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**Case Report**

# Ectopia cordis: a case report

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## ABSTRACT

The Ectopia cordis is a rare congenital condition. It is characterized by the abnormal position of the heart outside the thoracic cavity, associated with defects in the parietal pericardium, diaphragm, sternum, and, in most cases, cardiac malformations. The reported prevalence is 5.5 to 7.9 per million live births. The designation Ectopia cordis was first proposed by Abott in 1998, although cases of patients with similar defects had been reported in past. Only 267 cases have been reported as of 2001, most (95%) associated with other cardiac abnormalities. Ten hours old, normal, vaginally delivered, full-term male neonate, weighing 2020 grams was brought with an externally visible, beating heart over the lower chest wall. The antenatal period was uneventful, though no ultrasonography was done. The beating heart was covered with a membrane with greenish purulent matter over it, most probably, due to pseudomonas/proteus infection. Rest of the anterior abdominal wall was intact. Ectopia cordis is produced by segmental defects in mesodermal development in the third week of intrauterine life, and/or amniotic band syndrome that causes simultaneous cerebral and Thoraco-abdominal malformations. The existence of Ectopia cordis with severe congenital heart disease may be confirmed in the prenatal period by vaginal echocardiography at 10-12 weeks of gestation or by abdominal echocardiography by 20-22 weeks. Surgery is the only hope of survival, for such patients, although the overall success rate is very poor. In recent years, surgery has been attempted in one or two phases with variable results that depend, mainly on the type of associated heart disease.

**Keywords:** Ectopia cordis, Pentology of Cantrell, Tetralogy of fallots, Ventricular septal defect

## INTRODUCTION

The word Ectopia is derived from Greek ektopos, meaning away from place, and it implies an abnormality in the position of organ or a part of the body, often congenital in origin.

Ectopia cordis is a rare disease that is defined by the abnormal position of the heart outside the thoracic cavity, associated with defects in the parietal pericardium, diaphragm, sternum, and, in most cases, cardiac malformations. The reported prevalence is 5.5 to 7.9 per million live births.[1-3] It is generally a sporadic malformation, with reports, linking it to chromosomal abnormalities like trisomy 18 and turner syndrome and 46,XX,17q+.[4-6] The designation Ectopia cordis was first proposed by Abott in 1998, although cases of patients with similar defects had been reported in past.[7]

The condition is a rare congenital anomaly, the nomenclature is confusing, and pathology is so obscure that that it seems justifiable to report further cases. In this paper an attempt is made to present this rare congenital anomaly with discussion of relevant literature.

***History***

Ectopia cordis, first observed 5000 years ago.[8] As described by Engum and Kaplan et al, the first report of EC was by Haller in 1706.[9-10] Byron classified Ectopia cordis into four types: cervical, Thoraco-cervical/thoracic, Thoraco-abdominal and abdominal.[11]

*Cervical:* The heart is entirely in cervical region and the sternum is usually intact (5%).

*Thoraco-cervical/thoracic*: The Heart is partially in the cervical region and the cranial end of the sternum is defective. In the thoracic variety sternum is defective and the heart lies outside the thorax, partially or completely (65%).

*Thoraco-abdominal*: this subdivision was first suggested by Byron (1948); the condition should be partially accompanied by a diaphragmatic defect, and by a midline abdominal defect of diastases recti or omphalocoel (20%).

*Abdominal*: This entails a diaphragmatic defect allowing the heart to enter the abdominal cavity (10%).

In 1958, Cantrell reported a syndrome with five defects, which constitutes pentology of Cantrell. It is the combination of Thoraco-abdominal EC, lower sternal defect, anterior diaphragmatic hernia, midline supraumblical defect along with pericardial and intra-cardiac defects.[12-14] Because of its rarity and associated abnormalities, EC is a challenging congenital anomaly. As of 2001, 267 patients have been reported.[15-16] Ninety percent of the infants died in the first year of life. Cases of the cervical type rarely survived a single day. Most (90%) newborns have associated cardiac malformations. The most frequent cardiac malformation is ventricular septal defect, which is present in the 59% of cases, followed by atrial septal defect in 35%, pulmonary stenosis or atresia in 36%, tetralogy of fallots in 22%, right ventricular diverticulum in 13%, left superior vena cava in 12%, and double right ventricular outflow tract in 13%. Other cardiac malformations like single ventricle, complete transposition of great arteries, and atrio-ventricular septal defect, occur only rarely.

***Embryology***

The heart develops as paired primordia in the visceral mesoderm overlying the anterior end of the yolk sac, while the parietal pericardium and septum transversum are formed simultaneously from the somatic mesoderm adjoining the developing heart. With the folding of the embryo the paired cardiac tubes come to lie in the ventral and cranial part of the foregut and fuse to form the single heart tube. Similarly, a single pericardial cavity is soon established and the septum transversum now comes to lie caudal to heart. The septum transversum forms the major part of the diaphragm.

The development of the ventral body wall begins by eighth day of embryonic life with differentiation and proliferation of mesoderm followed by its lateral migration. The heart originally develops in a cephalic location and reaches its definitive position by the lateral folding and ventral flexing of the embryo at about 16$^{th}$-17$^{th}$ day. Midline fusion and formation of the thoracic and abdominal cavities is complete by the 9$^{th}$ embryonic week.[17-18] Complete or incomplete failure, of midline fusion at this stage, results in disorders varying from isolated EC to complete ventral evisceration. Genesis of EC has not been fully explained, although several theories have been offered. Popular theories are early rupture of the chorion and/or yolk sac, and amniotic band syndrome.[19-21]

The amnion rupture theory states that during early embryonic development, the amnion surrounding the embryo ruptures, and stringy, sticky, fibrous bands of amnion become 'entangled' with the forming embryo, and causes a disruption in the developing parts of the foetus which may lead to various deformities like EC, midline sternal cleft, front nasal dysgenesis, a midfacial cleft, limb deformities etc. The spectrum of defect corresponds to the timing of its rupture. The findings in the literature suggest that its rupture in the third week of gestation causes an arrest of cardiac descent which may be the cause of such defects. EC with amniotic bands appears to have aetiology distinct from isolated EC. This suggests several different etiologies for EC.

Various diagnostic techniques are now available to screen for EC and PC. The prenatal diagnosis of these two defects is typically noted at the beginning of the second trimester; the earliest diagnosis was made at 10 weeks of gestation using Doppler sonography.[22]

The use of three-dimensional ultrasound and its combination with doppler allows for a more accurate early diagnosis. Magnetic resonance imaging is also becoming commonplace in prenatal evaluation to document and plan for management of complicated congenital anomalies.[23]

## CASE REPORT

Ten hours old, normal vaginally delivered, full-term male neonate, weighing 2020 grams was brought with an externally visible, beating heart over the chest wall. There was no history of consanguineous marriage, infection, intake of any teratogens, drugs or exposure to radiation, etc. in antenatal period. The antenatal period was uneventful, though no ultrasonography was done. There was no family history of any such or related congenital abnormality. Examination revealed heart rate-150/min, respiratory rate-76/min and SPO 2-88%. The lower half of sternum was bifid, with 5 cm inter-ridge distance. The heart, lying outside the thoracic cavity was covered with a thin membrane through which beating heart can be seen. It was also infected, since the membrane was coated

with green coloured purulent material, most probably due to pseudomonas/proteus organisms. Rest of the abdominal wall was intact and there was no omphalocoel, or any other anterior abdominal wall abnormality.

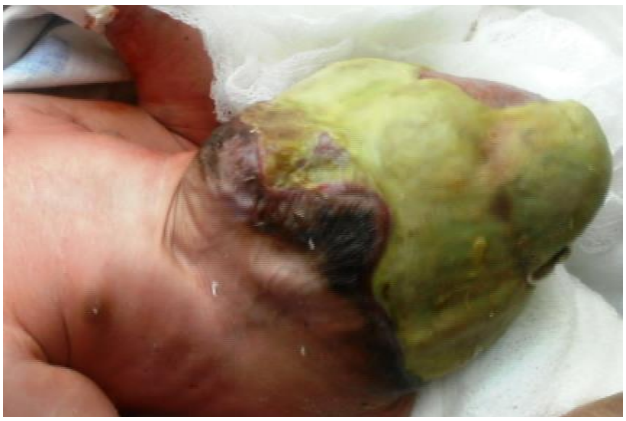

**Figure 1: The photograph showing heart outside thoracic cavity.**

*Investigations*: Haemoglobin (12 gm%), total leukocyte count (8200/cumm), differential leukocyte count (P-48%), L-42%, E-4%, M-2%, B-0%. Platelet count (2.3 lakhs/cumm), blood group (B+ve) were done. X-ray chest showed crowding of ribs with bifid lower sternum. Echocardiography showed ventricular septal and pericardial defects. The newborn baby remained alive for 72 hours and the relatives refused for post mortem examination.

## DISCUSSION

Ectopia cordis is produced by segmental defects in mesodermal development in the third week of intrauterine life, and amniotic band anomalies that produces simultaneous cerebral and Thoraco-abdominal malformations.[24-26] Ectopia cordis is rarely associated with chromosomal abnormalities. The existence of Ectopia cordis with severe congenital heart disease may be confirmed in the prenatal period by vaginal echocardiography at 10-12 weeks of gestation or by abdominal echocardiography by 20-22 weeks.[27,28] As of 2001, 267 patients have been reported, 102 (39.2%) of the thoracic type and 99 (38%) of the Thoraco-abdominal type. Ninety percent of the infants died in the first year of life. Most newborns had associated cardiac malformations.

Ectopia cordis can be complete, with the skin and parietal pericardium absent, or partial if there is pericardium under the sternum or skin over the sternum. The existence of an upper or lower partial sternal defect without a complete opening and covered by parietal pericardium and skin facilitates surgical treatment and alleviates the thoracic compression that results from introducing the heart in the cavity. In recent years, surgery has been attempted in one or two phases with variable results that depend mainly on the type of associated heart disease.[29,30]

The overall surgical objective of EC (all variety) management includes to closure of the chest wall defect (either by doing primary chest wall closures or by using bone/cartilage as tissue graft or artificial prosthesis like acrylic plaques, marlex mesh). The closure of the sternal defect and repair of the associated omphalocoel. Placement of the heart into the thorax and repair of the intracardiac defect.

The first attempted repair of EC was performed in 1925 by Cutler and Wilens.[31] Koop in 1975 achieved the first successful repair of thoracic EC in two stages.[32] Amato et al reported successful single stage repair of thoracic EC in 1995.[33] Conclave et al at Brazil reported a successful repair of uncomplicated EC in June 2007.[34]

Hence it may be concluded that EC is a lethal anomaly requiring prompt medical and surgical interventions. Surgery on these patients with life-threatening complex intracardiac anomalies, owes the only chance of survival, which should still be attempted despite poor outcomes.

### *Highlights of this article*

- It discusses a rare cardiac anomaly i.e. Ectopia cordis.
- Almost all EC cases are associated with other anomalies pertaining to heart and body wall.
- The cases may be diagnosed early in intrauterine life by intra- vaginal or abdominal ultrasound.
- The only chance of survival in the cases of EC is surgical intervention although the outcome is unsatisfactory, but anyway that is the only chance of survival.

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