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Thoracoabdominal Ectopia Cordis - A Case Report

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ABSTRACT

We report a case of thoracoabdominal ectopia cordis in a newborn female child with complete displacement of the heart outside the thoracic cavity, sternal defect in lower part, atrial septal defect, omphalocele, bilateral pleural effusion and bluish discoloration of cornea and moderate ascites.

Keywords : Thoracoabdominal ectopia cordis, sternal defect, atrial eptal defect, omphalocele

Introduction:
The term 'Ectopia cordis' was derived from Greek word 'ektopos' meaning away from a place (1). Ectopia cordis is defined as complete or partial displacement of the heart out side the thoracic cavity. It is a rare congenital defect in fusion of the anterior chest wall resulting in extra thoracic location of the heart. It occurs in 5.5 to 7.9 per 1 million live births (2).

Most reported cases of ectopia cordis are either thoracic (60%) or thoracoabdominal (40%); rarely, a case may be cervical or abdominal. The thoracic type is characterized by a sternal defect, absence of the parietal pericardium, cephalic orientation of the cardiac apex, epigastric omphalocele, and small thoracic cavity. The thoracoabdominal type has partial absence or cleft of the lower sternum, an anterior diaphragmatic defect through which a portion of the ventricle protrudes into the abdominal cavity, a defect of the parietal pericardium, and an omphalocele. Intracardiac abnormalities are common but not invariable; ASD, VSD, TOF, and tricuspid atresia are the most common intracardiac defects (3).

Case report:
A preterm (36 week) female child weighing 2.5 kg with Ectopia Cordis was delivered in our hospital to a 22 year primigravida mother by uncomplicated vaginal delivery. Antenatal period was adequately supervised. Ectopia cordis was diagnosed in fourth antenatal ultrasound scan done at 35 wk 6 days of gestation. The ultrasound scan was performed in our hospital and it revealed ectopia cordis with atrial septal defect, bilateral fetal pleural effusion and moderate amount of fetal ascites, defect in the upper ventral abdominal wall (width 3.2 cm) with lower sternal defect through which bulging of right hepatic lobe and peritoneal membranes could be seen beneath the herniated ectopia cordis. (Fig. 2)

On initial examination, the baby had heart rate of 140/min and respiratory rate of 50/min. The heart of baby was lying outside the thoracic cavity & devoid of pericardium with a sternal defect mainly in the lower segment. The great vessels were seen to enter the thoracic cavity from the externally placed heart. The apex of heart was pointing anteriorly. Baby also had associated supraumblical omphalocele which contained the right lobe of the liver with a thin membrane covering the omphalocele. (Fig. 1) There was bluish discoloration of cornea.
After resuscitation for poor respiratory efforts, baby was put on ventilator but baby expired after 6 hours.

Figure 1

Fig. 1 Baby with ectopia cordis

Figure 2

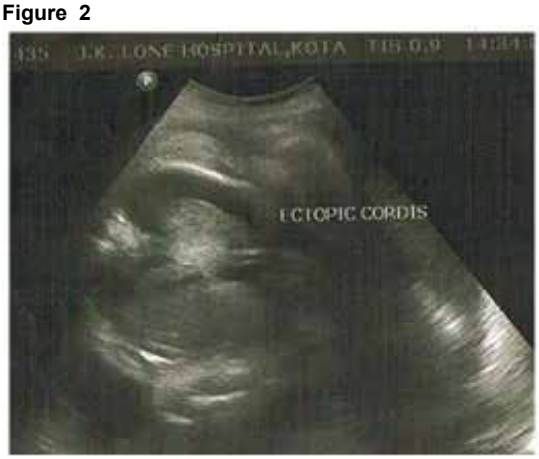

Fig. 2 Antenatal USG of patient

Discussion:

Thoracoabdominal ectopia cordis is also referred to as: Cantrell - Heller - Ravitch syndrome, pentalogy syndrome; and peritoneopericardial diaphragmatic hernia. Pentalogy of Cantrell (thoracoabdominal ectopia cordis) is a rare congenital syndrome of abdominal wall defect, lower sternal defect, diaphragmatic pericardial defect, anterior diaphragmatic defect, and intracardiac abnormalities. First described by Cantrell in 1958, the syndrome occurs sporadically with variable degrees of expression (4). Diagnosis of the complete syndrome requires the five criteria described by Cantrell, but incomplete variant forms exhibiting three or four of the features have been described. The sternal defect can range from absence of the xiphoid to cleaving, shortening, or absence of the entire sternum. The abdominal defect can range from a wide rectus muscle diastasis to a large omphalocoele (5). The exact aetiology is unknown and it occurs due to the developmental failure of a segment of the mesoderm, between 14 to 18 days after conception (6).

Kim et al classified ectopia cordis into 5 types based on the position of the misplaced heart: 1) cervical, in which the heart is located in the neck with sternum that is usually intact; 2) thoracicocervical, in which the heart is partially in the cervical region but the upper portion of the sternum is split; 3) thoracic, in which the sternum is completely split or absent, and the heart lies partially or completely outside the thorax; 4) thoraco - abdominal, which usually accompanies Cantrell's syndrome; 5) abdominal, in which the heart passes through a defect in the diaphragm to enter the abdominal cavity (7).

CNS, Face, Pulmonary, Skeletal, and Abdominal systems may also be involved.

The ultimate survival of these patients depend more on the presence or absence of intrinsic cardiac defects. Termination of pregnancy may be considered if ultrasound diagnosis is made before viability. Most of infants are stillborn or die within first few hrs or days of life (8, 9). Among survivors, early surgical corrective repair should be considered.

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