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A RARE CASE OF COMPLETE ECTOPIA CORDIS WITH COMPLEX CONGENITAL CARDIAC ANOMALIES

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ABSTRACT

Ectopia cordis is a very rare form of congenital malformation in which the heart is located outside the thorax, partially or totally. This abnormally located heart may be found in different anatomical locations, such as thoracic, abdominal, thoraco-abdominal, cervical or cervicothoracic. In majority, the heart projects outside the chest through a breach in sternum. This unique rare case report presents a male infant born at a peripheral hospital which was later referred to our tertiary care cardiac research institute. In our case thoracic ectopia was associated with complex cardiac anomalies. The baby had good APGAR scores at birth. The baby died 10 days after birth as a result of cardiac failure and septicaemia.

INTRODUCTION

Ectopia cordis is very rare with incidence varying from 5.5 to 7.9 per 1 million live births [1]. The survival beyond neonatal age is extremely rare without surgical treatment [1]. Apart from ectopic heart being more vulnerable to infections and trauma, complications like endocarditis, pericarditis, thromboembolism, arrhythmias cardiac rupture leading to tamponade and sudden death are also observed [2]. This condition has male preponderance. Though the aetiology is unknown, it is believed that failure of the development of midline mesoderm and ventral body wall during fetal development causes ectopia cordis. Most of the foetuses with this condition may result in spontaneous abortion in early weeks of pregnancy. The anomaly can be detected by antenatal ultrasound as early as 9th to 12th week of the pregnancy [3-5]. MRI is increasingly being used in antenatal detection. This abnormally located heart may be found in different anatomical locations, such as: thoracic (60%), abdominal (30%), thoraco-abdominal (7%), cervical (3%) and cervicothoracic (<1%) [2, 6, 7].

Case Summary

A 2 day-old male neonate born at a peripheral hospital was referred to our tertiary care cardiac research institute with complete thoracic ectopia cordis. The mother and father were illiterate labourers from poor economic sections of the society. The mother had not undergone any prior antenatal ultrasound examination. No family history of congenital malformations was found. The baby was breast fed by mother after 30 minutes of delivery as per guidelines.

The patient was vitally stable at the time of admission and had normal APGAR scores at the time of birth. The exposed heart was covered with saline-soaked gauze pads wrappings to prevent desiccation and heat loss [8]. The baby was admitted in pediatric ICU department and underwent echocardiography which revealed associated complex congenital cardiac anomalies. For better understanding of the complex anatomy it was decided to perform computed tomography angiography (CTA) before the decision of surgery.

CT findings (Figures 1 – 4) revealed ectopia cordis thoracic with absent sternum. Complex cardiac abnormalities included superior-inferior relation of ventricles with double outlet of morphological right ventricle, D-posed aorta, large PDA and changes of pulmonary hypertension. Atrial septal defect and perimembranous ventricular septal defect were present. Pulmonary venous drainage was normal. Persistent left SVC was noted. The great vessels were longer in length than normal.

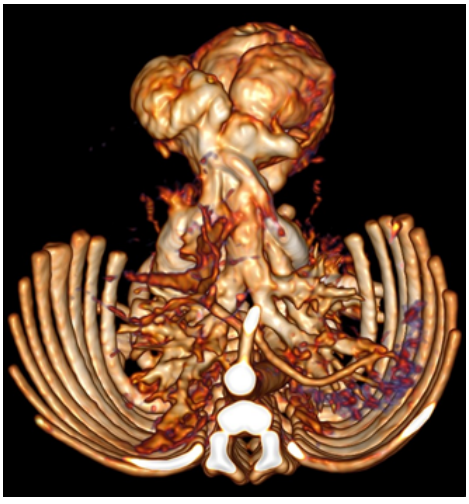

Figure 1. Volume-rendered (VR) CT image (caudal view) showing ectopia cordis with absent sternum.

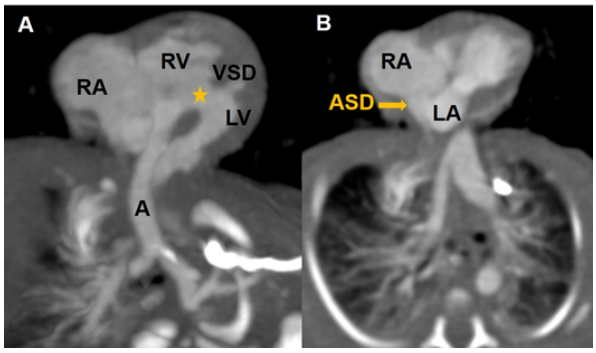

Figure 2. Ectopia Cordis. Axial oblique maximum-intensity-projection (MIP) CT images show VSD (yellow star) in image A) and ASD (yellow arrow) in image B). RA = Right atrium. LA = Left atrium. RV = Right ventricle. LV = Left ventricle. ASD = Atrial Septal Defect. VSD = Ventricular Septal Defect. A = Aorta.

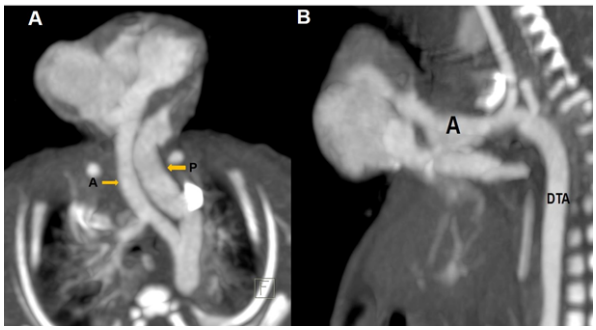

Figure 3. Ectopia Cordis. A) Axial and B) Sagittal oblique maximum-intensity-projection (MIP) CT images show abnormally long great vessels and their relationship. A = Aorta. P = Pulmonary artery.

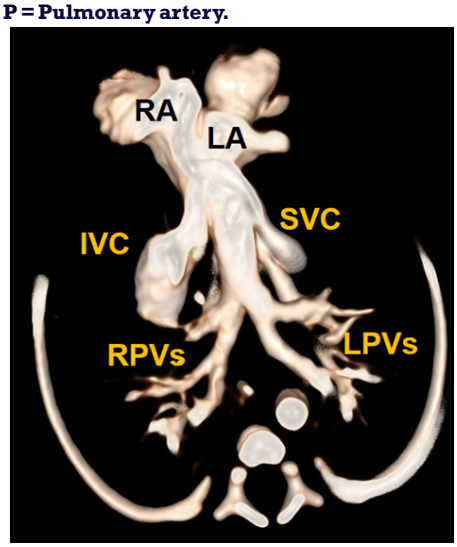

Figure 4. Ectopia Cordis. Volume-rendered (VR) CT image shows systemic venous drainage to respective atria. RA = Right atrium. LA = Left atrium. SVC = Superior Vena Cava. IVC = Inferior Vena Cava. RPVs = Right side pulmonary veins. LPVs = Left side pulmonary veins.

DISCUSSION

Ectopia cordis word is an amalgamation of two words from two different languages ("Greek: ectopia" and "Latin: cordis"). It simply means the heart is outside its normal position. There is herniation of heart through a defect in the anterior chest wall [9].

Embryologically, failure of the sternal fusion results in a variety of cleft sternal disorders, ranging from complete to partial ectopia cordis. Midline fusion and formation of the thoracic and abdominal cavities are complete by the 9th embryonic week [10]. Aberrations of midline fusion at this stage results in ectopia cordis. According to amniotic rupture theory, during early embryonic development, the amnion surrounding the embryo ruptures causing stringy, sticky, fibrous bands of amnion becoming entangled with forming embryo resulting into disruption article of fetal development leading to deformities such as ectopia cordis, midline sternal cleft, frontonasal dysgenesis, mid facial cleft, and limb deformities, among others. The spectrum of defects corresponds to the timing of rupture. Abdominal ectopia cordis has a slightly better prognosis, possibly because less incidence of coexisting intracardiac abnormalities [11].

Based on aberrations in the development, two subtypes are described.

1] As an outcome of embryological developmental failure of a segment of the mesoderm consisting of primary three defects. [12-14].

- Defect in the Diaphragm (due to partial/complete failure of development of transverse septum)
- Defect in the Pericardium (due to aberrant development of transverse septum)
- Intracardiac anomalies (due to faulty development of epicardium and/or myocardium- derivatives of splanchnic mesoderm)

2] Other subtype consists of sternal and abdominal wall defects (due to failure of migration of paired primordial structures). Variants of Pentalogy of Cantrell occur due to these defects [12, 14]

This abnormally located heart may be found in different anatomical locations, such as: thoracic (60%), abdominal (30%), thoraco-abdominal (7%), cervical (3%) and cervicothoracic (<1%). Table 1 shows subtypes of ectopia cordis [15]. Table 2 describes associated cardiac and non-cardiac defects.

Table 1: Subtypes of the ectopia cordis.

Subtype	Description
Partial ectopia cordis	The heart is located outside the chest wall but is covered by skin, pericardium. Heart can be seen beating through the skin.
Complete ectopia cordis	The heart is not covered by sternum, pericardium and lies unprotected completely outside the thoracic cavity.

Table 2: Congenital defects associated with ectopia cordis.

Intracardiac defects	Atrial septal defect Ventricular septal defect Tetralogy of Fallot Tricuspid atresia Double outlet right ventricle
Non-cardiac malformations	Pentalogy of Cantrell Omphalocele Anterior diaphragmatic hernia Cleft palate

Definitive treatment is multistage surgical repair. Repositioning of the heart results in elevated intrathoracic and abdominal pressures in postoperative period [16]. The great vessels are longer in length than normal which result in their kinking during repositioning. These abnormally long vessels may even compress the heart compromising cardiac output [17]. Differential diagnosis are as shown in Table-3 [18, 19].

Table 3: Differential diagnoses of ectopia cordis

Pentalogy of Cantrell (also considered thoracoabdominal ectopia cordis)	Abdominal wall defect Lower sternal defect Congenital heart malformations Absence of the diaphragmatic pericardium Anterior diaphragmatic defect
Amniotic band syndrome	Amputations, Bands, Random defects, Constriction rings
Cyllosomas-Limb body wall complex	Limb anomalies, Spinal anomalies, Complex-looking mass entangled with membrane.
Beckwith-Wiedemann syndrome	Macroglossia, Large omphalocele, Organomegaly, Polyhydramnios

CONCLUSION

Ectopia cordis is an extremely rare congenital anomaly with very poor survival rates. Very few surgical procedures are available for management without proven efficacy due to rare nature of disease. The outcome also depends on associated cardiac and extracardiac anomalies. There are rare cases of neonates with ectopia cordis surviving who had normal heart anatomy and functionality. The patient in our case could not undergo surgery and died on 10th day of life. Most cases of ectopia cordis die in early stage of life due to infection, septicaemia and heart failure.

Conflicts of Interest

The authors declare that they have no conflicts of interest.

Acknowledgments

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