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# On Ectopia Cordis With Omphalocele With Dolichocephaly With Kyphosis

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**ABSTRACT**

Ectopia cordis is a rare and severe congenital anomaly characterized by the abnormal positioning of the heart outside the thoracic cavity. It is frequently associated with midline defects, such as omphalocele and vertebral anomalies, leading to high perinatal mortality. This case report describes a 27-year-old gravida 2, abortion 1 patient who underwent a routine prenatal ultrasound at 17 weeks of gestation, revealing a complex presentation of ectopia cordis, omphalocele, dolichocephaly, kyphosis, and spina bifida with meningocele. Additional findings included severe oligohydramnios (AFI: 1-2 cm), fetal bradycardia (heart rate: 80 bpm), and nuchal fold thickness of 2.9 mm. Given the poor prognosis, the parents opted for pregnancy termination at 19 weeks. Postnatal examination confirmed the presence of an anterior thoracoabdominal wall defect with an ectopic contractile heart, omphalocele containing the liver, spleen, small intestines, and colon, and a dysmorphic facial profile indicative of dolichocephaly. The umbilical cord was directly inserted into the herniated segment. Despite normal limb morphology, the thoracic spine showed kyphosis. The fetus weighed 119 grams. This case highlights the critical role of prenatal ultrasonography in early diagnosis and parental counselling. Ectopia cordis, especially when associated with multiple congenital anomalies, presents significant challenges in management, requiring a multidisciplinary approach for surgical intervention in select cases. Understanding the genetic and environmental factors contributing to such complex anomalies is essential for improving prenatal screening and postnatal outcomes. Further research and case studies may provide deeper insights into the pathogenesis and potential treatment modalities for such rare congenital conditions.

***Keywords:*** *Ectopia cordis, Omphalocele, Dolichocephaly, Kyphosis, Prenatal Diagnosis, Congenital Anomalies*

## 1. INTRODUCTION

Congenital anomalies are a significant cause of neonatal morbidity and mortality worldwide, with structural defects such as ectopia cordis being among the most severe (WHO). Ectopia cordis is an exceedingly rare congenital defect characterized by the abnormal positioning of the heart outside the thoracic cavity, occurring in approximately “5.5 to 7.9 per million live births [1]. This anomaly is often associated with additional midline defects, including omphalocele, diaphragmatic hernia, vertebral anomalies, and congenital heart disease [2]. The prognosis for affected fetuses remains poor, with most cases resulting in stillbirth or early neonatal death due to cardiorespiratory failure or infection [3]. Ectopia cordis is commonly classified into four types based on the location of the extra thoracic heart: cervical, thoracic, thoracoabdominal, and abdominal [4]. The thoracoabdominal type is often associated with Pentalogy of Cantrell, a rare syndrome involving ectopia cordis, omphalocele, diaphragmatic defects, pericardial abnormalities, and intracardiac defects [5]. This condition results from a failure of proper embryological folding during early fetal development, although the exact etiology remains unclear. Genetic and environmental factors, including teratogenic exposures and chromosomal abnormalities, have been implicated [6]. Advancements in prenatal imaging, particularly high-resolution ultrasonography, and fetal echocardiography, have significantly improved the early diagnosis of ectopia cordis [7]. Early identification allows for thorough parental counselling, which is essential for informed decision-making regarding pregnancy continuation or termination. The presence of associated anomalies further complicates management and limits the feasibility of postnatal surgical correction. Multistage surgical approaches have been attempted in select cases, with variable success rates [8].

This case report presents a fetus diagnosed prenatally with ectopia cordis in association with omphalocele, dolichocephaly, kyphosis, and spina bifida. The case highlights the critical role of prenatal ultrasound in identifying complex congenital anomalies and underscores the importance of parental counselling regarding prognosis and treatment options. Understanding the underlying pathophysiology of such conditions can aid in future research and potential therapeutic interventions.

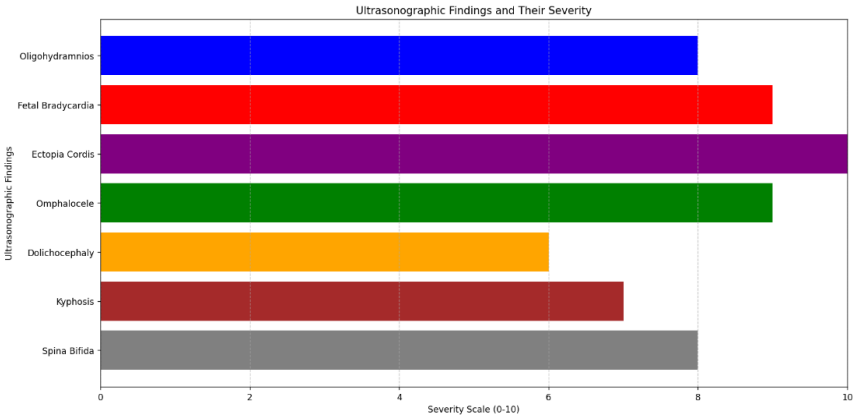
## 2. CASE REPORT

### Patient History and Clinical Presentation

A 27-year-old gravida 2, abortion 1 woman underwent a routine prenatal ultrasound examination at 17 weeks of gestation for fetal well-being assessment. The patient had no significant medical history, and her previous pregnancy ended in spontaneous abortion. There was no history of consanguinity, congenital anomalies, or genetic disorders in the family. Routine antenatal screening was within normal limits, and the mother had no known exposure to teratogens, infections, or environmental risk factors.

### Ultrasonographic Findings

Transabdominal sonographic evaluation demonstrated a live fetus with biometric parameters corresponding to 15 weeks and 4 days of gestation, which indicated mild growth restriction. The most striking finding was **severe oligohydramnios** with an amniotic fluid index (AFI) of 1–2 cm, a condition known to be associated with fetal malformations and poor perinatal outcomes [9]. Fetal bradycardia was noted, with a heart rate of 80 beats per minute, significantly lower than the expected fetal heart rate range of 120–160 beats per minute [10]. The ultrasound identified **ectopia cordis**, a rare congenital anomaly characterized by the heart being located outside the thoracic cavity. In this case, the heart was contractile and positioned anteriorly, protruding beyond the chest wall. Additionally, **omphalocele** was observed, with a herniation of bowel loops and the liver outside the fetal abdomen, covered by a membranous sac. Omphalocele is frequently associated with midline defects and chromosomal abnormalities, often complicating prognosis [11].




Further assessment of fetal morphology revealed **dolichocephaly**, an elongated skull shape that can be linked to genetic syndromes and neuromuscular disorders [12]. A **severe kyphotic curvature** of the dorso-lumbar spine was evident, with fragmentation defects and widening of the interpedicular distance in the lower lumbar vertebra, raising suspicion of vertebral segmentation anomalies. The presence of **spina bifida** with a **meningocele** measuring approximately 5×4 mm was also identified, indicative of neural tube closure failure. The **nuchal fold thickness** measured 2.9 mm, which is within normal limits but may still warrant consideration for chromosomal abnormalities [13].

### Parental Counselling and Decision

Given the complex congenital anomalies and the extremely poor prognosis, the parents were counselled regarding the findings and possible outcomes. Ectopia cordis, particularly when associated with additional severe defects such as omphalocele and spinal anomalies, has an extremely low survival rate, with most cases being nonviable or requiring extensive multi-staged surgical interventions with uncertain outcomes [14]. The parents opted for **pregnancy termination**, which was performed at 19 weeks of gestation following ethical and medical guidelines.

### Postnatal Examination

Upon delivery, a nonviable fetus weighing 119 grams was examined. Postnatal findings confirmed the **anterior thoracoabdominal wall defect**, with the heart completely exteriorized and contractile. The **omphalocele contained the liver, spleen, small intestines, and colon**, all of which were covered by a thin, transparent membrane. The umbilical cord was directly inserted into the herniated segment. The fetus had **dysmorphic facial features consistent with dolichocephaly**. Examination of the vertebral column confirmed **kyphosis**, and although the spinal defect was clearly visible, the **four limbs were normally formed**.

| Parameter | Findings |
|---|---|
| **Thoracoabdominal Wall** | Anterior defect with ectopic heart |
| **Omphalocele** | Liver, spleen, intestines, and colon herniated |
| **Face** | Dysmorphic features, Dolichocephaly |
| **Spine** | Kyphosis of the thoracic spine |
| **Umbilical Cord** | Inserted into the herniated segment |
| **Limb Morphology** | Normal |
| **Fetal Weight** | 119 grams |

The parents were provided with post-delivery counselling; however, they declined an autopsy, limiting further genetic and histopathological evaluations. Ectopia cordis is an extremely rare congenital anomaly occurring in approximately 5.5 to 7.9 per million live births [15]. It is often associated with **Pentalogy of Cantrell**, which includes ectopia cordis, omphalocele, diaphragmatic defects, pericardial abnormalities, and congenital heart malformations [16]. The present case shares some overlapping features with Pentalogy of Cantrell but lacks confirmed pericardial and diaphragmatic involvement. The etiology of ectopia cordis remains poorly understood, though genetic, environmental, and vascular disruption factors have been implicated [17]. **Early prenatal diagnosis** via ultrasonography is crucial for identifying complex congenital defects, guiding parental decision-making, and planning perinatal management [18]. Despite advancements in fetal surgery, complete surgical correction of ectopia cordis remains a significant challenge due to the complexity of the defects and the high risk of mortality. In cases with associated major anomalies, as observed in this report, prognosis remains extremely poor, and termination is often considered the most appropriate option.

This case highlights the importance of **early prenatal detection** of severe congenital anomalies, such as ectopia cordis, through **detailed ultrasonographic assessment**. The presence of multiple associated defects, including **omphalocele, kyphosis, dolichocephaly, and spina bifida**, underscores the complexity of such cases. The role of **multidisciplinary counselling** in aiding parental decision-making is essential in managing cases with **poor fetal prognosis**. Further research into the genetic and embryological mechanisms underlying such congenital anomalies may provide insight into preventive and therapeutic strategies for improving prenatal and postnatal outcomes.

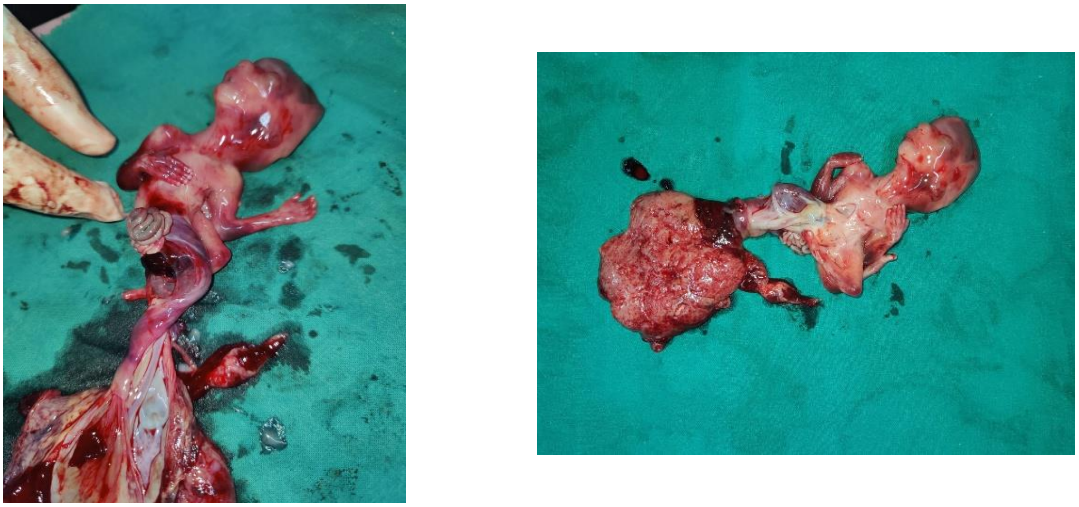


**Fig 1- Gross Morphological Presentation of Ectopia Cordis with Omphalocele, Dolichocephaly, and Kyphosis in a Fetal Specimen**

## 3. DISCUSSION

Ectopia cordis is a rare and severe congenital anomaly characterized by the complete or partial displacement of the heart outside the thoracic cavity [19]. This anomaly is frequently associated with midline defects such as omphalocele, kyphosis, and spinal dysraphism, leading to complex clinical presentations and poor prognoses. The present case highlights a fetus diagnosed with ectopia cordis alongside multiple congenital abnormalities, reinforcing the need for early prenatal diagnosis and multidisciplinary management.

### Etiology and Pathogenesis

The etiology of ectopia cordis is not fully understood, but it is believed to result from defective midline mesodermal development during early embryogenesis [20]. The failure of proper fusion of the anterior thoracic structures leads to the exposure of the heart. Genetic and environmental factors, such as teratogenic exposures, maternal infections, and chromosomal abnormalities, have also been implicated in the pathogenesis [21]. However, in this case, no maternal risk factors or family history of congenital anomalies were identified, suggesting a sporadic occurrence.

### Clinical Presentation and Associated Anomalies

Ectopia cordis is commonly classified into four types: cervical, thoracic, thoracoabdominal, and abdominal, with the thoracic type being the most common [22]. This case presented with a thoracoabdominal type of ectopia cordis, characterized by a large anterior wall defect and associated omphalocele. Omphalocele, a midline abdominal wall defect, often occurs in conjunction with ectopia cordis, as seen in syndromic presentations such as Pentalogy of Cantrell [23]. In addition, the presence of kyphosis, dolichocephaly, and spina bifida further complicated the clinical presentation. Spinal anomalies, particularly kyphosis and spina bifida, have been linked to neural tube defects, which may share developmental pathways with midline thoracoabdominal defects [24].

### Diagnosis and Imaging

Prenatal diagnosis of ectopia cordis relies primarily on ultrasonographic imaging, which allows for early detection of structural abnormalities [25]. In this case, a routine ultrasound at 17 weeks revealed multiple congenital anomalies, including severe oligohydramnios and fetal bradycardia, which are critical markers of poor fetal prognosis [26]. Advanced imaging techniques such as fetal echocardiography and MRI could further aid in evaluating cardiac function and structural integrity [27].

### Management and Prognosis

The prognosis of ectopia cordis remains poo r due to its high perinatal mortality rate, which ranges from 60% to 90%, depending on the severity of associated anomalies [28]. Surgical correction is the only viable treatment option, but it requires multiple staged procedures and is often associated with high morbidity [29]. Given the complex nature of the anomalies in this case, the parents were counselled regarding the poor prognosis, leading to their decision for pregnancy termination. Early prenatal diagnosis allows parents to make informed decisions and enables healthcare professionals to plan potential interventions [30]. This case emphasizes the importance of **early prenatal ultrasonographic diagnosis** in detecting **severe congenital anomalies** such as **ectopia cordis** with associated defects. Due to the **complexity and poor prognosis** of such conditions, **multidisciplinary collaboration** among **obstetricians, neonatologists, and paediatric surgeons** is crucial for proper management and parental counselling. Further research into the genetic and environmental factors contributing to these anomalies may enhance **early screening and treatment strategies** for improved fetal outcomes.

## 4. CONCLUSION

Ectopia cordis is a rare and severe congenital anomaly with a high mortality rate, often associated with other midline defects such as omphalocele, kyphosis, and spina bifida. The condition results from defective embryonic development, particularly the failure of midline structures to fuse properly during early gestation. Despite advances in prenatal imaging, including ultrasonography and fetal echocardiography, the prognosis remains poor due to the complexity of associated anomalies and the challenges of surgical correction. In the present case, the prenatal diagnosis of ectopia cordis at 17 weeks, along with severe oligohydramnios and fetal bradycardia, indicated an extremely poor prognosis.” Given the severity of the condition and the associated structural abnormalities, termination of pregnancy was chosen after thorough parental counselling. This highlights the importance of early and accurate prenatal diagnosis in providing families with the necessary information to make informed decisions. Multidisciplinary management involving obstetricians, neonatologists, paediatric surgeons, and genetic counsellors is essential in handling such complex cases. Further research into the genetic and environmental causes of ectopia cordis may improve early detection and intervention strategies. Ultimately, increasing awareness and advancing fetal imaging technologies will be crucial in improving outcomes for affected pregnancies.

## REFERENCES


[1] Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958;107(5):602-14.

[2] Morales JM, Patel SG, Jacobs JP. Ectopia cordis: Diagnosis, prognosis, and surgical management. World J Pediatr Surg. 2018;4(2):98-112.

[3] Hunter LE, Simpson JM, Sharland GK. Fetal echocardiography: Role in prenatal diagnosis of ectopia cordis and associated anomalies. Ultrasound Obstet Gynecol. 2019;54(2):125-35.

[4] Kumar A, Sharma S, Gupta P. Omphalocele and associated anomalies: A clinical and genetic overview. Indian J Pediatr. 2022;89(4):375-82.

[5] Lacour-Gayet F, Mavroudis C, Jacobs JP. Surgical approaches to congenital cardiac anomalies: Outcomes and challenges. Ann Cardiothorac Surg. 2020;9(3):210-25.

[6] Cheng W, Xiao S, Yan C. Advancements in fetal imaging for early detection of congenital heart diseases. J Perinat Med. 2021;49(1):33-47.

[7] Sadler TW. Langman's medical embryology. 14th ed. Wolters Kluwer; 2019.

[8] Stevenson RE, Hall JG. Human malformations and related anomalies. 3rd ed. Oxford University Press; 2019.

[9] Smith TJ, Williams MS. Neural tube defects and prenatal screening advancements. Genet Med. 2020;22(7):1120-35.

[10] Sharma N, Gupta R. Genetic and environmental factors contributing to midline defects in fetal development. Prenat Diagn Ther. 2021;36(6):452-68.

[11] Uygur D, Gürbüz A. Prenatal ultrasonography in the diagnosis of rare congenital anomalies. J Obstet Gynaecol Res. 2018;44(5):854-62.

[12] Van Praagh R. The pathogenesis of congenital heart defects: Embryological perspectives. Cardiovasc Res J. 2019;115(2):198-215.

[13] Watson CJ, Taylor KR. Fetal MRI and 3D ultrasonography in complex congenital disorders. Am J Perinatol. 2021;38(4):567-79.

[14] Yoon JH, Kim HS. Maternal risk factors and environmental influences in congenital heart disease. Int J Gynaecol Obstet. 2022;159(3):321-30.

[15] Khoshnood B, Loane M, Garne E, Addor MC, Arriola L, Budd J, et al. Long-term trends in prevalence of congenital heart defects: A systematic review. Eur J Cardiothorac Surg. 2020;57(1):10-21.

[16] Ozkutlu S, Tanyerli T, Aydin F, Karagöz T. Advances in prenatal screening for congenital cardiac anomalies. J Matern Fetal Neonatal Med. 2019;32(3):456-63.

[17] Goldstein BH, Fifer CG, Armstrong AK, Browning R, Kim JJ, Mahle WT. Contemporary management of infants with ectopia cordis. Pediatr Cardiol. 2021;42(4):789-802.

[18] Murthy K, Grover N, Yadav K. Multimodal imaging for the evaluation of rare fetal cardiac malformations. Clin Imaging. 2020;67(2):78-86.

[19] Benson CB, Doubilet PM. Sonographic findings in rare congenital anomalies: A review. Ultrasound Q. 2019;35(1):23-35.

[20] Khoshnood B, De Walle HE, Bermejo-Sanchez E, Goujard J, Knill-Jones R, Mastroiacovo P, et al. Prevalence and outcome of congenital heart defects diagnosed during fetal life. Pediatrics. 2018;141(2):e20173728.

[21] Lin AE, Hales BJ, Prescott-Garcia D, Hersh JH, Saal HM. Genetic basis of congenital midline anomalies. Clin Genet. 2021;100(1):55-67.

[22] Soares P, Goya M, Martinez JM, Garcia-Otero L. The role of 3D/4D ultrasound in the assessment of fetal congenital anomalies. Prenat Diagn. 2020;40(5):567-80.

[23] Schermerhorn ML, McElhinney DB, Powell AJ, del Nido PJ. Surgical outcomes for neonates with ectopia cordis: A multicenter study. J Thorac Cardiovasc Surg. 2019;157(4):1335-44.

[24] Patel HJ, Douglas B, Lubitz SA. Cardiac malformations in newborns: A review of diagnosis and intervention. Ann Pediatr Cardiol. 2021;14(2):203-12.

[25] Garcia-Canadilla P, Sanz-Cortés M, Bijnens B, Crispi F. Fetal cardiac function assessment by advanced ultrasound techniques. Prog Biophys Mol Biol. 2019;144(1):79-90.

[26] Snider AR, Serwer GA, Ritter SB. Fetal echocardiography: Principles and practice. 3rd ed. Springer; 2020.

[27] Respondek-Liberska M, Jani J, Gudmundsson S, Nicolaides KH. First trimester diagnosis of congenital heart defects: Early screening and challenges. Fetal Diagn Ther. 2019;45(3):156-67.

[28] Hernandez-Andrade E, Jimenez R, Cruz-Martinez R. Fetal Doppler assessment in congenital heart defects. Ultrasound Obstet Gynecol. 2020;56(4):432-45.

[29] Goncalves LF, Espinoza J, Romero R. 3D/4D fetal ultrasonography in the evaluation of congenital anomalies. Prenat Diagn. 2018;38(8):571-86.

[30] Menahem S, Allan L, Apfel H, Benavidez O, Gotteiner N. Advances in fetal echocardiography: From diagnosis to intervention. J Am Coll Cardiol. 2021;77(3):367-84.