The parents were non-consanguineous, and there was no known family history of congenital malformations or inherited disorders.

The referral was made solely for a routine fetal anatomical survey, and there had been no prior clinical suspicion of fetal structural abnormalities. Maternal physical examination findings and obstetric observations accompanying the referral were unremarkable, although detailed maternal vital signs and obstetric examination findings were not available for review.

TABLE 1: TIMELINE OF EVENTS

TIME	CLINICAL EVENT
PREVIOUS PREGNANCIES	Three uncomplicated term pregnancies resulting in healthy live births. No history of congenital anomalies, recurrent pregnancy loss, stillbirth, or neonatal death.
CURRENT PREGNANCY (FIRST AND SECOND TRIMESTERS)	Routine antenatal care. No documented maternal hypertension, diabetes mellitus, chronic medical illness, teratogenic exposure, alcohol intake, cigarette smoking, consanguinity, or family history of congenital anomalies.
16 JULY 2026 (26 WEEKS' GESTATION)	Routine second-trimester obstetric ultrasonographic examination performed at the Department of Radiology, Federal Teaching Hospital, Birnin Kebbi.
ULTRASOUND FINDINGS	Live singleton fetus with ectopia cordis, membrane-covered omphalocele containing the liver, posterior encephalocele, and unilateral microphthalmia. Gestational age estimated from a femur length of 45.3 mm. Fetal heart rate measured 128 beats/minute.
AFTER DIAGNOSIS	Multidisciplinary counselling provided regarding the prenatal diagnosis, anticipated poor prognosis, and available management options.
PREGNANCY OUTCOME	Following informed counselling and shared decision-making, the parents elected to undergo medical termination of the pregnancy.
POST-TERMINATION	Post-delivery examination, fetal autopsy, postmortem imaging, chromosomal analysis, and genetic testing were not available at the time of manuscript preparation.

A comprehensive two-dimensional prenatal ultrasonographic examination was subsequently performed on 16 July 2026 using a GE Versana Essential ultrasound system equipped with a curvilinear transducer. Gray-scale imaging and spectral Doppler evaluation were undertaken with systematic assessment of fetal viability, biometry, cranial structures, face, spine, thorax, heart, anterior abdominal wall, abdominal organs, extremities, placenta, and amniotic fluid in accordance with established obstetric ultrasonography recommendations (2–4).

The examination demonstrated a live singleton intrauterine fetus with preserved cardiac activity. Spectral Doppler confirmed a fetal heart rate of 128 beats per minute.

Because the posterior encephalocele distorted the normal cranial contour and the anterior abdominal wall defect rendered abdominal circumference unreliable, gestational age was estimated from the femur length, which measured 45.3 mm and corresponded to approximately 26 weeks' gestation (3,11).

Systematic fetal anatomical assessment revealed multiple major congenital anomalies. The fetal heart was located outside the thoracic cavity, extending into the upper abdominal and lower cervical regions while maintaining cardiac activity, consistent with ectopia cordis (5). A large midline anterior abdominal wall defect containing the liver within a membrane-covered sac was also identified, consistent with a membrane-covered liver-containing omphalocele (6,12). Sagittal imaging demonstrated a posterior calvarial defect with extracranial herniation of intracranial contents, consistent with a posterior encephalocele (13). Evaluation of the fetal face further demonstrated marked asymmetry of the globes, with one globe appearing significantly smaller than the contralateral side, consistent with unilateral microphthalmia (14).

No gross abnormalities of the spine, kidneys, urinary bladder, or extremities were identified. Likewise, there was no sonographic evidence of severe scoliosis, limb reduction defects, or abnormal placental attachment.

Based on these findings, a prenatal diagnosis of multiple severe congenital fetal anomalies comprising ectopia cordis, a membrane-covered liver-containing omphalocele, posterior encephalocele, and unilateral microphthalmia was established. The coexistence of ectopia cordis and omphalocele strongly suggested an incomplete form of pentalogy of Cantrell (7-10). However, complete diagnostic confirmation was not possible because fetal echocardiography, fetal magnetic resonance imaging, prenatal genetic testing, and postnatal pathological examination were unavailable.

Following multidisciplinary counselling involving the obstetric and radiology teams, the patient and her husband were informed of the imaging findings, anticipated prognosis, and available management options. After shared decision-making, they elected to proceed with medical termination of the pregnancy in accordance with institutional practice and applicable national regulations. No prenatal fetal intervention was undertaken because of the multiplicity and severity of the fetal anomalies and the absence of fetal surgical services.

At the time of manuscript preparation, post-delivery examination, fetal autopsy, postmortem imaging, chromosomal analysis, and molecular genetic testing had not been performed. Consequently, pathological confirmation of the prenatal diagnosis and definitive syndromic classification were not possible. The parents were counselled regarding the likely sporadic nature of the anomalies and advised that future pregnancies should include preconception counselling, early antenatal booking, first-trimester ultrasonography, and a detailed mid-trimester fetal anatomical survey, with referral for fetal echocardiography and genetic evaluation if structural abnormalities are identified (2-4, 17, 18).

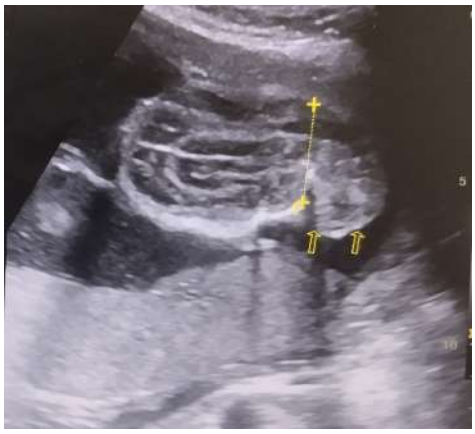

Figure 1: Posterior encephalocele. Sagittal prenatal ultrasonographic image obtained at 26 weeks' gestation demonstrating a posterior calvarial defect with extracranial herniation of intracranial contents (yellow arrows), consistent with a posterior encephalocele.

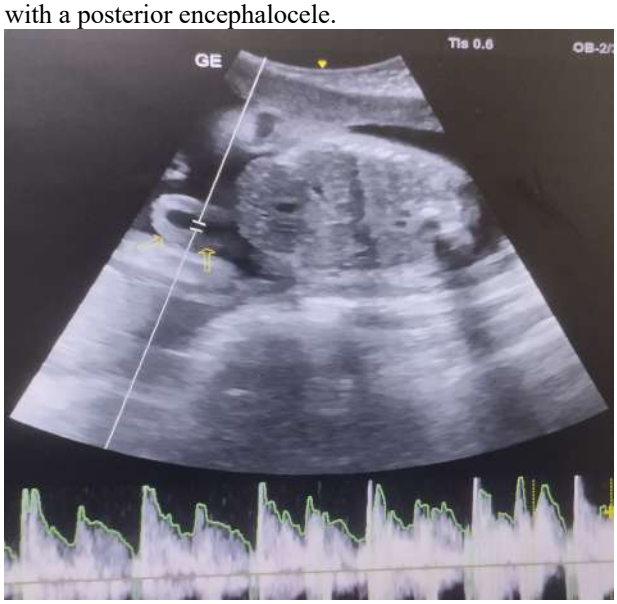

Figure 2: Ectopia cordis. Spectral Doppler ultrasonographic image demonstrating cardiac activity within the ectopic fetal heart. The yellow arrows identify the ectopic cardiac structure. The recorded fetal heart rate was 128 beats per minute, confirming fetal viability at the time of examination.

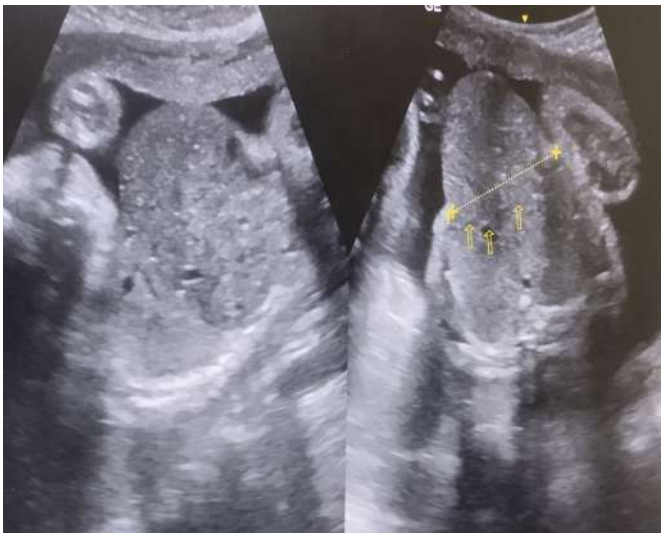

Figure 3: Membrane-covered liver-containing omphalocele. Prenatal ultrasonographic image demonstrating a midline anterior abdominal wall defect with herniation of abdominal contents, including the liver, into a membrane-covered sac (yellow arrows), consistent with an omphalocele. The maximal sac diameter measured approximately 4.5 cm.

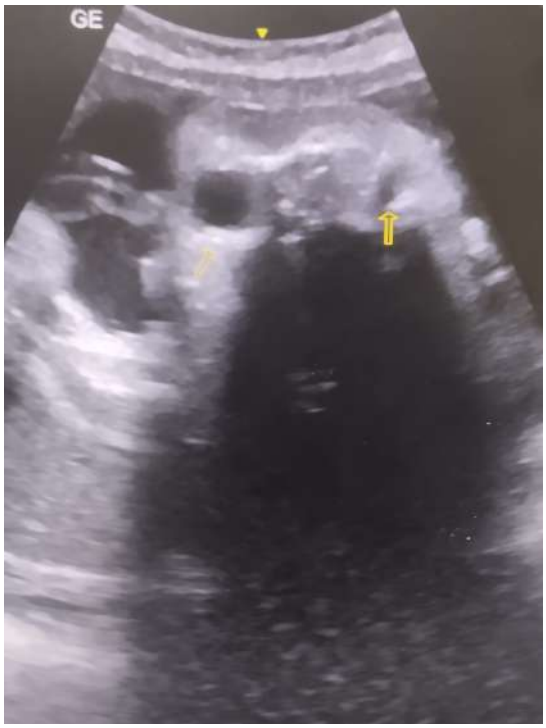

Figure 4: Unilateral microphthalmia. Axial prenatal ultrasonographic image of the fetal face demonstrating asymmetry of the orbits, with one globe appearing markedly smaller than the contralateral globe (yellow arrow), consistent with unilateral microphthalmia.

DISCUSSION

The prenatal diagnosis in this fetus was driven by the simultaneous identification of ectopia cordis and a membrane-covered liver-containing omphalocele during a routine second-trimester anomaly scan. Recognition of these two major abnormalities immediately narrowed the differential diagnosis to ventral midline developmental defects, particularly the pentalogy of Cantrell spectrum. The additional demonstration of posterior encephalocele and unilateral microphthalmia indicated a more extensive disturbance of embryogenesis and substantially broadened the diagnostic considerations. This uncommon constellation of anomalies highlights the importance of systematic fetal anatomical assessment whenever a major structural abnormality is detected.

In this fetus, routine second-trimester ultrasonography enabled recognition of multiple abnormalities involving the thoracoabdominal wall, central nervous system, and orbit. This observation reinforces the pivotal role of systematic fetal anatomical assessment during the routine mid-trimester examination, particularly where access to fetal magnetic resonance imaging, fetal echocardiography, and prenatal genetic testing is limited (2–4).

In the present case, the coexistence of ectopia cordis and a membrane-covered liver-containing omphalocele strongly suggested an incomplete form of pentalogy of Cantrell. Cantrell and colleagues originally described the syndrome as comprising five characteristic ventral midline defects, while Toyama subsequently recognized complete, probable, and incomplete forms of the condition (7,8). More recent literature continues to regard pentalogy of Cantrell as a developmental spectrum with considerable phenotypic variation rather than a uniform congenital syndrome (9,10). Although two major components were clearly demonstrated in this fetus, complete evaluation of the sternum, anterior diaphragm, diaphragmatic pericardium, and intracardiac anatomy was not possible because fetal echocardiography and advanced prenatal imaging were unavailable. Consequently, the findings are most appropriately interpreted as suggestive of an incomplete form of pentalogy of Cantrell rather than a definitive diagnosis.

The additional presence of posterior encephalocele and unilateral microphthalmia further distinguished this case from the classical description of pentalogy of Cantrell. These abnormalities are uncommon within the recognized spectrum of the syndrome and raise the possibility of a broader developmental field defect or an underlying chromosomal disorder. Similar associations have been described in fetuses with trisomy 13, trisomy 18, and other complex genetic syndromes, underscoring the value of prenatal genetic evaluation whenever multiple organ systems are affected (17,18).

Limb-body wall complex and amniotic band sequence were also considered in the differential diagnosis because both conditions may present with severe thoracoabdominal wall and craniofacial abnormalities. However, limb-body wall complex was considered less likely in the absence of severe spinal deformity, limb reduction defects, and abnormal placental attachment. Likewise, amniotic band sequence was

considered unlikely because no amniotic bands, constriction rings, or asymmetric disruption defects were identified (15,16).

A practical imaging issue in this fetus was the choice of biometric parameter. The encephalocele altered the cranial contour, while the omphalocele distorted the abdominal circumference. Reliance on these measurements could therefore have produced an inaccurate gestational age estimate. Femur length, which was not affected by the structural abnormalities, provided the most appropriate available estimate and corresponded to approximately 26 weeks' gestation (3,11).

The principal limitations of this report were the absence of fetal echocardiography, fetal magnetic resonance imaging, prenatal genetic testing, post-delivery examination, and autopsy. These investigations would have clarified the intracardiac anatomy, better characterized the encephalocele and ocular abnormality, and permitted more definitive syndromic classification. Accordingly, conclusions regarding incomplete pentalogy of Cantrell or a specific chromosomal syndrome should remain cautious.

Despite these limitations, the case demonstrates that careful prenatal ultrasonography can identify complex multisystem fetal malformations and provide clinically useful information in a resource-constrained setting. The examination supported prognostic counselling, multidisciplinary discussion, and informed parental decision-making when more advanced investigations were unavailable.

CONCLUSION

This case demonstrates the continuing importance of systematic prenatal ultrasonography in recognizing complex fetal malformations. The coexistence of ectopia cordis and a membrane-covered liver-containing omphalocele should prompt careful assessment for additional ventral midline defects, while associated cranial and ocular abnormalities should raise suspicion for a broader multisystem or genetic disorder.

When structural abnormalities make conventional fetal measurements unreliable, an unaffected parameter such as femur length may provide a more dependable estimate of gestational age. Although fetal echocardiography, magnetic resonance imaging, and prenatal genetic testing would improve diagnostic precision, meticulous ultrasonographic assessment remains central to prenatal diagnosis, prognostic counselling, and pregnancy management where access to these investigations is limited.

PATIENT PERSPECTIVE

Following detailed multidisciplinary counselling, the patient and her family indicated that they had a clear understanding of the nature of the fetal abnormalities, the anticipated prognosis, and the available management options. They expressed appreciation for the opportunity to discuss their concerns and participate in the decision-making process, which enabled them to make an informed choice regarding pregnancy management.

ETHICAL CONSIDERATION

Ethical approval for publication of this single case report was waived in accordance with institutional policy. The report was prepared in compliance with the ethical principles of the Declaration of Helsinki.

CONSENT FOR PUBLICATION

Written informed consent for publication of the clinical details and anonymized ultrasonographic images was obtained from the patient.

CONFLICT OF INTEREST

The authors declare that they have no competing interests.

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