THORACIC ECTOPIA CORDIS AT TERM WITH SEVERE INTRATHORACIC EFFUSION

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ABSTRACT

Ectopia cordis is an exceptionally rare congenital anomaly characterized by partial or complete displacement of the fetal heart outside the thoracic cavity. It represents a spectrum of defects encompassed within Cantrell's pentalogy, a constellation of five major ventral midline abnormalities. With an estimated incidence of 5.5–7.9 per one million live births, ectopia cordis remains a profound diagnostic and management challenge in prenatal medicine. We describe a 24-year-old primigravida at 36 weeks of gestation who presented with uncontrolled hypertension and suspicion of fetal structural abnormalities. Ultrasonography confirmed a severely malformed fetal heart located outside the thoracic cavity, accompanied by marked intrathoracic fluid accumulation and persistent fetal bradycardia. Given the non-survivable nature of the anomaly and absence of feasible postnatal interventions, a vaginal termination was performed. A female neonate weighing 2900 g was delivered with Apgar scores of 4 and 5, demonstrating complete thoracic ectopia cordis. The prognosis of ectopia cordis remains extremely poor and depends largely on the type of ectopia, degree of cardiac exposure, and the presence of intracardiac or associated structural malformations. Despite advances in fetal imaging and neonatal cardiac surgery, survival is exceedingly rare. In pregnancies with lethal features, conservative prenatal management represents an ethically appropriate approach. This case underscores the importance of early diagnosis, comprehensive counseling, and multidisciplinary decision-making.

Keywords: Ectopia Cordis, Congenital Anomaly, Cantrell's Pentalogy, Fetal Diagnosis, Pregnancy Management.

INTRODUCTION

Ectopia cordis is a rare congenital defect in which the fetal heart lies partially or completely outside the thoracic cavity due to failure of midline fusion during early embryogenesis. Its incidence ranges from 5.5 to 7.9 per one million live births and constitutes approximately 0.1% of all congenital cardiac anomalies (Kebalo et al., 2021). As part of Cantrell's pentalogy, ectopia cordis is associated with five characteristic defects: a supraumbilical abdominal wall

malformation, a lower sternal defect, a deficiency of the anterior diaphragm, a defect of the diaphragmatic pericardium, and congenital cardiac malformations (Mohammed J et al., 2021; Saxena, 2015) The condition is typically categorized into four anatomical subtypes based on the site of cardiac displacement: cervical (3%), thoracic (60%), abdominal (30%), and thoracoabdominal (7%). The present case represents the thoracic type, the most common variant, accompanied by omphalocele and suspected intracardiac anomalies. Given the rarity of the disorder, each case contributes valuable insights into its clinical spectrum, prognostic determinants, and management considerations (Chang et al., 2015; Shad et al., 2012).

LITERATURE REVIEW

However, outcomes remain poor due to limited thoracic capacity, risk of vascular kinking, hemodynamic instability, and the presence of associated anomalies. Existing literature including reports from Indonesia, Iraq, Turkey, Tanzania, and Togo demonstrates extremely high neonatal mortality despite attempts at staged or palliative surgical repair (Gabriel et al., 2014; Morello et al., 2024). Long-term survival is exceedingly rare and generally occurs only in cases of partial or incomplete ectopia cordis with minimal intracardiac involvement. In this case, the thoracic cavity was deemed too small to accommodate cardiac reduction, and no surgical intervention was considered feasible. This underscores the uniformly poor prognosis associated with complete thoracic ectopia cordis (Harring et al., 2015; Morello et al., 2024).

RESEARCH METHODS

CASE REPORT

A 24-year-old primigravida at 36+2 weeks of gestation was referred to Arifin Achmad Regional Hospital due to persistent hypertension and suspicion of major fetal structural abnormalities. The patient had attended regular antenatal care at a primary clinic and had no significant medical, surgical, or family history. She denied exposure to teratogenic medications, alcohol, smoking, or herbal supplements. Prenatal screening during the first trimester was unremarkable, and routine serologic tests for TORCH infection were negative. On admission, her blood pressure was 156/102 mmHg, pulse 92 beats/min, respiratory rate 20 breaths/min, and temperature 36.8°C. Fundal height corresponded to 34–35 weeks. There was no pedal edema or signs of severe preeclampsia. Laboratory findings revealed hemoglobin 11.2 g/dL, platelets 206 $\times 10^9$/L, normal liver function tests, and no proteinuria on dipstick evaluation. Fetal movements were still perceived, although the patient reported a subjective decrease over the last 24 hours. A detailed obstetric ultrasound was performed by a maternal–fetal medicine specialist. The fetus was in cephalic presentation with an estimated fetal weight of 2800 g (appropriate for gestational age).

RESEARCH RESULTS

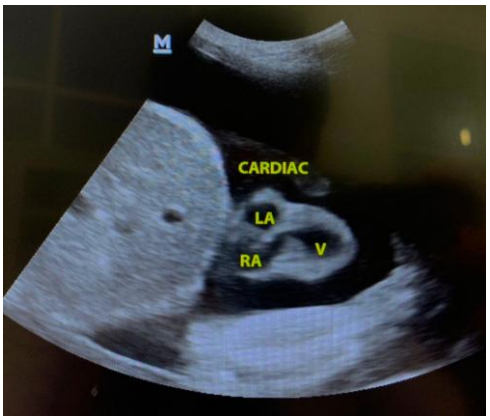

Fig. 1. Ultrasound Imaging Demonstrating The Fetal Cardiac Structures Visualized Outside The Expected Thoracic Cavity. The Four-Chamber View Shows Right Atrium (RA), Left Atrium (LA), And Ventricle (V) Displaced Anteriorly, Consistent With Thoracic Wall Defect And Ectopia Cordis.

A persistent fetal bradycardia of 88–95 bpm was documented. A large midline thoracic wall defect was observed, with the entire heart located outside the thoracic cavity and covered only by a thin membranous layer. Associated findings included marked intrathoracic effusion, absence of the sternum, and a small thoracic cavity. Preliminary cardiac assessment revealed suspected ventricular septal defect and a single-outflow–tract appearance, although full characterization was limited due to severe displacement of the heart. Additional anomalies included a small supraumbilical abdominal wall herniation consistent with omphalocele, mild polyhydramnios (AFI 25 cm), and reduced fetal breathing movement. No limb abnormalities, craniofacial malformations, or amniotic bands were identified. Umbilical artery Doppler velocimetry demonstrated elevated systolic/diastolic ratios with intermittent absent end-diastolic flow. The patient and family received counseling from a multidisciplinary team consisting of an obstetrician, neonatologist, and pediatric surgeon. Given the complete thoracic ectopia cordis, severe anatomical defects, compromised fetal hemodynamics, and absence of feasible postnatal surgical intervention, the condition was determined to be uniformly lethal.

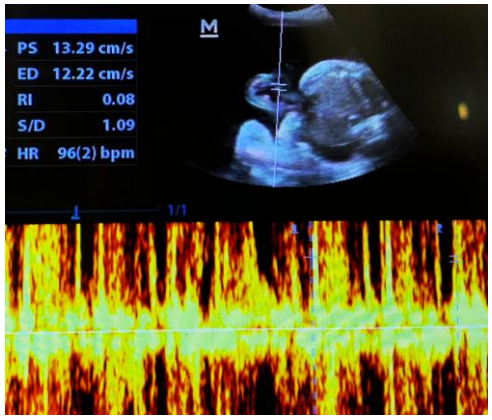

Fig. 2. Doppler Ultrasound Assessment Showing Fetal Heart Rate Tracing With Persistent Fetal Bradycardia (96 Bpm). The Spectral Doppler Waveform Also Reveals Altered Cardiac Flow Dynamics, Supporting Significant Hemodynamic Compromise.

After shared decision-making and ethical consideration, a plan for vaginal termination was agreed upon. Labor induction was initiated using a low-dose oxytocin regimen. After 6 hours of labor, a female neonate was delivered spontaneously with a birth weight of 2900 g. Apgar scores were 4 at 1 minute and 5 at 5 minutes. The neonate exhibited immediate respiratory distress, bradycardia, and cyanosis. Physical examination revealed complete thoracic ectopia cordis with the heart fully externalized, absence of the lower sternum, and a 3 × 4 cm omphalocele. The thoracic cavity was severely underdeveloped. Despite resuscitative efforts, the neonate expired shortly after birth due to cardiorespiratory failure. The placenta weighed 480 g and appeared grossly normal with a three-vessel umbilical cord. Placental histopathology was unremarkable. Postnatal chromosome analysis using FISH reported a normal 46,XX karyotype. The mother had an uncomplicated postpartum course and was discharged on day 2 with counseling regarding recurrence risk, future pregnancy planning, and early anomaly screening in subsequent gestations.

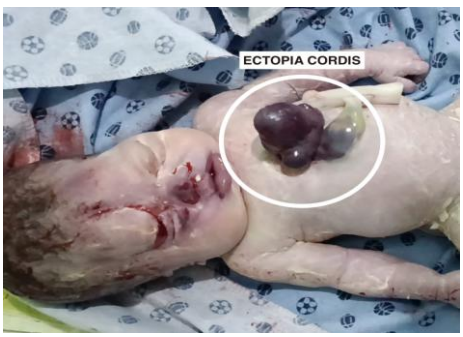

Fig. 3. Post-Delivery Clinical Photograph Demonstrating Complete Thoracic Ectopia Cordis. The Heart Is Fully Externalized From The Chest Cavity, Uncovered By The Sternum, With Associated Anterior Thoracic Wall Defect And Omphalocele

DISCUSSION

Ectopia cordis results from a failure of ventral midline fusion during early embryologic development, particularly during lateral mesoderm folding. Although its precise etiology remains uncertain, disruption of normal embryogenesis around day 21 of gestation has been implicated. Structural failure during this critical period may lead to sternal defects, thoracic wall malformations, and anterior diaphragmatic abnormalities (Achiron et al., 2011; Sepulveda et al., 2013). The anomaly is frequently associated with major intracardiac defects including ventricular septal defect, atrial septal defect, double-outlet right ventricle, tricuspid atresia, and tetralogy of Fallot and various non-cardiac malformations such as omphalocele, diaphragmatic hernia, cleft palate, and pulmonary hypoplasia. Chromosomal abnormalities (e.g., trisomy 18, trisomy 21, and Turner syndrome) have also been reported, although no cytogenetic abnormality was detected in this case (Achiron et al., 2011; Alphonso et al., 2013).

Prenatal diagnosis is usually achieved through ultrasonography during the first or early second trimester. Fetal echocardiography is essential for delineating intracardiac structures, although visualization may be limited by maternal obesity, fetal positioning, or acoustic shadowing (Çelik et al., 2015). Fetal cardiac MRI offers an additional diagnostic modality when ultrasound is suboptimal. Management of ectopia cordis is highly complex and requires a multidisciplinary team involving maternal–fetal medicine specialists, neonatologists, pediatric cardiologists, radiologists, anesthesiologists, and pediatric surgeons (Arnold et al., 2021). Surgical strategies, when feasible, typically include reduction of the heart into the thoracic cavity, reconstruction of chest wall defects, repair of omphalocele, and correction of intracardiac anomalies (Kabbani et al., 2002; Morales et al., 2000).

CONCLUSION

Ectopia cordis is a rare and typically fatal congenital anomaly with extremely poor prognosis. Outcomes depend on the type of ectopia, degree of cardiac exposure, thoracic capacity, and associated intracardiac or extracardiac malformations. In cases where the condition is unequivocally lethal, conservative prenatal management and pregnancy termination may be the most appropriate options. Early diagnosis, thorough parental counseling, and multidisciplinary care remain essential components of management.

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