

Early Prenatal Diagnosis of Ectopia Cordis: A Case Report

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Ectopia cordis is a very rare congenital anomaly with an incidence of 5.5-7.9 per million live births. In this manuscript we report an ectopia cordis diagnosed in a fetus at 12 weeks.

Key Words: Ectopia cordis, Pregnancy, Ultrasonography

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Introduction

Ectopia cordis is defined as a congenital anomaly in which the fetal heart is partially or completely displaced outside the thoracic cavity. It is a very rare congenital anomaly with an incidence of 5.5-7.9 per million live births.¹

In this manuscript we report an ectopia cordis diagnosed in a fetus at 12 weeks.

Case Report

A 26 year old, gravida 2, para 1 woman at 12 weeks pregnancy attended our outpatient clinic for a routine antenatal check-up. Her obstetric history revealed that she had had one singleton pregnancy resulting in one healthy male child without any congenital problems. There was no relationship between the patient and her husband.

Both the transabdominal and transvaginal ultrasonographic (USG) examinations (5.0 and 3.5-MHZ curved-array transducer, GE Medical, Voluson 730 Pro, Istanbul, Turkey) revealed that she had a singleton live fetus with crown-rump length (CRL) 65 mm consistent with a gestation of 12 weeks. Fetal heart pulsations were clearly seen outside the thoracic cavity via Doppler USG examination (Figure 1). Ectopia cordis was clearly seen and diagnosed. USG examination also revealed that a cystic hygroma was associated to the ectopia cordis (Figure 1). There was no associated anomaly seen in USG examination.

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Prenatal counseling was given to the parents and they decided to terminate the pregnancy. The pregnancy termination was successfully performed. Fetal karyotype revealed that the fetus was 46 XX.

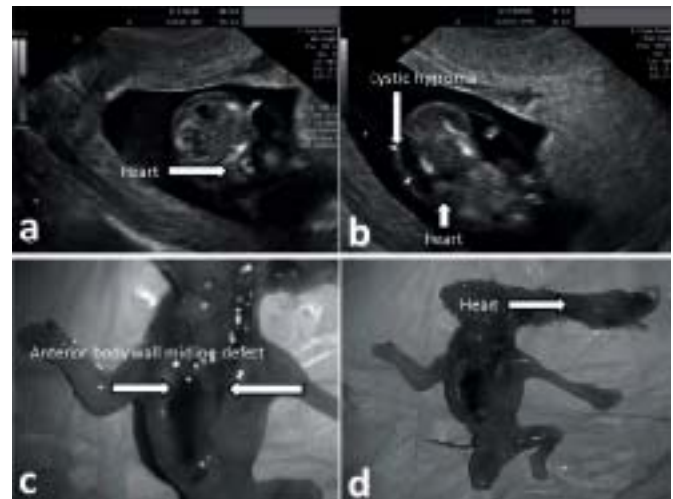


Figure 1: Ectopia cordis is clearly seen in the fetus (arrow). There is no skin and thoracic wall covering the heart.

Discussion

Ectopia Cordis (EC) is usually seen as a sporadic congenital anomaly. The etiology of EC is unknown. A failure in the closure of the thoracic region during embryologic development is the most commonly accepted hypothesis in the pathogenesis. The closure of the thoracic and abdominal region is completed at the 9th embryonic week.² Therefore the prenatal diagnosis of EC could be possible as early as 9-10 weeks. With the development of prenatal diagnostic instruments such as high resolution transabdominal or transvaginal ultrasonography, early diagnosis of congenital anomalies could be possible. Early prenatal diagnosis of EC in a pregnancy at 13 weeks became possible by 1989 according to Riboni and Agosti.³ The earliest diagnosis of EC was reported by Liang et al.⁴ in a pregnant patient at 10 weeks. We also diagnosed our EC in a fetus at 12 weeks.

Isolated EC is rarely seen. EC diagnosed in the first trimester is usually associated with several congenital anomalies such as omphalocele, cystic hygroma and Cantrell's pentalogy.⁵ Cantrell's pentalogy is characterized as an anterior body wall midline developmental anomaly which consists of a supraumbilical wall defect, a defect of the lower sternum, a deficiency of the anterior diaphragm, a defect of the diaphragmatic pericardium, and various intracardiac defects.⁶ EC is frequently found in association with Cantrell's pentalogy. In our case we only saw a cystic hygroma associated to the EC. There was no umbilical abdominal wall defect in USG examination, but we were not sure about the intracardiac anomalies and the deficiency of the anterior diaphragm since the fetus was at 10 weeks and therefore very small.

It is well known that cystic hygroma may be associated with congenital heart defects.⁷ Bennet et al.,⁸ Hsieh et al.⁶ and Peixoto-Filho et al.⁹ reported that EC was associated with cystic hygroma. In our patient EC was associated with a cystic hygroma also.

According to cordial location, EC can be classified into four types: cervico-thoracic and thoracic (65%), abdominal (10%), thoraco-abdominal (20%), and cervical (5%).¹⁰ In our case EC was located in the cervico-thoracic region.

In conclusion, first trimester diagnosis of EC is easy with USG examination. Since the prognosis of EC is very poor, when diagnosed in the first trimester, fetal termination should be offered to the parents.

Ektopia Kordisin Erken Tanısı: Olgu Sunumu

Ektopia kordis 5.5-7.9 milyon canlı doğumda 1 izlenen çok nadir bir doğuştan anomalidir. Bu olgu sunumunda 12 haftalık bir fetusta tespit edilen ektopia kordis sunulmuştur.

Anahtar Kelimeler: Ektopia kordis, Gebelik, Ultrasonografi

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