

## Cantrell Syndrome: A Rare Case Report

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### Abstract

Cantrell syndrome is a rare syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium and the heart. The spectrum of anomalies varies widely. Less than 160 cases have been described in the world literature. We reported a premature infant, with the syndrome. The case with a rare congenital malformation consisting of a pentad of findings: ectopia cordis and absent pericardium and a midline supraumbilical wall defect, evisceration of the intestines and liver, and short sternum. We presented this case because of its rarity and discuss the pathologic findings.

**Keywords:** Cantrell syndrome; Ectopia cordis; Midline defects

### Introduction

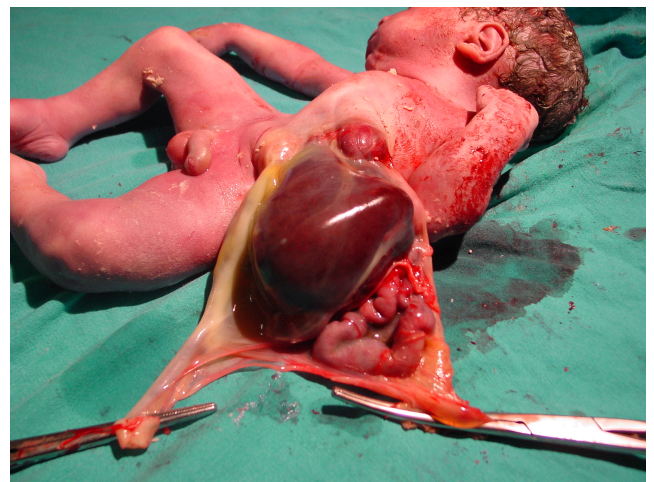
In 1958, Cantrell et al. [1] described a rare syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and congenital heart malformation. The characteristics of this syndrome are as follows: a midline supraumbilical abdominal wall defect; a defect of the lower part of the sternum; a deficiency of the anterior diaphragm; a defect in the diaphragmatic pericardium; and a congenital heart malformation. There are different statements in the literature about the incidence of this syndrome. The incidence of the malformation is one in 65,000 live births [2]. Carmi and Boughman reported that regional prevalence is 5.5/1 million liveborn infants for this disorder [3]. It is common in males. The exact etiology remains unknown to date. No familial tendency has been demonstrated. The basic defect behind these anomalies could be developmental failure of mesoderm at early embryonic life [2].

A newborn with the supraumbilical wall defect as diagnosed Cantrell syndrome is reported.

### Case Report

The patient was born as preterm from the mother's first pregnancy after a normal gestational period. There was no family history of congenital defects. In the story, there is not parent's consanguineous and sister. This pregnancy is the mother's first gestation and no stillbirths or pregnancy loss and no similar disease in the family history. At birth, the Apgar scores were 4 and 5 at 1 and 5 minutes, respectively. It was measured birth weights: 2800 g, height: 45 cm, head circumference: 34 cm. After birth, physical examination of the newborn showed a supraumbilical wall defect with ectopia cordis, absent pericardium, evisceration of the intestines and liver, short sternum (Figure 1). An omphalocele was immediately apparent. The sternum was short but complete, and beneath the xyphoid process. A pulsatile structure extending into the epigastrium was seen. Baby's palate was

intact. There was no systolic and diastolic murmur. Because the patient was breathing and had an adequate heart rate, he was transferred to the intermediate care unit for observation, and a mesh was placed over the exposed thoracoabdominal viscera as protection. While in the care unit, the patient had acrocyanosis and died within one hour after birth. The physical examination was otherwise normal (Figure 2). Chromosomal analysis revealed a 46 XY genotype.



**Figure 1:** Clinical photograph shows in the Cantrell's syndrome. Heart and liver was outside the thoracic cavity (General view).

### Discussion

Ectopia cordis is defined as the presence of the heart outside the normal confines of the thorax, the walls of which are composed of ribs, sternum, and diaphragm. According to Cantrell's pentad, it is accompanied by varying numbers of the following associated defects:

(I) a central abdominal wall defect, supraumbilical or omphalocele; (II) an abnormal lower sternum; (III) a “V” shaped defect in the anterior diaphragm; (IV) a defect in the diaphragmatic aspect of the pericardium; and (V) congenital heart disease (VI). Its pathogenesis has not been elucidated. Cantrell et al. [1] suggested a developmental failure of a segment of the lateral mesoderm around embryonic days 14 to 18. As a consequence, the transverse septum of the diaphragm does not develop, and the ventromedial migration of the paired mesodermal folds of the upper abdomen fails to occur. In the present case, it is difficult to propose the etiology. It is likely that hereditary and environmental factors may both be responsible.



**Figure 2:** Photograph of the newborn with features of Cantrell's syndrome (Front view).

The prognosis depends on significant extracardiac defects, complex cardiac anomalies, pulmonary hypoplasia, large abdominal wall defects, cerebral anomalies, and herniation of the bowel into the thoracic cavity because all these would likely worsen the overall prognosis of these patients [4]. The survival rate is 36 h on average, and all unoperated cases have died [4].

Onderoglu et al. [5] performed the diagnosis at the 16th gestational week, the pregnancy was terminated because of karyotype revealing trisomy 21 and the serious structural defects. Autopsy demonstrated an ectopia cordis without pericardium and an abdominal wall defect with an omphalocele. Fetus had no diaphragm or sternum, and pulmonary and extremity anomalies were also present. Because of the poor prognosis of Cantrell's pentalogy, early antenatal sonographic detection of it is important and allows for elective abortion before viability. Even though the pentalogy of Cantrell occurs sporadically as an isolated event, the family should receive genetic counseling. Unfortunately our patient has been made antenatal sonographic examination.

A review of world literature revealed 153 cases of Cantrell's syndrome, with sternal malformations in 91 patients. The abdominal wall was defective in 114 patients, the diaphragm in 87 and the pericardium in 64. Cardiac malformations were described in 127 patients, most commonly VSD (92 patients, 72%). Tetralogy of Fallot was present in 22 patients (17.3%) and left ventricular diverticulum in 41 (32.3%) [6-8]. Most of these malformations were present supraumbilical wall defect with ectopia cordis, absent pericardium,

evisceration of the intestines and liver, short sternum and omphalocele in our case.

Presence of a distal sternal defect, an omphalocele or omphalocele like partial abdominal defect, a crescentic anterior diaphragmatic defect, a pericardial defect allowing pericardio-peritoneal communication and an intracardiac defect usually involving ventricular septum constitutes classical Cantrell's pentalogy [1]. Intracardiac defects of the case were not determined due to no permission from the parent's, although Cantrell et al. noted that congenital intracardiac anomalies are constant elements of the pentalogy, with ventricular septal defects seen in every case (100%), atrial septal defects in 53%, pulmonary stenosis in 33%, tetralogy of Fallot in 20%, and left ventricular diverticulum in 20% [1].

Çelik et al. [9] reported that ectopia cordis is associated with other congenital heart diseases and various tissue and organ disorders. Common cardiac anomalies associated with ectopia cordis include ventricular septal defect, atrial septal defect, pulmonary stenosis, right ventricular diverticulum, double right ventricular outflow tract and tetralogy of Fallot. Extracardiac anomalies associated with ectopia cordis reported in the literature include omphalocele, gastrochisis, cleft lip and palate, scoliosis and central nervous system malformations. Çelik et al. [9] case's reported that heart was completely outside of the thoracic cavity, there was no pericardium on the heart. There was defect in the abdominal wall above the umbilicus, abdominal distention was present. In our case's was a supraumbilical wall defect with ectopia cordis, absent pericardium, evisceration of the intestines and liver, and short sternum and omphalocele (Figure 1).

In the existence of supraumbilical or thoracoabdominal defects, cases should be detailed evaluated in point of Cantrell syndrome. Furthermore; cases with omphalocele and/or together sternal malformations must be precise evaluated on account of other components of the Cantrell syndrome.

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