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A Case Report of Ectopia Cordis with Omphalocele

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Abstract **Case Report**

Ectopia cordis (EC) is defined as complete or partial displacement of the heart outside the thoracic cavity. It is a rare congenital defect in fusion of the anterior chest wall with an estimated prevalence of 5.5 -7.9 per million live births. We discuss one such of case of a 2-days old preterm female neonate weighing 1.3 Kgs with ectopia cordis and omphalocele. The patient died of cardiorespiratory arrest 1 day after attempt at surgical intervention was abandoned due to hemodynamic instability intra-operatively.

Keywords: Ectopia cordis, Thoracic cavity, Pentalogy of Cantrell.

INTRODUCTION

Ectopia cordis is a rare congenital malformation in which the heart is located partially or totally outside the thoracic cavity. It may occur as an isolated malformation or it may be associated with body wall defects of the thorax, abdomen or both. Ectopia cordis was first observed 5000 years ago and the term ectopia cordis was coined by Haller et al. in 1706. It can occur as a component of pentalogy of Cantrell which was described by James R. Cantrell in 1958. The malformation can be detected with antenatal ultrasound by 9th to 12th week of pregnancy. The treatment usually consists of a staged surgical repair. However, despite advances in neonatal cardiac surgery, complete ectopia cordis remains a surgical challenge with poor prognosis. Here we present a case report of Ectopia cordis with Omphalocele highlighting the importance of imaging in the detection as well as in assessment of the full extent of the pentalogy of Cantrell.

CASE REPORT

Two days old female baby born as twin to a primigravida mother of 31 years at 32 weeks of gestation. The baby was delivered by spontaneous vaginal delivery weighing 1.3 Kgs and immediately cried after birth, however, due to low APGAR and poor respiratory pattern the baby was intubated. On examination ectopia cordis was noted with heart completely outside the thoracic cavity and a midline anterior abdominal wall defect. Pulse rate 164/minute, respiratory rate 60/minute, Spo2 97%, RS and CNS examination was within normal limits.

A bedside echocardiography showed a large subaortic VSD with overriding of aorta >50%. Main pulmonary artery was seen arising from the right ventricle. The pulmonary valves were mildly thickened showing laminar flow. Both the great vessels were parallel with no crossover.

X-ray AP view of chest and abdomen showed an abnormal midline cardiac position with air-lucency around the ectopic heart.

Contrast enhanced CT scan of the thorax and abdomen showed ectopia cordis with a large midline sternal defect measuring approximately 3.2 x 2 cm (TR x SI) in size. A near contiguous abdominal wall defect was noted in the epigastric quadrant measuring approximately 4 x 2.9 cm (TR x SI) in size. Herniation of part of the liver was noted through this defect together with the left branch of portal vein and umbilical vein. Situs solitus was noted. Superior and inferior vena cava was seen draining into the right atrium. Two superior pulmonary veins were seen draining into the left chamber. Left sided aortic arch was noted with normal calibre of ascending, arch and descending aorta. The pulmonary vasculature appeared normal (pulmonary trunk -4.2mm, RMPA-2.7 mm and LMPA-2.6 mm). Lung parenchyma showed lower lobar collapse-consolidation and subsegmental atelectasis in both upper lobes. Bilateral pleural effusion was noted around both lower lobes.

Reduction of ectopia cordis with skin closure was planned under general anaesthesia. Baby was

operated on the 3rd day. However, in view of hemodynamic instability intraoperatively, the procedure was abandoned.

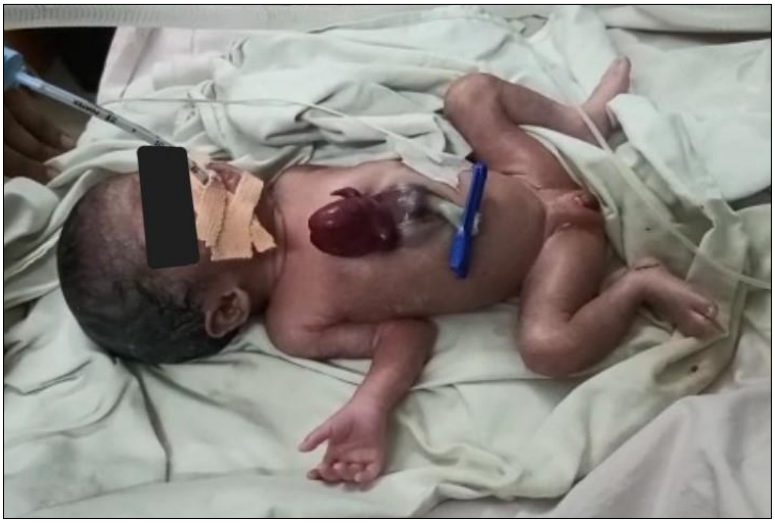

Fig. 1: Patient has an externally visible heart with no skin or pericardial covering (complete ectopia cordis). Omphalocele is present inferiorly to the ectopic heart with umbilical cord inserted at the inferior end of abdominal wall defect.

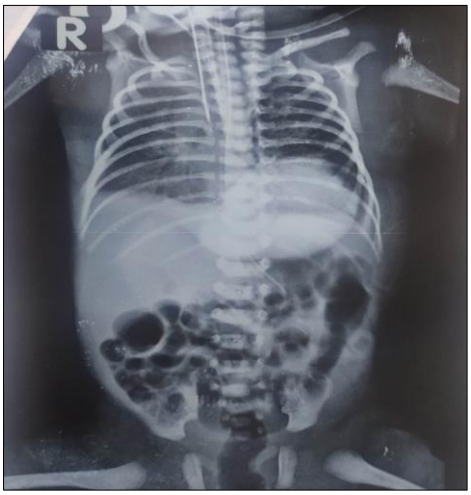

Fig. 2: CXR of ectopia cordis shows the heart as an ovoid opacity with an abnormal cardiac position. A deficient sternum is also noted.

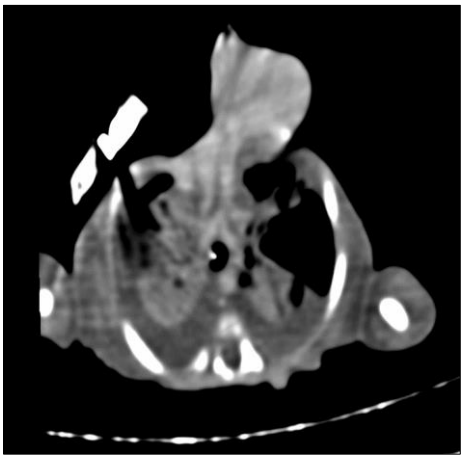

Fig. 3: Axial image of ectopia cordis shows heart extending through a midline defect in the sternum and lying outside the thorax.

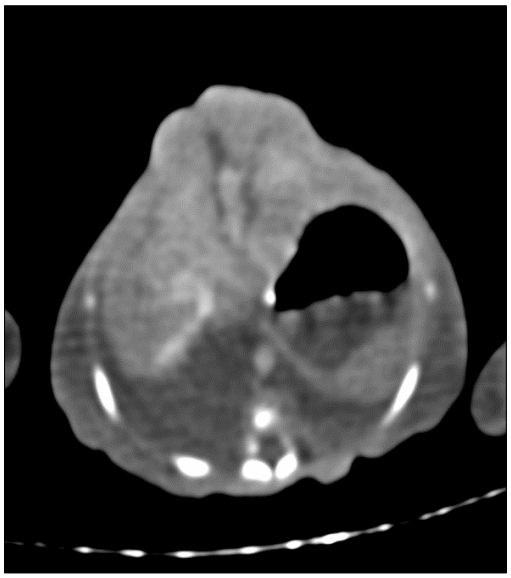

Fig. 4: Axial image of omphalocele showing herniation of part of the liver and left branch of portal vein.

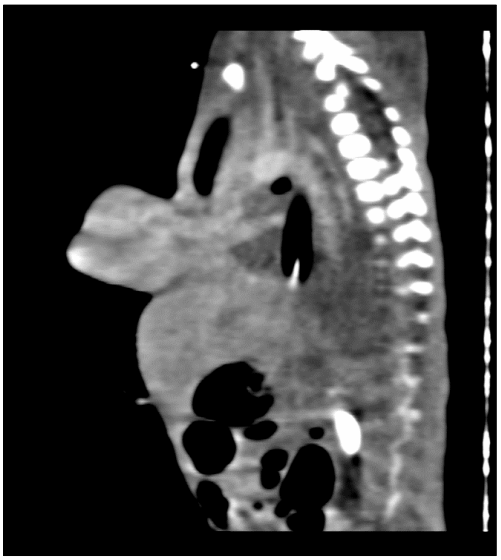

Fig. 5: Sagittal image showing ectopia cordis and omphalocele.

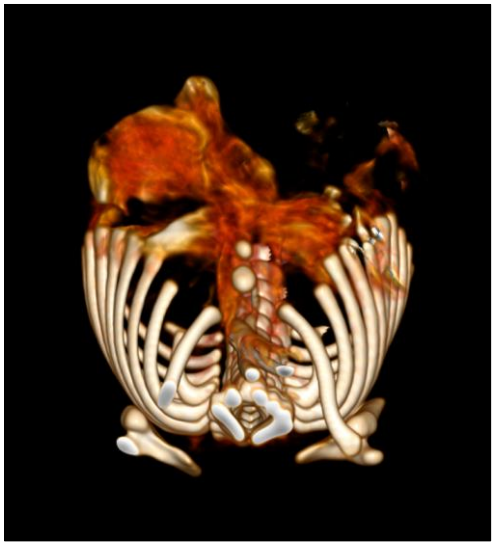

Fig. 6: Volume-rendered (VR) CT image (cranial view) showing ectopia cordis with deficient sternum.

Table 1: 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	Omphalocele Anterior diaphragmatic hernia Cleft palate

Table 2: Differential diagnosis of ectopia cordis

Pentalogy of Cantrell	Omphalocele with a midline defect at the umbilical cord insertion.
Body stalk anomalies	Eccentric large lateral defect and adherence of the placenta to the defect.
Amniotic band syndrome	Ventral wall defect with extremity deformity with an adherent band.

DISCUSSION

EC is defined as complete or partial displacement of the heart outside the thoracic cavity. It is a very rare anomaly with an estimated prevalence of 5.5 -7.9 per million live births [3]. It is related to the malformation of the anterior wall of the thorax, with an extra thoracic location of the heart. Cantrell described this syndrome which can occur with variable degrees of expression. The five defects described by Cantrell are as follows (i) Supraumbilical omphalocele (ii) Lower sternal cleft (iii) Defect in diaphragmatic pericardium – *leading to ectopia cordis* (iv) Anterior diaphragmatic defect(v) Congenital heart malformations [6].

Depending upon the location of the heart, it can be classified into five types: cervical (5%), cervicothoracic and thoracic (65%), thoracoabdominal (20%) and abdominal (10%). Ectopia cordis can also be classified into partial or complete ectopia cordis. In partial ectopia cordis, the heart is located outside the chest wall but is covered by skin and pericardium. Heart can be seen beating through the skin. In complete ectopia cordis, the heart is not covered by sternum or pericardium and lies unprotected completely outside the thoracic cavity [1,3,5].

The development of the ventral body wall begins by 8th day of embryonic life with differentiation and proliferation of mesoderm followed by its lateral migration. Midline fusion and formation of the thoracic and abdominal cavities is completed by the 9thembryonic week [1,3,5]. Complete or incomplete failure of midline fusion at this stage result in disorders varying from isolated EC to complete ventral evisceration [3,6].

Theories for genesis of EC include amnion rupture theory and amniotic band syndrome. The amnion rupture theory states that during early embryonic development, the amnion surrounding the embryo ruptures, and stringy, sticky, fibrous bands of amnion becomes 'entangled' with the forming embryo and causes a disruption in the developing parts of the fetus which may lead to various deformities like EC, midline sternal cleft, frontonasal dysgenesis, a midfacial cleft, limb deformities etc [1,3,5]. The spectrum of defect corresponds to the timing of its rupture. EC with amniotic bands appears to have aetiology distinct from isolated EC.

The differential diagnosis of ectopia cordis includes isolated thoracic ectopia cordis, amniotic band syndrome and body stalk anomalies. The key features for distinguishing these conditions is the position of abdominal wall defect in relation to the umbilical cord insertion, eviscerated organs, the presence or absence of membranes or bands, and associated anomalies [2].

The prenatal diagnosis of EC is made with ultrasound by visualizing the heart outside the thoracic cavity. In view of the poor prognosis, termination of pregnancy can be considered if ultrasound diagnosis is made before viability. The prognosis depends on the degree of the intracardiac involvement and associated malformations, as well as the degree to which the heart is exposed. Most infants are stillborn or die within the first hours or days of life. Attempts at surgical correction are widely performed, with immediate covering of the heart and exposed abdominal contents using silastic prosthesis being recommended [2]. Definitive treatment is a multistage surgical repair.

CONCLUSION

Ectopia cordis is an extremely rare congenital anomaly with very poor survival rates. In utero and neonatal imaging studies play a key role in not only prenatal counselling, but also in assisting surgeons to formulate better operative plans for patients with this condition [4]. The outcome also depends on associated cardiac and extra-cardiac anomalies. Most cases of ectopia cordis die in early stage of life due to infection, septicaemia and heart failure [5].

While some suggest early intervention, others believe a step-wise procedure should be followed to manage this condition. While the physical exam may reveal the apparent defects in the ventral abdominal wall, imaging is necessary to assess the full extent of the pentalogy of Cantrell [6].

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