# Cantrell syndrome in one of a set of monozygotic twins

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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. A full syndrome, probable syndrome and incomplete expression have been described. Less than 160 cases have been described in the world literature. Only one set of monozygotic twins concordant with the syndrome has been reported. To our knowledge, the syndrome affecting only one of a set of twins has not been reported. We report a premature infant, the first-born twin, with the syndrome. The omphalocoele was large, and heart abnormalities included dextrocardia with atrial septal defect. The co-twin is normal.**


**Keywords: atrial septal defect, Cantrell syndrome, dextrocardia, monozygotic twins, omphalocoele**

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## INTRODUCTION

Cantrell syndrome consists of congenital defects of abdominal wall, heart, sternum, diaphragm and pericardium and is due to developmental failure of mesoderm at an early embryonic period. The syndrome occurs in various degrees of severity, ranging from incomplete to severe expression with involvement of other organ systems[1,2]. This syndrome has been reported in only one set of twins by antenatal ultrasonography[3]. We present a case of Cantrell syndrome with a giant hepatomphalocoele affecting only the first-born of a set of premature monozygotic twins. An anomalous pinna was an additional interesting finding in our case.

## CASE REPORT

A 35-week-old premature baby, the first of a pair of twins born to a second gravida mother by lower segment caesarean section was noticed to have a huge intact omphalocoele. It measured 10 cm × 10 cm over the anterior abdominal wall (Fig.1). Apgar score was eight at one minute. Birth weight was 2,310 g. The placenta was monochorionic and diamniotic. The co-twin is normal, with the same sex and blood group. Antenatal ultrasonography showed polyhydramnios and suggested the presence of an omphalocoele. The base of the abdominal wall defect was 10 cm. Both lobes of the liver, small and large intestines constituted the contents of the sac. The cardiac apex was felt on the fourth intercostal space on the right side. There were no murmurs. An anomaly of pinna was present but the physical examination was otherwise normal.

Radiograph showed dextrocardia and a huge omphalocoele (Fig. 2). The sternum was short, with defective formation of its lower third. Echocardiography revealed dextrocardia and an ostium secondum atrial septal defect of 0.4 cm. The diagnosis of Cantrell syndrome was made. The baby was managed conservatively as the omphalocoele was large. During subsequent follow-up, the baby was thriving well. The skin covering turned the defect into a ventral hernia. The baby underwent surgery at 11 months of age. The sac contained the liver, small and large intestines. A small defect of the diaphragm was also noted at surgery.

## DISCUSSION

In 1958, Cantrell et al[4] described this rare syndrome. The incidence of the malformation is one

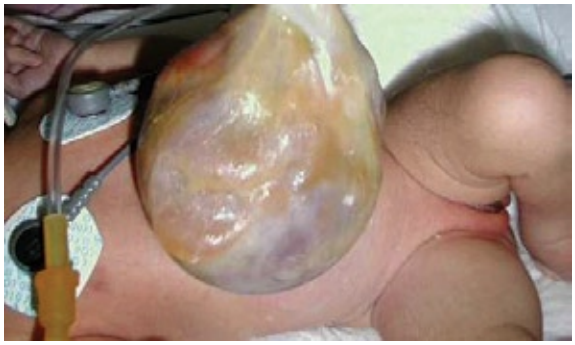

**Fig. 1** Clinical photograph shows giant hepatomphalocele in Cantrell syndrome.

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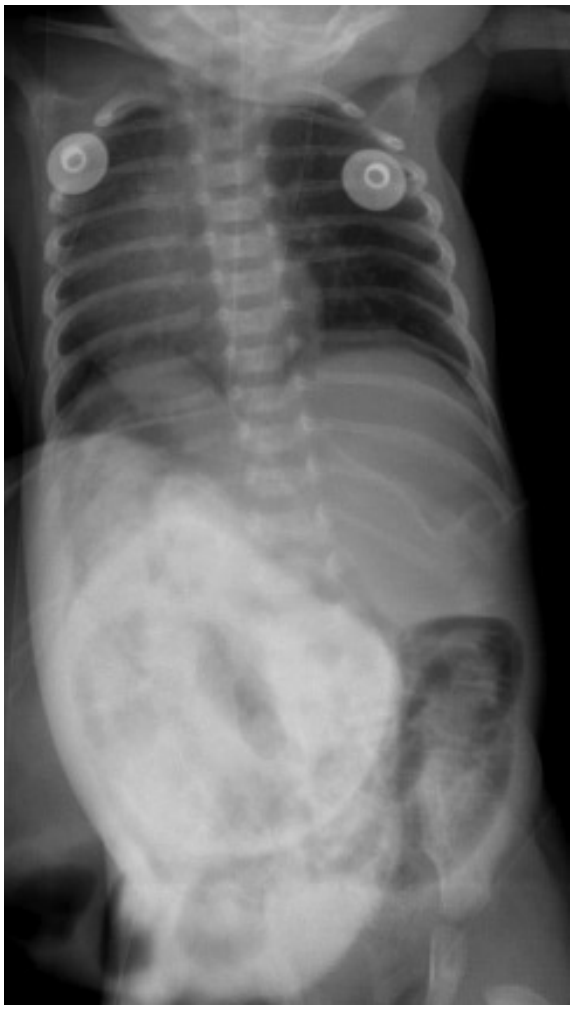

**Fig. 2** Radiograph shows giant omphalocele with dextrocardia.

in 65,000 live births. It is common in males. The characteristic pentology of the syndrome are midline supra-umbilical abdominal wall defect, congenital heart malformation, defect of lower sternum, deficiency of anterior diaphragm, and a defect in the diaphragmatic pericardium. The exact aetiology 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 between 14 and 19 days[5]. Differences in timing may explain the variability. The extent of individual defects and their combinations varies considerably. The involvement of the mesoderm prior to its differentiation into splanchnic and somatic layers results in a complete pentology, while an insult after the differentiation does not involve the heart.

A detailed review of the literature makes it apparent that the syndrome occurs in various degrees of severity from incomplete to severe expression with involvement of other organ systems[2,6]. The syndrome is rare among twins. Similar congenital defect gastroschisis in one pair of dizygotic twins has been reported in 1984[7]. The authors discussed the possible role of maternal alcohol consumption as the aetiology. For the Cantrell syndrome in the present case, it is difficult to propose the aetiology. It is likely that hereditary and environmental factors may both be responsible. We could not find any aetiological factor in our case.

Abdominal wall defects include omphalocoele, distasis recti, epigastric hernia, umbilical hernia, and combined defects. However, the most common abdominal wall defect is omphalocoele. A giant omphalocoele[6] refers to an abdominal wall defect greater than 5 cm in diameter. The term hepatomphalocele implies a liver-containing omphalocoele. In our case, the abdominal wall defect was 10 cm in diameter. The omphalocoele contained both lobes of the liver and intestines. Cardiac lesions may vary widely. Common lesions include ventricular septal defect, tetralogy of Fallot, ostium secondum atrial septal defect, dextrocardia, left ventricular diverticulum, pulmonary stenosis or atresia, and complex malformations. Sternal malformations commonly seen are defective formation of the lower third, bifid sternum, absent xiphoid process and short sternum. Ventral defects of the diaphragm and absent pericardium are the common diaphragmatic and pericardial defects, respectively.

The Cantrell syndrome must be actively sought in every patient with an omphalocoele. Chest radiographs may demonstrate cardiac dextro-rotation and associated thoracic anomalies. Echocardiography and cardiac catheterisation will lead to the diagnosis of the intra-cardiac anomalies. Treatment of this syndrome depends on the size of the abdominal wall defect and the other associated malformations[5,6,8]. When the omphalocoele is large and contains the liver, immediate reduction becomes difficult. There are various approaches and staged repair. In the present case, delayed surgery was planned and performed.

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