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Antenatal Diagnosis of a Case of Ectopia Cordis with Complex Congenital Heart Defects in a Resource Limited Set Up in a Remote Part of Eastern India

Case Report

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Abstract

Ectopic cordis is a rare congenital anomaly. Here, the fetal heart is displaced outside the thoracic cavity partially or completely. We describe a case of antenatal diagnosis of Thoracic Ectopia Cordis (EC) with congenital heart disease (CHD) without any other extracardiac malformations. Fetal Echocardiographic examination showed large VSD, double outlet right ventricle and unrestricted pulmonary and systemic blood flow. The diagnosis of Ectopia cordis and associated cardiac and non-cardiac anomalies may be difficult in the fetal period due to multiple factors, so meticulous attention should be paid for proper diagnosis.

Introduction

Ectopia cordis (EC) is a rare congenital anomaly with an incidence of 5.5 to 7.9 per one million live birth. In this condition, the foetal heart is projected outside the thoracic cavity either partially or completely. It contributes around 0.1% to congenital heart disease (CHD). Infants with EC usually have a poor prognosis. Factors affecting prognosis include type of EC and severity of other cardiac and extracardiac congenital anomalies. [1, 2]. It affects all races equally with male preponderance [3, 4].

Most newborns (90%) with EC are born with CHD. The most frequent being ventricular septal defect (VSD) in 59% of cases. Other anomalies include pulmonary atresia or stenosis, atrial septal defect, tetralogy of Fallot, right ventricular diverticulum, double outlet right ventricle (DORV), complete transposition of great arteries, and rarely atrioventricular septal defect. [4]

The EC may be associated with other midline defects called the Pentalogy of Cantrell syndrome. This syndrome consists of thoraco-abdominal EC, anterior diaphragmatic hernia, lower sternal defect, midline supra-umbilical defect (omphalocele) with associated pericardial and intra-cardiac defects. [5,6] Ectopia cordis may also be accompanied with few extracardiac anomalies like amniotic band syndrome, cleft lip and palate, omphalocele, diaphragmatic hernia, body stalk syndrome and skeletal malformations such as kyphosis. [7] Normal karyotype is most often reported in screened patients, however, abnormal karyotypes like XXY, trisomy 18 and trisomy 21 and Turner syndrome are also been reported. [4, 8]

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Case Report

A 22-year-old woman (Gravida3, para 2) was referred for fetal echocardiographic evaluation at 22nd weeks of gestation. The pregnancy was spontaneous. She had regular prenatal care including folic acid supplementation after pregnancy confirmation. There was history of two abortions, but no family history of congenital heart disease, genetic disorders or any anomaly related to ectopia cordis. There was also no history of exposure to radiation, teratogenic drugs, toxins or any physical trauma. Routine sonography and anomaly scan at 20th weeks of gestation revealed thoracic ectopia cordis. Diagnosis of cardiac defects associated with EC was made by fetal echocardiography. It showed complete thoracic EC, large malalinged VSD, double outlet right ventricle i.e. both the great arteries arising from right ventricle (anterior ventricle with an AV valve placed more apically) with aortic override, unobstructed pulmonary and systemic outflow, disproportionately dilated right atrium and ventricle with a large patent foramen ovale. (Figure 1-3) Parents were explained in details about the anomalies, multi staged treatment plans and possible outcome. After detailed discussion with obstetrician, parents opted for abortion of the foetus, but they did not agree to do any genetic testing and autopsy of the abortus. We have taken written consent from the patient before publishing this case.

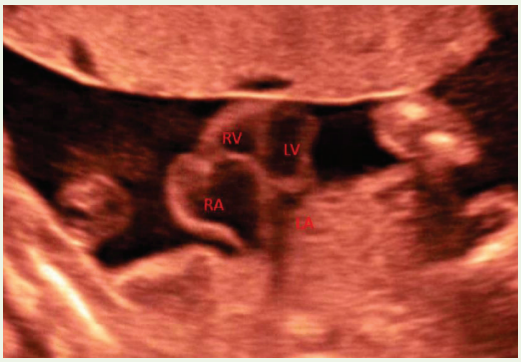

Figure 1: Heart outside the chest cavity

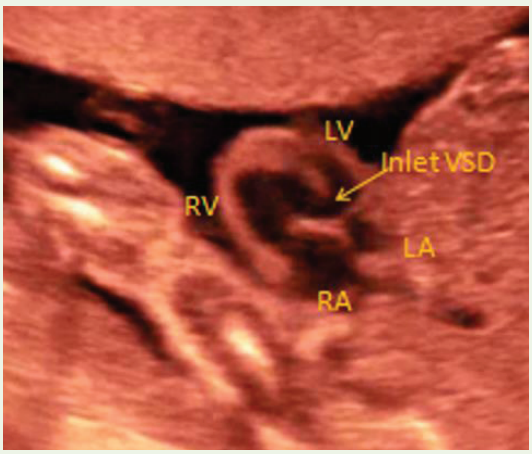

Figure 2: Heart outside chest cavity showing large inlet VSD

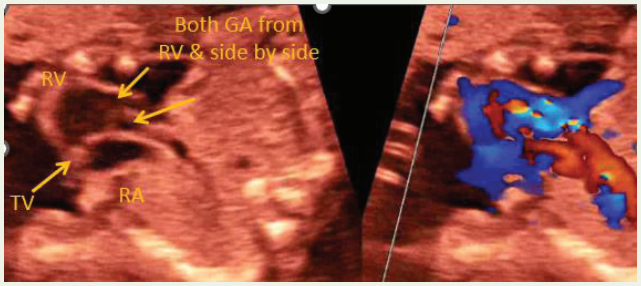

Figure 3: Double outlet right ventricle with un obstructed systemic and pulmonary output, both great arteries seen in a single plane; and parallel; arising from anterior ventricle

Discussion

There are several theories about the genesis of ectopia cordis but none of them are fully proven. Most of the theories state that lateral body folds cannot descend 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 of gestation. [2,8,9]It has been stated that Bone Morphogenetic Protein 2 gene (BMP2) knock out (KO) defects cause impairment in Ventral Folding Morphogenesis (VFM) resulting in extra thoracic displacement of the heart. [4] The complexity and severity of associated cardiac disorders determine the prognosis of EC. [10]Here the heart lost its protection by the sternum and predisposed to direct trauma. Another complication is frequent chest infections due to the paradoxical movement of the lungs. [4,8] This anomaly results in stillbirth or death in the early neonatal period in most cases. [7]

Given the high mortality rate of EC, therapeutic abortion prior to the age of viability may be considered in most cases. [10] In our case, therapeutic abortion before viability was chosen by the parents. The combination of fetal MRI and fetal echocardiography are often used for follow-up of these cases if pregnancy is continued. [6]

In a study on seven fetuses with ectopic cordis (out of six pregnancies), abdominal wall defects were detected in six fetuses. Other abnormalities were kyphoscoliosis, encephalocele, clubfoot and short umbilical cord. Five pregnancies were terminated, one died in utero, and others died on the second day of life. This emphasized the poor outcome of EC. [11]

A major difficulty of performing foetal echocardiography in EC is the poor echo window resulting from cardiac malposition, squeezing of heart by chest wall ostium and hypermobility due to lack of supporting adjacent structures. This makes the actual diagnosis more difficult. [6]

Conclusion

The awareness among obstetricians and parents about the utility of doing timely fetal echocardiography is increasing nowadays, even in the remote parts of eastern India. As a result, complex congenital heart defects are diagnosed before reaching viability. As a result, parents can get time to take the decision regarding the continuation of pregnancy after getting informed about the disease and its prognosis in detail.

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