

Prenatal Diagnosis of a Rare Congenital Anomaly, Isolated Ectopia Cordis: A Case Report

Prenatal Dönemde Tanısı Konulan Nadir Bir Konjenital Anomali; İzole Ektopia Kordis

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ABSTRACT Ectopia cordis is a very rare congenital malformation in which the heart is located outside the thorax. Reported prevalence of ectopia cordis is 0.5-1/100.000. Ectopia cordis may present as an isolated malformation; however, it may also be accompanied by Cantrell's pentalogy. Termination of pregnancy has been offered for the cases diagnosed at early gestational age since neonatal surgery outcomes are generally poor. Some genetic abnormalities associated with ectopia cordis, have been described. Fetuses with ectopia cordis diagnosed prenatally should be evaluated for genetic abnormalities, which guide prenatal and postnatal management of cases. Here, a case of isolated ectopia cordis diagnosed by prenatal ultrasonography that was performed in the 18th week of pregnancy has been presented along with the discussion of the case based on the literature review.

Key Words: Ectopia cordis; ultrasonography, prenatal

ÖZET Ektopia kordis, 0.5-1/100.000 sıklıkta nadir görülen, kalbin toraks dışında bulunması ile karakterize, bir konjenital anomalidir. Ektopia kordis izole bir anomali olabildiği gibi Cantrell pentalojisine eşlik eden bir anomali olarak da bulunabilir. Prenatal dönemde tanı konulan olgulara uygulanan neonatal cerrahi sonuçları genelde kötüdür ve erken gebelik haftalarında tanı alan olgulara gebelik sonlandırması önerilmektedir. Ektopia kordia anomalisine eşlik eden çeşitli genetik anormallikler tanımlanmıştır. Prenatal dönemde ektopia kordis tanısı alan fetüslere, prenatal ve postnatal takibin belirlenmesi için genetik değerlendirme yapılmalıdır. Bu yazıda kliniğimize 18. gebelik haftasında başvuran bir gebenin prenatal ultrasonografisinde saptanan izole ektopia kordis malformasyonlu fetus sunuldu ve konu literatür bilgileri eşliğinde tartışıldı.

Anahtar Kelimeler: Ektopia kordis; ultrasonografi, prenatal

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Ectopia cordis (EC) is a very rare congenital malformation in which the heart is completely or partially located outside the thorax.¹ It is more frequent in female fetuses and reported prevalence of EC is 0.5-1/100.000.^{1,2} Etiology of the malformation is unknown. EC may present as an isolated malformation or it may be associated with anterior body wall defects. The condition may be a component of Cantrell's Pentalogy.¹⁻⁵ Neonatal prognosis of the fetuses with ectopia cordis malformation are generally poor since intrinsic cardiac defects and other system abnormalities generally accompany the malformation.^{1,2}

CASE REPORT

A 17-year-old primigravid patient referred to our out-patient clinic in the 18th week of pregnancy on April 28, 2008 due to high alpha fetoprotein determined during screening in the second trimester. Ultrasonography was performed, and an 18-week old female fetus with a defect in the anterior thorax and thoracic ectopia cordis malformation was observed (Figure 1). All the other structures were normal. The history of the patient was non-specific except elevated alpha feto-protein level (2.639 mom) in the second trimester of her pregnancy determined on April 24, 2008. The patient was informed and referred to a larger medical center for further evaluation. On May 3, 2008, the patient was readmitted to our clinic with the complaint of vaginal bleeding. She never applied to the center she had been referred to. Ultrasonography was performed and an in utero-ex fetus was detected. The written informed consent of the patient was obtained, and the pregnancy was terminated on May 5, 2008. The macroscopic evaluation of the fetus revealed an anterior thoracic defect and extrathoracally located heart (Figure 2). The fetus was sent in for pathologic examination for investigation of any intrinsic cardiac defects and other possible abnormalities. A sample of placenta was sent to the genetic laboratory. Pathologic investigation showed a female fetus, weighing 320 gram, having partially distal sternal defect, and thoracic ectopia cordis.

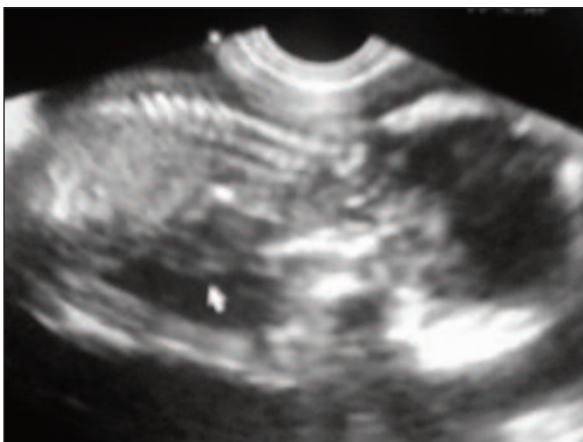


FIGURE 1: Longitudinal ultrasound section of the fetus; the arrow shows extrathoracic location of the heart.



FIGURE 2: The appearance of the female fetus with thoracic ectopia cordis, after termination of pregnancy.

There were no intrinsic cardiac defects or any other abnormalities. Genetic investigation presented a normal 46, XX karyotype.

DISCUSSION

EC refers to an anterior thoracic defect and extrathoracic location of the heart.¹ It is a very rare congenital malformation with an estimated prevalence of 0.5-1/100.000 births and it is more frequent in females.^{1,2} The formation of the thoracic and abdominal walls is complete in the ninth week of pregnancy and of the heart in the eighth week. Complete or incomplete failure of midline fusion at this embryonic stage can result in a variety of disorders ranging from isolated ectopia cordis to complete ventral evisceration.³ EC can easily be diagnosed by prenatal ultrasonography after tenth weeks of pregnancy.³ EC is classified in five categories according to the location of the defect: Cervical, cervicothoracic, thoracic, thoracoabdominal, and abdominal. Thoracic and thoracoabdominal types are 85% of all EC cases.⁴ Thoracoabdominal EC is usually accompanied by Cantrell's pentalogy, which consists of sternal defect, ectopia cordis, diaphragmatic defect, diaphragmatic pericordial defect, and anterior abdominal wall defect.^{5,6} Hence, all cases with EC diagnosed by prenatal ultrasonography should be investigated for omphalocele and other concomitant malformations. In our case, we did not detect any malformations except isola-

ted sternal defect and thoracic EC. Eighty percent of fetuses with EC also have intrinsic cardiac malformations, which are related with poor neonatal prognosis.⁴ The most common cardiac malformations accompanying EC are atrial septal defect, ventricular septal defect and tetralogy of Fallot.^{4,7} EC is considered an isolated and sporadic malformation. Nevertheless, various chromosomal abnormalities related with EC have been reported. Trisomy 18 is the most common chromosomal abnormality accompanying EC.⁸ In the light of these reports, a diagnosis of EC by prenatal ultrasonography should also be considered genetic investigation. Prognosis of EC varies depending on the presence of accompanying structural abnormalities and intrinsic cardiac defects. Antenatal or neonatal death from EC

is common. An atraumatic caesarean birth followed by corrective neonatal surgery has been suggested for fetuses with normal karyotype and diagnosis of isolated EC.⁹ Few successful outcomes have been reported in neonates with isolated EC after corrective surgery;¹⁰ however, the procedure often fails secondary to kinking of the great vessels or mechanical compression of the heart, resulting in decreased cardiac output.^{11,12}

In conclusion, prenatal diagnosis of isolated EC is a very rare occasion. Isolated EC requires appropriate prenatal and postnatal management. Genetic counseling and multidisciplinary clinic evaluation by obstetricians, pediatricians, and pediatric cardiovascular surgeons should be planned.

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