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Case Report

Radiology Section

# Ectopia Cordis with Omphalocele in First Trimester Pregnancy: A Case Report of Incomplete Type of Pentalogy of Cantrell

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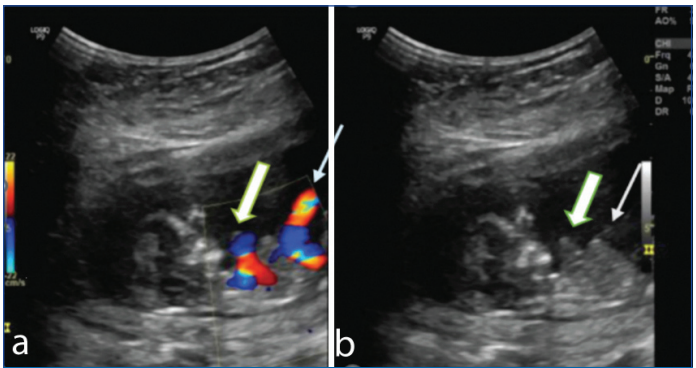
## ABSTRACT

Ectopia Cordis (EC) is a rare congenital malformation in which the heart is located partially or totally outside the thoracic cavity. It results from the failure of migration of the lateral mesoderm into the midline. The four main ectopic positions are cervical, thoracic, thoracoabdominal and abdominal. EC can manifest as an isolated deformity or as part of a broader group of ventral body wall defects affecting the abdomen, thorax, or both. Pentalogy of Cantrell is a well known association that comprises EC, omphalocele (typically supraumbilical), congenital diaphragmatic hernia, sternal cleft and congenital heart disease. A 27-year-old primigravida came for a routine antenatal ultrasound at 13 weeks of gestation. There was no family history of congenital anomalies, genetic abnormalities, or exposure to teratogenic agents. The ultrasound showed a single foetus corresponding to 13 weeks of gestation with an anterior thoracic defect and an extrathoracic heart, along with partial herniation of the liver near the midline in the epigastric region. These findings were confirmed by foetal Magnetic Resonance Imaging (MRI). An unfavourable prognosis for the foetus was explained to the parents, and medical termination of pregnancy was performed. Therefore, prenatal ultrasonographic diagnosis of EC should be followed by a thorough search for associated defects, as the prognosis may vary.

**Keywords:** Antenatal ultrasonography, Anterior abdominal wall defects, Congenital anomalies, Foetus

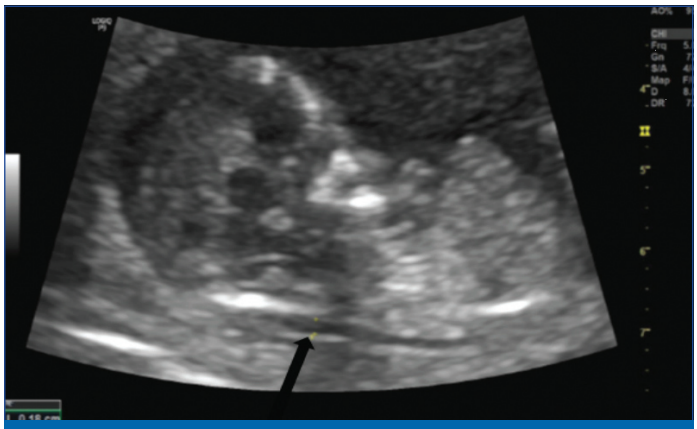
## CASE REPORT

A 27-year-old primigravida came for a routine antenatal ultrasound at 13 weeks of gestation. The patient conceived naturally and had regular antenatal check-ups. She had no relevant past or present medical or surgical history, and there was no family history of congenital anomalies, genetic abnormalities, or exposure to teratogenic agents. The patient was sent to the Radiology Department for a Nuchal Translucency (NT) scan. A 2 Dimensional (2D) and 3D ultrasound revealed a thoracic defect, with the foetal heart lying completely outside the thoracic cavity, protruding into the amniotic cavity. Additionally, mild protrusion of the foetal liver into the amniotic cavity was observed near the midline in the supraumbilical region, with an umbilical cord inserted at the lower end of the omphalocele and covered by a thin membrane [Table/Fig-1]. The NT value was within normal limits (<95$^{th}$ percentile), but the nasal bone was not visualised [Table/Fig-2].

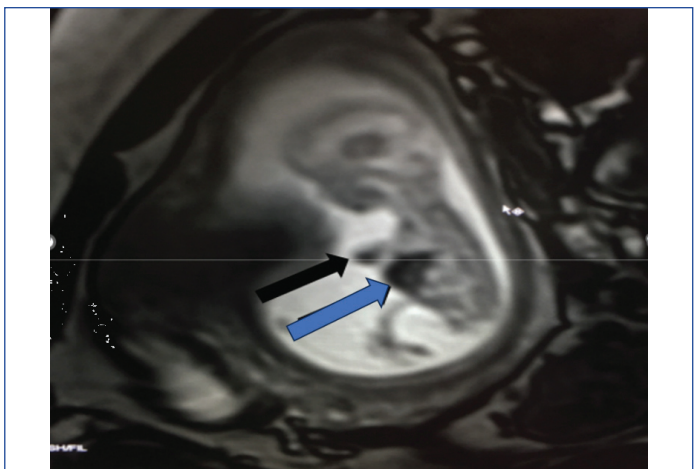



**[Table/Fig-1a,b]:** Colour doppler and B-mode Ultrasonogram (USG) shows thoracic defect with cardiac chamber lying outside thoracic cavity (block arrow) and mild protrusion of liver into amniotic cavity with umbilical cord inserted at the lower end of omphalocele (open arrow).

For further delineation of the anomalies, a foetal MRI was suggested. The MRI confirmed the thoracoabdominal type of EC and supraumbilical omphalocele with the liver as content [Table/Fig-3]. These findings raised the suspicion of the pentology of Cantrell; however, it was difficult to demonstrate intracardiac anomalies by ultrasonography or MRI at 13 weeks of gestation.

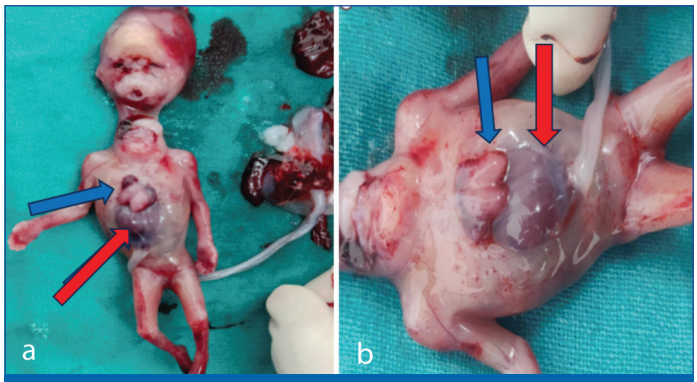


**[Table/Fig-2]:** NT: 18 mm. The nasal bone was not visualised. (Arrow shows nuchal thickness).



**[Table/Fig-3]:** T2 Weighted (T2W) MRI sagittal section shows sternal defect with T2 hypointense heart lies completely outside thorax (black arrow), T2 hypointense liver (blue arrow) is also seen protruding into amniotic cavity.

An unfavourable prognosis for the foetus was explained to the parents, and medical termination of pregnancy was performed. The foetal abortus revealed a heart outside the thoracic cavity with a sternal defect and a liver lying outside the abdominal cavity with an anterior abdominal wall defect [Table/Fig-4a,b]. Foetal karyotyping was later performed and found to be normal. The parents were informed about the potential risks in subsequent pregnancies and advised to seek genetic counselling.




**[Table/Fig-4]:** Foetal abortus revealed heart outside the thoracic cavity (blue arrow) with sternal defect and liver (red arrow) lying outside the abdominal cavity covered with thin membrane.

## DISCUSSION

Embryologically, the sternum develops from two parallel bands formed by the longitudinal fusion of mesenchymal layers. By the sixth week of gestation, the sternum consists of two parallel primordia, with sternal bars positioned away from the midline. The sternal bars fuse to create a median cartilaginous plate, progressing from the cephalad to the caudal end, as they gradually undergo chondrification and move toward the midventral line. Failure of sternal fusion can lead to various cleft sternal disorders, which manifest as complete or partial EC [1].

With an estimated frequency of 0.079 per 10,000 births, EC is an extremely rare defect that may be more common in females. It is associated with defects of the anterior wall of the thorax, resulting in the extrathoracic location of the heart [2]. EC can be divided into five types: 1) Cervical; 2) Thoracocervical; 3) Thoracic; 4) Thoracoabdominal; and 5) Abdominal [3,4].

In 1958, Cantrell JR et al., reported five cases of EC associated with congenital defects, including diaphragmatic hernia, omphalocele involving the abdominal wall, partial absence of the pericardium, Ventricular Septal Defect (VSD) and a partial opening in the sternum due to defective closure of the midline [5].

Kylat RI, reported a case involving a primigravida at 39 weeks of gestation, whose foetus was diagnosed with Cantrell's pentalogy. A prenatal ultrasound showed a large omphalocele with liver herniation, anterior diaphragmatic hernia, absence of the lower sternum (EC) and a VSD. After birth, the large omphalocele was enclosed, along with closure of the sternal defect, mediastinal defects and anterior abdominal wall defects. The infant required increased respiratory support and was haemodynamically unstable, necessitating resuscitation twice due to severe decompensation. At 19 days, the parents elected to withdraw critical care support, leading to the infant's demise [6].

Türkçapar AF et al., published a case study discussing the complementary roles of transvaginal sonography and colour Doppler in the diagnosis of Cantrell syndrome in early pregnancy. The transvaginal scan revealed significant lumbosacral scoliosis and a midline supraumbilical abdominal wall deformity, which included a herniated liver and EC, as well as, a massive omphalocele containing the intestines. The abdominal aorta could not be visualised, which could indicate an intracardiac defect [7].

Shenoy HT et al., reported a case of incomplete (Class 3) pentalogy of Cantrell, where a multigravida at 20 weeks of gestation was diagnosed with a large abdominal wall and partial chest wall defect. There was a spinal deformity (kyphoscoliosis), along with pelvic deformity and rotation of the lower limbs. No cardiac malformation was noted [8]. EC has been linked to several congenital defects, including cranial and facial deformities, cleft lip and palate, anencephaly, neural tube defects, pulmonary hypoplasia, genitourinary malformations, gastrointestinal defects and chromosomal abnormalities [9].

In a few cases, the spectrum of the pentalogy of Cantrell was not fully present. Hence, in 1972, Toyama WM proposed proposed the following classification for the pentalogy of Cantrell: Class 1: conclusive diagnosis, with all five anomalies present; Class 2: probable diagnosis, with four defects present, including intracardiac and ventral wall anomalies; Class 3: incomplete diagnosis, with combinations of defects, including a sternal anomaly [10]. In the present case, thoracoabdominal EC with a sternal defect, extrathoracic heart, and omphalocele was observed, an incomplete version of Cantrell's pentalogy.

Differential diagnosis include isolated thoracic EC, amniotic band syndrome and body stalk anomalies. The position of the abdominal wall defect relative to the umbilical cord, the eviscerated organs, the presence of membranes or bands, and any concomitant anomalies will help distinguish these conditions. The omphalocele in Cantrell's pentalogy is typically a midline anomaly at the umbilical cord insertion. Body stalk anomalies are recognised by a large lateral defect with placental attachment to it. Amniotic band syndrome is characterised by an unexplained ventral wall defect and extremity deformities accompanied by an adhering band [11].

## CONCLUSION(S)

The prenatal diagnosis can be easily established using 2D/3D ultrasound to visualise the heart outside the thoracic cavity. However, small abnormalities in the diaphragm and the pericardium can be extremely difficult to diagnose accurately. In these cases, MRI can be useful. The prognosis depends on the degree of intracardiac involvement and any accompanying abnormalities, as well as, the extent to which the heart is exposed. Therefore, a thorough assessment is essential to provide appropriate prenatal and postoperative management.

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