Case Report

Journal Homepage: http://crcp.tums.ac.ir

A Case of Antenatal Diagnosis of Ectopia Cordis with Cardiac Disease

Fariba Rashidighader [1], Mohammadrafie Khorgami [1], Elio Caruso[2], Farruggio Silvia[2], Reza Masihi[1]

1. *Rajaie Cardiovascular Medical and Research Center, Iran University of Medical Sciences, Tehran, Iran.*
2. *Department of Pediatric Cardiology,Mediterranean Pediatric Cardiology Center "Bambin Gesù", San Vincenzo Hospital, Taormina, Italy.*

Use your device to scan and read the article online

Citation Rashidighader F, Khorgami M., Caruso E, Silvia F, Masihi R. A Case of Antenatal Diagnosis of Ectopia Cordis with Cardiac Disease. Case Reports in Clinical Practice. 2023; 8(1):34-38.

Running Title Antenatal Diagnosis of Ectopia Cordis

ABSTRACT

Ectopic cordis is a rare anomaly with incidence of 5.5 to 7.9 per one million. In this anomaly, fetal heart is displayed towards outside of the thoracic cavity partially or completely. We describe a case of antenatal diagnosis of Thoracic Ectopia Cordis (EC) with CongenitalHeartDisease (CHD) without any other extracardiac malformations. The mother was referred to our center at 18 weeks of gestation due to abnormal sonography. Fetal Echocardiographic examination showed isolated thoracic ectopia cordis with Double Outlet Right Ventricle (DORV), large inlet to outlet Ventricular Septal Defect (VSD), malposition of Aorta, Pulmonary Atresia with retrograde flow, narrow Pulmonary Artery (PA) and PA branches. Amniocentesis and sonography revealed no other anomaly or chromosomal derangement. Because the diagnosis of Ectopia cordis may be difficult in the fetal period due to multiple factors, meticulous attention should be paid for true diagnosis.

Article info:
Received: 09 Jan 2023
Revised: 30 Jan 2023
Accepted: 16 Febr 2023

Keywords:
Ectopia cordis; Pregnancy; Prenatal diagnosis; Heartdisease

*** Corresponding Author:**
Reza Masihi, MD.
Address: *Rajaie Cardiovascular Medical and Research Center, Iran University of Medical Sciences, Tehran, Iran.*
E-mail: *farhad.heidary@yahoo.com*

Introduction

Ectopic cordis is a rare anomaly with incidence of 5.5 to 7.9 per one million. In this anomaly, fetal heart is displayed towards outside the thoracic cavity partially or completely and includes 0.1% of CHD. Ectopic cordis may be accompanied with extracardiac and intracardiac congenital disorders. The infants with EC usually have poor prognosis and many factors affect it including type of ectopic cordis and severity of congenital and cardiac disorders. (1, 2). It affects all races equally with boys more commonly affected than girls (3, 4).

Based on the position of the heart, the following classification was suggested: thoracic (60% of cases), abdominal (30%), thoraco-abdominal (7%), cervical (3%), and cervicothoracic (<1%) (2, 4, 5). Among different types of ectopic cordis, the thoraco-abdominal type has the best prognosis and the cervical type has the worst prognosis and those affected usually do not survive. (6) Most newborns (90%) with Ectopia cordis are born with associated cardiac malformations. The most frequent CHD is VSD in 59% of cases. Other anomalies include pulmonary atresia or stenosis, atrial septal defect, tetralogy of fallots, RV diverticulum, left superior vena cava and DORV (4). Other cardiac malformations like single ventricle, LV diverticulum (7), complete transposition of great arteries, and atrioventricular septal defect occur rarely (4). Ectopic cordis is a segmental defect in mesodermal development occurring in the third week of fetal period. Another theory is amniotic band syndrome leading to thoraco-abdominal and cervical malformations.

The ectopic cordis usually is an isolated disorder but it may be associated with other midline defects called the Pentalogy of Cantrell syndrome (7, 8). Pentalogy of Cantrell was reported first by Cantrell in 1958. This syndrome consists of five defects: thoraco-abdominal EC, anterior diaphragmatic hernia, lower sternal defect, midline supra-umbilical defect (omphalocele) with pericardial and intra-cardiac defects (Table 1). Ectopic cordis may be accompanied with Extracardiac anomalies including amniotic band syndrome, cleft lip and palate, omphalocele, diaphragmatic hernia, body stalk syndrome and skeletal malformations such as kyphosis (Table 1). The thoracoabdominal type is frequently associated with Cantrell's pentalogy (9). Normal karyotype is most often reported in screened patients, however, abnormal karyotypes reported include XXY, trisomy 18 and trisomy 21 & Turner syndrome (4, 10).

Case Presentation

A 38-year-old woman (gravida 5, para 1) was referred for more evaluation at 18th week of pregnancy. She had regular prenatal care during gestation. At 14th gestational week, extra thoracic heart with anterior thoracic defect was recognized. The pregnancy was spontaneous (not product of assisted reproduction). There was history of 3 abortions and one healthy 9year-old girl and family history of CHD in mother's brother which was operated on successfully. There was no family history of genetic disorders or any history related to ectopia cordis. The mother was receiving folic acid before pregnancy and there was no history of X-ray exposure, drugs, toxins and physical trauma. Amniocentesis was normal, sonography revealed no other congenital anomalies except for thoracic ectopia cordis, however, NT was elevated. Diagnosis of congenital heart anomaly was made by fetal echocardiography (figure 1and 2) including: complete thoracic heart ectopia in a male fetus, ASD, large inlet to outlet VSD with aortic overriding, DORV, anterior malposition of aorta, RVH, dilatation of LA resembling to aneurysmal change, pulmonary atresia with retrograde flow, narrow pulmonary artery increased turbulancy of ascending aorta and pericardium covering only ventricles which were confirmed on surgery. Given an unfavorable fetus prognosis during consultation with parents, they decided to terminate the pregnancy Unfortunately blood tests and genetic tests were not performed during D&C despite of our recommendation.

Table 1. Extracardiac anomalies associated with Ectopia cordis

Syndromes	Presentations
Pentalogy of Cantrell	Sternal, pericardial and diaphragmatic defects, omphalocele
Beckwith-Wiedemann syndrome	Organomegaly, polyhydramnios, macroglossia, large omphalocele
Amniotic band syndrome	Random defects, constriction rings, amputations bands
Limb-body wall complex	Complex-looking mass entangled with membrane, limb anomalies, spinal anomalies

Discussion

There are several theories about genesis of ectopic cordis but none of them are fully proven. Predominant theory states that lateral body folds cannot descent appropriately and midline fusion doesn't occur. Also, premature rupture of the chorion and/or yolk sac leads to failure of midline fusion at around 3rd week (5, 6, and 2). Early gestation rupture of the amniotic membrane explainthe amniotic band disruption. These bands result in amputation and intrauterine growth disruption. (4). Complete development of the thorax is necessary for normal descent of the heart. If this development doesn't happen it may lead to EC. Other mechanical restrictions explain heart defects. It has been stated that the initial characterization of the BMP2 (Bone Morphogenetic Protein 2 gene) knock out (KO) defects cause impairment in Ventral Folding Morphogenesis (VFM) such as the extra thoracic placement of the heart (4).

Other studies have stated that defect in the anterior body wall and the sternum result in lack of maturation of the midline mesodermal components and displacement of the heart outside of the thoracic cavity. The complexity and severity of cardiac disorders determine the prognosis of ectopic cordis (11). It has been also observed that the ultimate survival of these patients depends more on the presence or absence of intrinsic cardiac defects rather than surgical techniques. This disorder results in stillbirth or death in early neonatal period in most cases (9). Causes of cardiac complications or cardiac death include cardiac rupture,tamponade , endocarditis, arrhythmias and sudden cardiac death(4,5).

We think that our case was complicated because aorta and PA was under pressure of twisting and malpositioning of the heart, causing increased turbulent flow as well as dilatation of LA and left atrial appendage (LAA). Dilatation of LA and LAA might be an associated incidental finding or as a result of cardiac malposition as compressed by the xiphoid process (13). In addition to usual cardiac defects in EC, there are some other less common problems such as diverticulum of Left Ventricle (13 ,14) and Right Ventricle (13,15), and dilatation of Right Atrium (13) making them prone to sudden cardiac death due to spontaneous rupture or tachyarrhythmia (16). Increased morbidity might be explained by abnormal course and positioning of great vessels of the heart which result in kinking and further compromising circulation (18). In this disorder, the heart lost its protection by the sternum and the heart was predisposed to direct trauma. Other complication is frequent chest infections due to the paradoxical movement of the lungs.

The most common causes of death are cardiac failure, infection and hypoxemia,therefore, these events need appropriate and prompt therapy (4). The diagnosis can be conducted before second trimester between 9 and 13 weeks by sonography (2 and 3 dimensions). Given the high mortality rate of ectopia cordis, therapeutic abortion prior to the age of viability may be considered (10). The combination of MRI and fetal echocardiography are often used for follow-up of these cases if pregnancy is continued (8).

In these cases, due to risk of cardiac compression with vaginal delivery, caesarean should be planned (4). For determination of associated congenital disorders or abnormal Karyotype, amniocentesis and diagnostic tools may be considered (8) as it was done for our patient. Ectopia cordis is a rare congenital malformation which may require a staged procedure to achieve complete repair. The two tage surgical repair is usually recommended. In the first stage, surgery is done immediately after birth; the skin and soft tissue cover the heart with surgical process and with aim of prevention of fluid loss, cardiac desiccation, and trauma to the heart. The second stage is usually planned between 6 months and two years, the heart is returned to thoracic cavity and the anterior chest wall is reconstructed and cardiac defects re repaired (4,9,11). The single staged surgical repair is considered only for severe cardiac disease with hemodynamic instabilities (4).

In a study on seven fetuses with ectopic cordis (six pregnancies), abdominal wall defects were detected in six fetuses. Other abnormalities were Kyphoscoliosis, cephalocele, clubfoot and short umbilical cord. Five pregnancies were terminated, one fetus died in utero, and one neonate died on second day of life which emphasizes the poor outcome for these fetuses. After birth, cleft lip/palate was detected in two fetuses and tetralogy of Fallot in one (12). These finding were contrary to our study in which there were no extracardiac anomalies and abdominal wall defects as shown in fetal echo, sonography and amniocentesis (all were normal). An important problem with performing fetal echocardiography in ectopia cordis is presence of poor echo window resulting from cardiac malposition, squeezed by chest wall ostium and hypermobility due to lack of support of adjacent structures which makes the actual diagnosis more difficult. Thus, meticulous attention should be considered in this regard.

Our study had some limitations; we did not take blood test or genetic test for more evaluation. Also we could not take image of the stillbirth baby after termination of pregnancy or do autopsy.

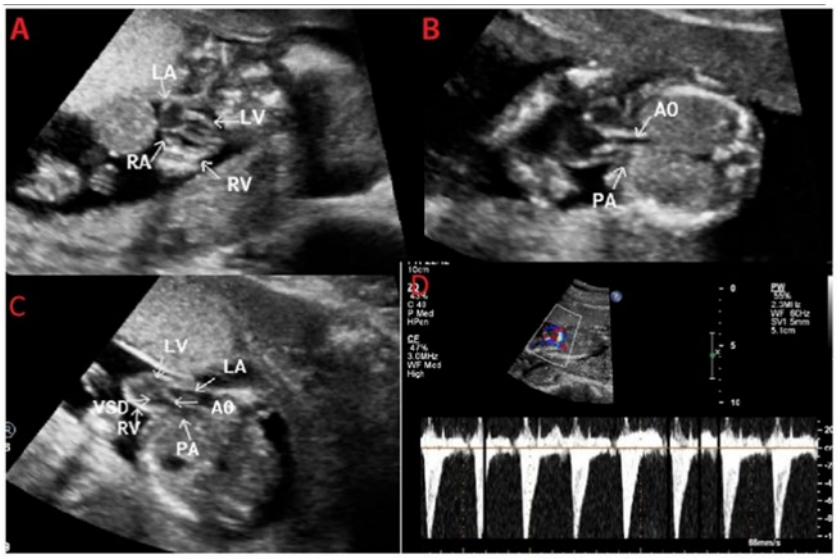

Figure 1: Fetal Echocardiography of isolated thoracic Ectopia Cordis (EC): A.4chamber view showing well developed Left Atrium(LA),Right Atrium(RA),Right Ventricle(RV) and Left Ventricles(LV) B and C. Double Outlet Right Ventricle and Ventricular Septal Defect(VSD), small pulmonary artery (PA) and PA branches, aortic overriding, anterior malposition of Aorta (AO) or D-Transposition of Great Arteries., LV: Left Ventricle, LA: Left Atrium. Doppler flow profile: Increased turbulancy of ascending aorta exiting from the heart entering toward the body.

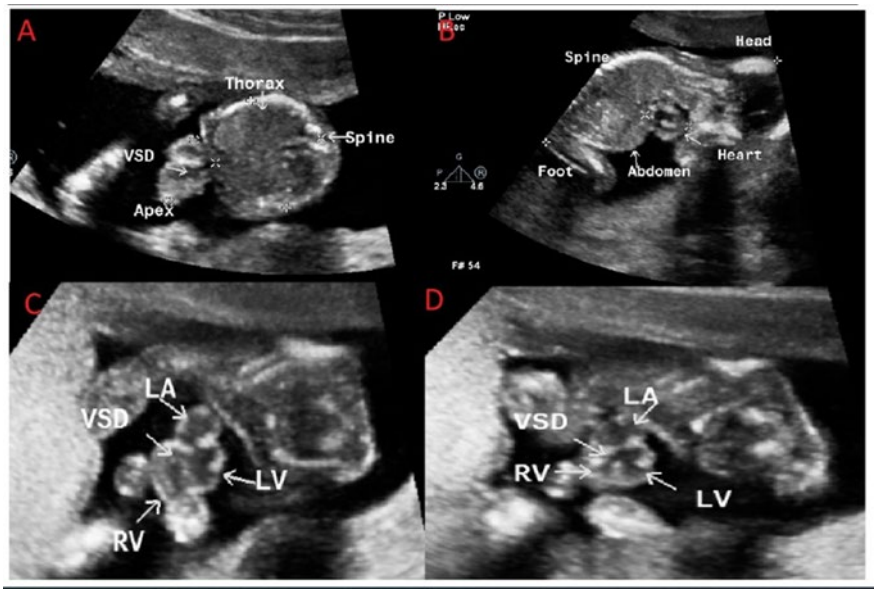

Figure 2 : Fetal Echocardiography :A.Short axis of heart and thorax: Thoracic form of Ectopia Cordis with written sizes of base to apex of heart(=1.8 cm) compared to short axis of thorax(=3.2cm x 2.7cm) .B. Long Axis of body:Thoracic Ectopia Cordis with written sizes of base to apex of heart (=1.75cm) compared to head to buttock(=9.54cm).C. Nearly 4 chamber view illustrating Right Ventricle(RV), Left Ventricle(LV), Ventricular Septal Defect(VSD) and dilated Left Atrium(LA) nearly in diastole. Nearly 4 chamber view illustrating Right Ventricle (RV), Left Ventricle (LV), Ventricular Septal Defect (VSD) and dilated Left Atrium (LA) in systole

Conclusion

Ectopia cordis is as a rare congenital malformation requiring comprehensive medical care and immediate surgical intervention.The prognosis is poor and related to associated cardiac and extracardiac anomalies and needs an appropriate approach with suitable timing. Given the high mortality rate of ectopia cordis, therapeutic abortion prior to age of viability may be considered.

Ethical Considerations

Compliance with ethical guidelines

The authors certify that they have received all appropriate consent forms. In this form, consent was given to report images and other clinical information in the journal. The patient knows that their names and initials will not be published and that every effort will be made to conceal their identity.

Funding

This research did not receive any grant from funding agencies in the public, commercial, or non-profit sectors.

Conflict of interest

The authors declared no conflict of interest.

Acknowledgements

The authors appreciate the insightful cooperation of pediatric cardiac echocardiography staff of Shahid Rajaee Hospital

References

[1]Taksande MA, Vilhekar YK. A case report of ectopia cordis and omphalocele. Indian J Hum Genet 2013 Oct-Dec; 19(4): 491–493.

[2]Ramasauskaite D, Snieckuviene V, Zitkute V, Vankeviciene R, Lauzikiene D, and Drasutiene G. A Rare Case Report of Thoracic Ectopia Cordis:An Obstetrician's Point of View in Multidisciplinary Approach .Case Reports in Pediatrics 2016, Article ID 5097059 | https://doi.org/10.1155/2016/5097059(19804 Jul 2016).

[3]Pannová J, Kováčová H, Matura D, Gruszka T, Pavlíček J, Delongová P. Ectopia Cordis-Case Report. Ceska Gynekol, 2015 Jun; 80(3):214-217.

[4]Abigail G, Donnelly J, Kuc A, Daniel G, Doros G, Matusz P, Loukas M.Ectopia Cordis: A Rare Congenital Anomaly . Clin. Anat. 2014; 27:1193– 1199

[5]Oumarou H, Amadou MI, Harissou A, Ousseini A.Ectopia Cordis: A Case Report and Review of Literature. Journal of Case Reports, 2017;7(2):143- 145

[6]Engum SA, "Embryology, sternal clefts, ectopia cordis, and Cantrell's pentalogy".Seminars in Pediatric Surgery, 2008(17);no.3: 154–160.

7] Agrawal SN, JoshiVK, Sanapale S,Tiwari PC. Ectopia cordis: a case report. International Surgery Journal 2018 Feb;5(2):739-742.

[8]Madhavi D, and Rajasree TK. Thoracic Ectopia Cordis - A CASE REPORT IJBR, 2012;3(01):69-73.

[9]Apte AV. Thoraco-Abdominal Ectopia Cordis: A Rare Entity. Case Report and Review of Literature People's Journal of Scientific Research 2008; July (1):31-33.

[10] Barakat A Animasahun,Akpoembele D Madise W.Thoracic Ectopic Cordis in a Nigerian Child. Journal of Cardiology & Current Research, 2016;6(6):00227.

[11] Malik R1, Zilberman MV, Tang L, Miller S, Pandian NG. Ectopia Cordis with a Double Outlet Right Ventricle, Large Ventricular Septal Defect, Malposed Great Arteries and Left Ventricular Hypoplasia. Echocardiography, 2015 Mar;32(3):589–591.

[12] .Turkyilmaz G, Avci S,Sivrikoz T,Ertrk E,Altunoglo U,Turkyilmazlmaz SE,Kalelioglu IH,Has R,Yuksel A.Prenatal Diagnosis and management of Ectopia Cordis:Varied Presentation Spectrum. Journal of Fetal and Pediatric pathology, 2019(38), Issue 2:127-137.

[13] Piraste A,Carcano C,Kirsch J,Mohammed TLH. Pentalogy of Cantrell with ectopia cordis:CT findings. Journal of radiology case reports, 2014;8(12):29-

[14] Madi JM,José Festugatto JR, Rizzon M, Agostini AP, de Araújo BF,Rahmi Garcia RM.Ectopia Cordis associated with pantalogy of Cantrell,A case report. Rev. Bras. Gynecol. Obstet. 2019;41(5):352-356.

[15] Mendoza J. Cardiac abnormality ectopia cordis. Internal J of pregnancy and child birth, 2020;6(1)

[16] Hakim K,Ayari J,Jebri F,Msaad H,Ouarda F,Chaker L,Boussaada R. Cardiac anomalies in cantrell's pantalogy from ventricular diverticulum to complete thoracic thoracic ectopia cordis. Cardiol Tunis, 2013; 9(1): 94–97. 237

[17] Shad J,Budhwani K,Biswas R.Thoracic ectopia cordis. BMJ casereports 2012 http://dx,doi.org/10.1136/bcr.11.2011.5241.PMCID: PMC4543290, PMID:23035158(published online 2012 sep 30)