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Case Reports: Thoracic ectopia cordis with gastroschisis.

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Abstract

Ectopia cordis is a rare congenital defect in which the heart is displaced partially or completely, out of the chest. We present a case of term male neonate, delivered by elective caesarean section, asphyxiated and transferred to neonatal intensive unit at one hour of age. The baby had thoracic ectopia cordis and gastroschisis. The baby was unstable, cyanosed, required vasopressors and ventilatory supports, eventually succumbed after 12 hrs, before echocardiography and any surgical intervention could be performed.

Keywords: Ectopia cordis; gastroschisis,

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Introduction

Ectopia cordis is a rare congenital malformation, occurring in 5.5 to 7.9 per million live births (1). It occurs because of the failure of maturation of the midline mesodermal components of the chest and abdomen. It is commonly associated with anterior diaphgramatic hernia, omphalocele or gastroschisis, sternal and pericardial defects, and congential heart disease (2, 3). Depending upon the location of the heart, it could be classified into five types: cervical, cervico-thoracic, thoracic, thoraco-abdominal and abdominal (4,5). Thoracic and thoraco-abdominal forms account for majority of the case. The thoraco-abdominal type is frequently associated with Cantrell's pentalogy, which include bifid sternum, deficiency of the diaphragm, defect of diaphragmatic pericardium, defect of the anterior abdominal wall, and intracardiac defects (6). Incomplete expression of pentalogy can occur. Management is mainly surgical, may require a staged procedure to achieve a complete repair. Historically, the prognosis is poor, depends on associated defects. We present this case because of rarity of the condition.

Case report

This was a male baby, weighing 2.6 kg, intramural, born by elective caesarean section to a primigravida mother. The antenatal period was uneventful and there was no history of unusual infection or medication. Antenatal ultrasound was done at a gestational age of 28-30 wks showing upper abdominal wall defect with heart seen outside the thorax. There was no family history of any major congenital anomaly. The amniotic fluid was clear of meconium. At birth, the baby was limp with no spontaneous respiratory effort, and resuscitated. The newborn was intubated and given positive pressure ventilation for 10 minutes at birth. Apgar score was 7at 10 minutes. He was transferred to our neonatal intensive care unit at one hour of age. At admission, the baby was looking sick, lethargic, hypothermic, tachypnic, having poor perfusion and central cyanosis; and had shallow breathing. The heart rate was 168/ min. The peripheral pulses were feeble but regular with no radio-femoral delay. There was a mid sternal defect having beating heart protruding out from thoracic cavity, covered with a serous membrane, with the apex pointing upwards (Figure 1). Both atrial appendages were visible. The origin of the great arteries dipped into the thoracic cavity. The liver and spleen were not palpable. The lower part of sternum and upper abdominal wall was also deficient with a protruding mass covered with a membrane (gastroschisis) (Figure 1). The structure within mass could not be identified. The umbilical cord looks in an elevated position and was attached below the abdominal defect. The umbilical cord had two arteries and one vein. The first and sound heart sounds were normal with no murmurs. The breath sounds were normal and there were no added sound on auscultation. There were no any other external anomalies.

The initial management included stabilization, warmer care, O2 supplementation, intravenous fluid administration and orogastric decompression. The exposed heart and the abdominal mass were covered with sterile gauze moistened with normal saline at intervals. Blood gas analysis revealed combined acidosis and hypoxemia. The baby was put on vasopressor and mechanical ventilation. Consultation with pediatric and cardiothoracic surgeons was done, and the baby was initiated on broad spectrum antibiotics. The haemoglobin was 16.8 gm/dl; and initial blood sugar, urea and electrolytes were normal. However,

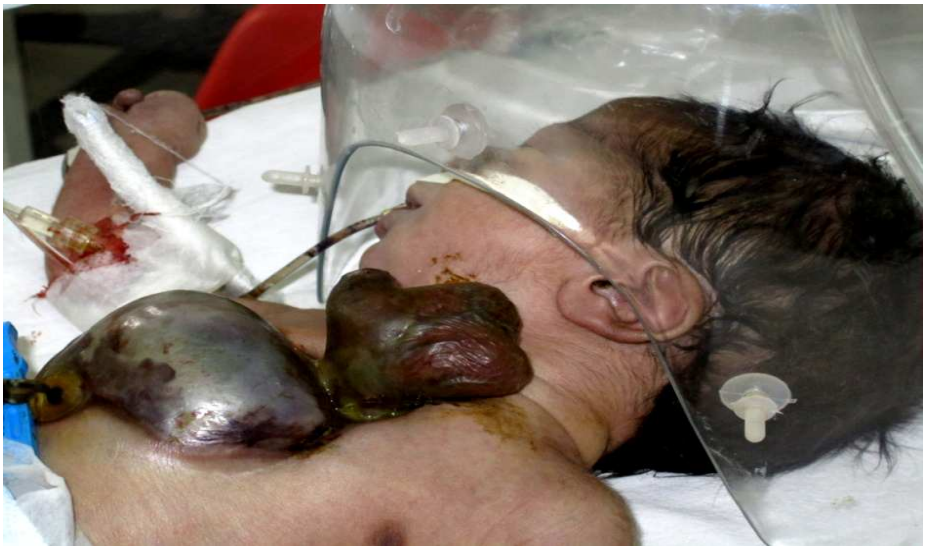

Figure 1. *Neonate with thoracic Ectopia Cordis showing naked heart outside the chest wall & associated gastroschisis*

the baby ran a downhill course and succumbed within 12 hours. Echocardiography could not be performed. Autopsy could not be done.

Discussion

The term ectopia cordis was first coined by Haller in 1706 (7). The defect is described as malposition of the heart, partially or completely outside the thorax. The etiology of ectopia cordis and gastroschisis is not known but may be linked to abnormalities in the lateral body wall folds responsible for closing the thoracic and abdominal portion of ventral body wall during 4-5 weeks of gestation (8). Omphalocele is separate entity, attributed to a failure of gut loops to return to the body cavity after their normal physiological herniation into the umbilical cord from 6-10 week of development (8). Davies et al (9), has proposed two distinct mechanisms leading to ectopia cordis: (i) a reverse diaphragmatic hernia in the case of a large diaphragmatic defect and an omphalocele; (ii) through a sternocostal defect with gastroschisis or a supraumbilical abdominal defect.

Cervical form of ectopia cordis is rare, while thoracic form is the commonest. Our case was thoracic type with complete sternal cleft which allowed protrusion of the heart outside the thoracic cavity. In our patient, the abdominal defect was associated with gastroschisis, and not seems to be omphalocele, as umbilical cord was not attached onto the ventral defect.

Majority of ectopia cordis patients have associated congenital heart defects, commonly ventricular septal defect, atrial septal defect, tetralogy of fallot, diverticulum of ventricle, and other anomalies (10,11). Our patient appeared to have cardiac defect as there was circulatory compromise and desaturation; however, the patient was asphyxiated, and it could not been confirmed by echocardiography. Other associated anomalies with ectopia cordis include cleft lip and palate, meningocele, encephalocele and skeletal deformities.

The management of ectopia cordis apart from supportive care involves: closure of the chest wall defect, including sternal defect, repair of the associated abdominal defects, placement of the heart into the thorax and repair of the intracardiac defect. This may be achieved in single or multiple stages (10, 12). Prognosis of ectopia cordis is largely related to severity and the complexity of the intracardiac defect (10). Many of infants are stillborn or die within few hours of life (11).

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