

## INTRODUCTION

Ectopia cordis is a rare congenital condition in which the heart is located partially or totally outside of the chest cavity. It is estimated to occur in 5.5–7.9 per 1,000,000 live births, with a high mortality rate in the first few days of life. The condition can be classified into four types based on the position of the heart: cervical, thoracic, thoracoabdominal, and abdominal.

## AIM AND OBJECTIVE

The aim of this study was to present the clinical and radiological findings of a patient with thoracic ectopia cordis and discuss the management and outcomes of this rare anomaly. This case highlights the critical role of first-trimester ultrasound in diagnosing severe fetal anomalies and the importance of early recognition and prompt referral for the best possible outcomes.

## PROCEDURE

A 28 year-old woman, primi gravida conceived with ovulation induction with clomifene citrate in our hospital and underwent a routine prenatal ultrasound at 12 weeks of gestation. The patient had no significant medical history and no family history of congenital anomalies. Ultrasound showed a single, live, intrauterine gestation corresponding to a gestational age of 12 weeks. The fetal heart was visualized outside the chest through a defect in the lower sternum in suggestive of thoracic variety of ectopia cordis. The patient was advised double marker test and amniocentesis but was not affordable. After extensive discussions with the patient and her family, the decision was made to offer termination of pregnancy due to the poor prognosis of the fetus and the potential risks to the mother's health during continued pregnancy. The patient was provided with counselling and support throughout the decision-making process. Medical termination of pregnancy was performed under close monitoring and supervision. Expulsion of the fetus was achieved safely. When examined macroscopically, it was observed that the heart was completely outside the thorax with the proximal sternum defect. These findings were observed to be consistent with isolated thoracic-type EC. Since the family did not accept an autopsy, further examination could not be performed. The patient was discharged after receiving a psychological care.



## DISCUSSION

EC is a rare malformation that occurs as an isolated lesion or as part of the pentology of Cantrell which is characterized by midline closure defects. Although its etiology is unknown, the hypothesis that it originates from the defects of the lateral mesoderm in early gestational weeks. As the newest theory in etiology suggests; it is caused by mutation in bone morphogenetic protein (BMP2) gene mediating the cardiac tube formation and ventral body wall closure. EC is considered an isolated and sporadic malformation. With the widespread implementation of first-trimester sonographic examination as a screening tool for aneuploidy in current clinical practice, it is possible to detect EC at an early gestational age.

## CONCLUSION

Prenatal diagnosis of isolated thoracic-type EC is a very rare condition. Antenatal ultrasound scan is of great value in the prenatal assessment of this defect. Cardiac screening should be considered in the early weeks to diagnose this malformation in the first trimester. Due to its poor prognosis in the postnatal period, termination may be offered to these patients as an option.

## REFERENCE

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