Thoracoabdominal Ectopia Cordis: A Report of Two Cases

RAJENDRA K. GHRITLAHAREY, K.S. BUDHWANI, JYOTI SRIVASTAVA

ABSTRACT

Ectopia cordis (EC) is defined as the complete or partial displacement of the heart outside the thoracic cavity. It is a rare congenital defect in the fusion of the anterior chest wall, resulting in the extra thoracic location of the heart. We are describing here, two cases of thoracoabdominal EC. One died after emergency surgery and another is awaiting definitive procedures.

Key Words: Ectopia cordis, Ectocardia, Thoracoabdominal ectopia cordis, Cantrell's pentalogy

INTRODUCTION

EC, ectocardia or exocardia is a rare congenital defect in the fusion of the anterior chest wall, resulting in the extra thoracic location of the heart [1]. The estimated prevalence of EC is 5.5-7.9 per million live births [2], [3]. Depending upon the location of the heart, it could be classified into five types: cervical, cervicothoracic, thoracic, thoracoabdominal and abdominal [2], [3]. Thoracic and thoracoabdominal EC account for about 85% of the cases [2], [3]. Cantrell, et al, in 1958, first described this syndrome in which anterior diaphragmatic hernia occurred in association with an omphalocele. Thoracoabdominal ectopia cordis is also referred to as: Cantrell-Heller-Ravitch syndrome, pentalogy syndrome; and peritoneo-pericardial diaphragmatic hernia [4].

CASE 1

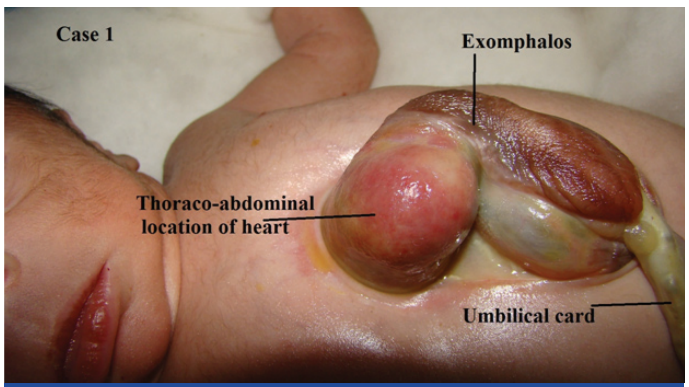
A full term, 2-day-old boy, weighing 3 kg, was admitted with an ectopic heart. Clinical examination revealed the thoracoabdominal location of the heart, with associated exomphalos major [Table/Fig-1]. He had cyanosis and his general condition was poor. Under anaesthesia, a skin cover was given to the heart and the exomphalos was repaired. He succumbed few hours after the surgery. No other investigations were possible and the intracardiac lesions could not be known, but we presume that he had severe intra cardiac lesions.

CASE 2

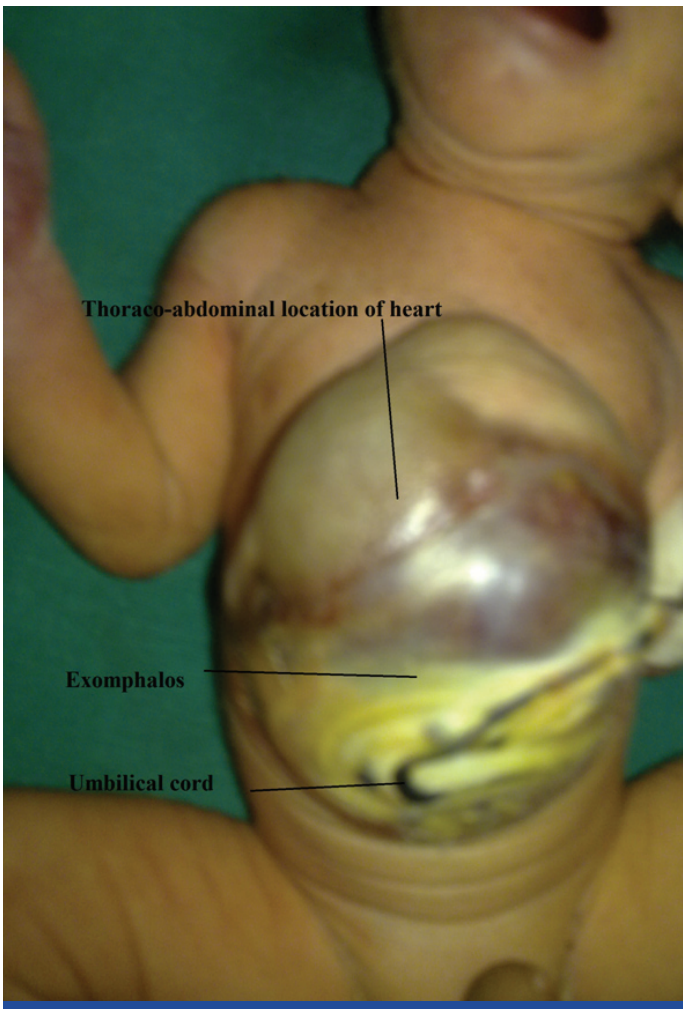
A boy was born at 37 weeks of gestation to a G I, P 0, 23 years-old mother. The antenatal period and the hospital delivery were uneventful. The clinical examination revealed that he weighed 1.7 kg and that he did not have cyanosis or respiratory distress. He had thoracoabdominal EC with exomphalos major [Table/Fig-2 and video of case 2]. The X-ray of his chest / abdomen revealed a sternal defect and a soft tissue shadow in the thoracoabdominal region. Ultrasonography (USG) of the abdomen was suggestive of the thoracoabdominal location of the heart, the exomphalos contained intestines and a part of the heart and the liver. Similar findings were confirmed by computed tomography (CT) scans of the abdomen and chest [Table/Fig-3 & 4]. Echocardiography reported ventricular septal defect (VSD) and patent ductus arteriosus (PDA) [Table/Fig-5]. As he had intra-cardiac lesions, the exomphalos was treated conservatively and other definitive procedures were deferred. He was discharged on request on the 13th post admission day in a good condition with an advice about the need of multiple surgeries and he was gaining weight as well.

DISCUSSION

EC is defined as the complete or partial displacement of the heart outside the thoracic cavity. It is a rare congenital defect in the fusion of the anterior chest wall, resulting in the extra thoracic location of the heart. Depending upon the location of the heart, it can be classified into five types: cervical, cervicothoracic, thoracic, thoracoabdominal and abdominal. Thoracic and thoracoabdominal EC account for about 85% of the cases [2], [3]. 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 [4]. Thoracoabdominal ectopia cordis is also referred to as: Cantrell-Heller-Ravitch syndrome, pentalogy syndrome; and peritoneopericardial diaphragmatic hernia [4]. The complete syndrome is characterized by two major defects: ectopia cordis and an abdominal wall defect [most commonly an omphalocele, but gastroschisis can also be present). The other three defects of the pentalogy are the disruptions of all the interposing structures: the distal sternum, the anterior diaphragm and the diaphragmatic pericardium. Incomplete expressions of pentalogy have also been reported [4], [5].



[Table/Fig-1]: Clinical photograph of case 1, showing thoracoabdominal heart and exomphalos

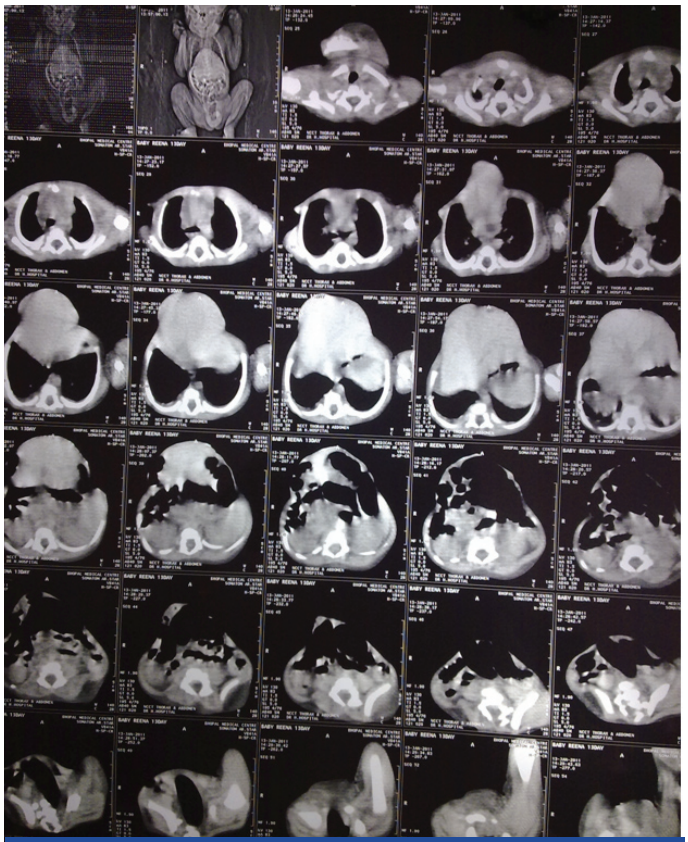


[Table/Fig-2]: Clinical photograph of case 2, showing thoracoabdominal heart and exomphalos

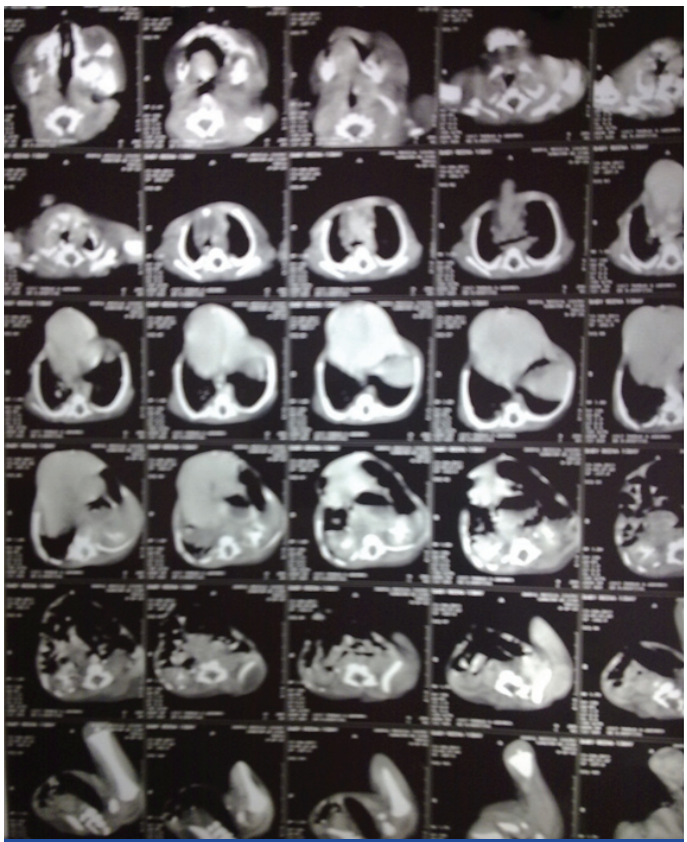
Thoracoabdominal EC may be associated with other intracardiac anomalies, cranial and facial anomalies, chromosomal abnormalities, clubfeet, malrotation of the colon, hydrocephalus, and anencephaly [2], [4]. Intracardiac anomalies are described in the pentalogy of Cantrell, including VSD (100%), ASD (53%) and tetralogy of the Fallot (20%), and the ventricular diverticulum (20%). Most of the cases are sporadic and no familial tendency or recurrence has been demonstrated, but thoracoabdominal EC has also been reported in twins [4].

The prenatal diagnosis of thoracoabdominal EC is possible in the first trimester of pregnancy by USG and this depends on the size and extent of the defects. The diagnosis of the complete syndrome requires the five criteria which are described by Cantrell, but incomplete variant forms exhibiting three or four of the features may also present. Ultrasound, echocardiography, CT scan / multi detector computed tomography (MDCT) scan and magnetic resonance imaging, when necessary, should be applied to search for this combination of malformations [4], [6], [7], [8]. The surgical correction of these defects is complex and it generally requires a staged closure, but a single stage repair has also been reported, with success [2], [9], [10]. The staged repair includes: [1] covering the heart, [2] correction of the intrinsic cardiac defects prior to the orthotopic location of the heart and [3] placement of the heart into the thoracic cavity and sternal / thoracic reconstruction. The potential advantages of the staged repair are; minimization of the compression of the heart and the great vessels, and allowing the thoracic cavity to expand gradually.

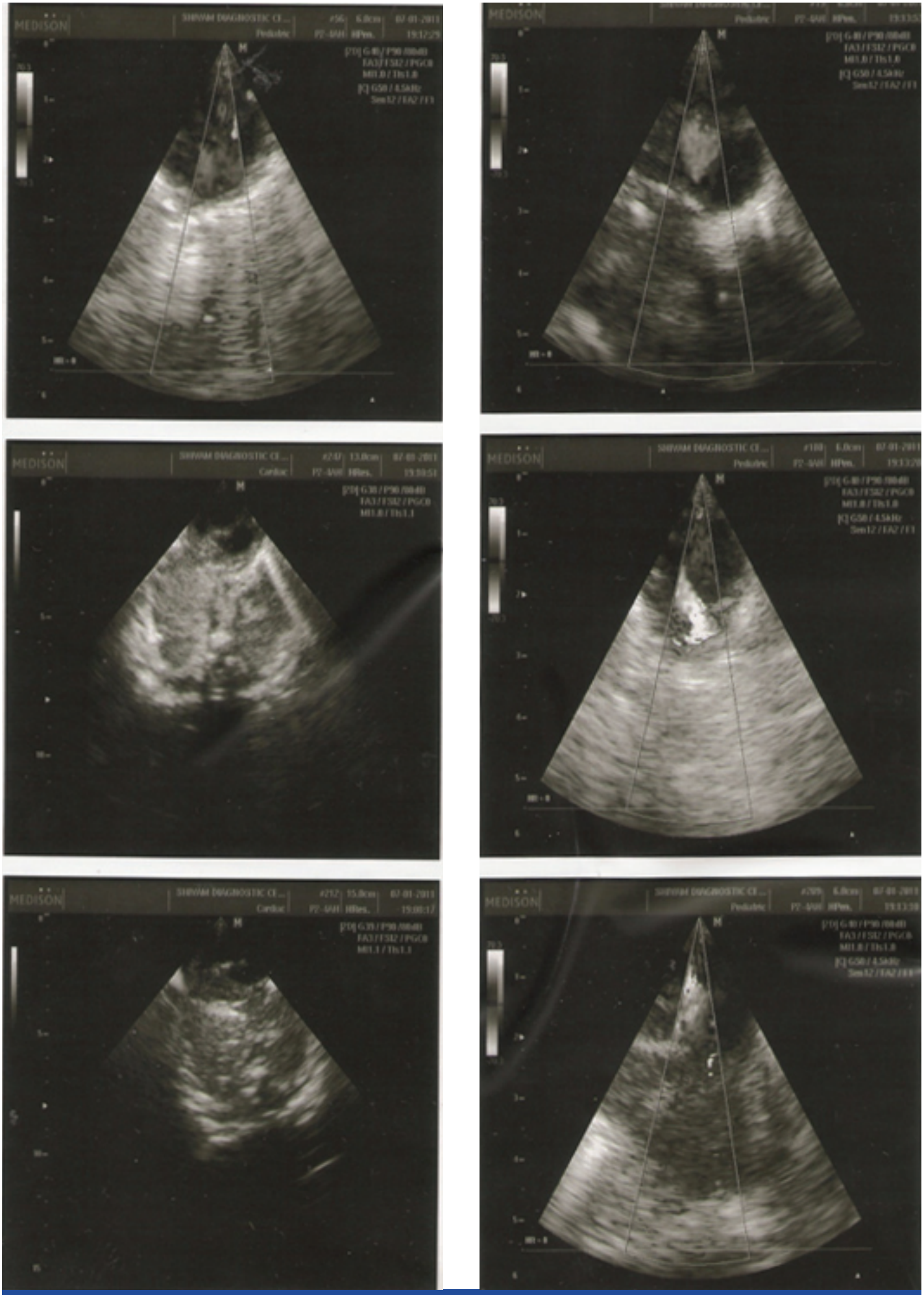
Our case one had thoracoabdominal EC, exomphalos and sternal defect, but no diaphragmatic hernia and intracardiac lesions could not be detected. A skin cover for the heart was provided and the exomphalos was repaired. Case 2 also had incomplete pentalogy of Cantrell, ie, thoracoabdominal EC, exomphalos major, sternal defect, VSD and PDA, but no diaphragmatic hernia. In view of the



[Table/Fig-3]: CT scan of abdomen and chest of case 2



[Table/Fig-4]: CT scan of abdomen and chest of case 2



[Table/Fig-5]: Echocardiography of case 2, showing VSD

intracardiac lesions and the presence of the skin cover over the heart, the repair of the exomphalos and the definitive procedures were deferred.

The successful repair of such anomalies is dictated by the presence and the severity of the intrinsic cardiac defects and the other associated congenital anomalies, rather than the type of surgical approach itself. Advances in the foetal ultrasound techniques have aided in the early recognition of such anomalies and the termination of the pregnancy prior to viability should be considered and discussed with the parents.

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