

## **Pentalogy of Cantrell: A case report**

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**Abstract:** Cantrell's pentalogy is a rare congenital malformation that consists of supra umbilical abdominal wall defect, the sterna lower part defect and agenesis of the anterior portion of the diaphragm, an absence of the diaphragmatic part of the pericardium and a malformation of cardia. This case report presents a male neonate who was born at 37 weeks of conception, weighing 2500 g in Mahatma Gandhi Hospital. He had the five anatomical defects known as Cantrell's pentalogy.

**Keywords:** Ectopia cordis, Sternal cleft, Supraumbilical abdominal wall defect.

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### **INTRODUCTION**

Cantrell *et al.*; in 1958 reported 5 cases with a pentad of findings that included supraumbilical wall defect, defect of the lower sternum, deficiency of the anterior diaphragm, defect of diaphragmatic pericardium and cardiac anomalies [1]. Pentalogy of Cantrell is a rare congenital anomaly having an incidence of 1: 100,000 with a male preponderance (M: F = 2: 1.2)

Toyama WM (1972) described three categories of Pentalogy of Cantrell-

Category 1: exact diagnosis with 5 defects present

Category 2: probable diagnosis with 4 defects present (including intra cardiac and abdominal wall defects)

Category 3: incomplete diagnosis with combination of defects where sternal defect is always present

There are some case reports that have been classified as full spectrum and incomplete forms with lesser defects of Pentalogy of Cantrell have also been

reported[2-4]. There are few reported survivors after corrective surgery [2].

### **CASE REPORT**

An unbooked antenatal patient G3P2 term pregnancy with labour pains was admitted to the maternity ward of Mahatma Gandhi Medical College and Hospital Sitapura Jaipur on 5<sup>th</sup> October 2013. She had a single ultrasound report done at 30 week which was s/o gastroschisis. The patient had a normal vaginal delivery. She delivered a male child of 2.5 kg at 1.50 pm on 5/10/2013. The apgar of the baby was 1/10 at 1 minute and 1/10 at 5 minutes; heart rate was less than 20 beats and gasping respiration. The major congenital anomalies noted were ectopia cordis, sternal cleft, pleuro- peritoneal defect, anterior diaphragmatic defect, supraumbilical wall defect with evisceration of stomach, small and large intestine, liver and spleen, kyphoscoliotic vertebral defect, aplastic right upper limb and right lung aplasia. Our suspected diagnosis is Cantrell's pentalogy. Prognosis was poor and well explained to the parents and no active intervention was done.

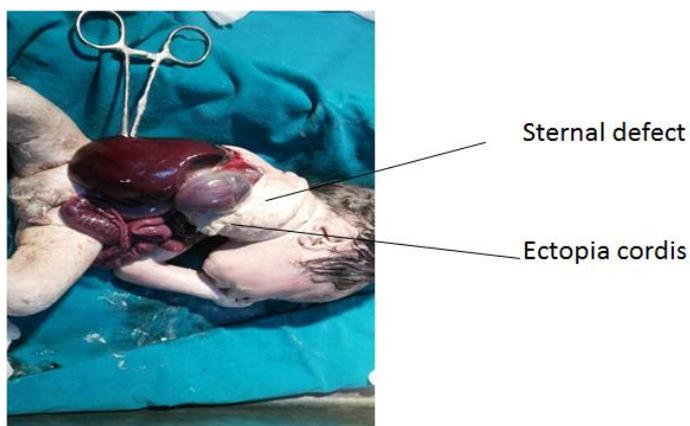


Fig-1



Fig-2

## DISCUSSION

Cantrell pentalogy is a rare congenital thoraco-abdominal disruption, first described by Cantrell *et al* with five characteristic:

- 1) Ectopia cordis and intracardiac anomalies;
- 2) lower sternal defect;
- 3) midline supraumbilical thoraco-abdominal wall defect;
- 4) anterior diaphragmatic defect; and
- 5) defect of diaphragmatic part of pericardium that results in relation between pericardial cavity and peritoneum [1].

Prevalence of pentalogy of Cantrell is about 1 per 65000 live births and classified as a developmental defect of midline anterior body wall. Full pentalogy of Cantrell is a severe and rare syndrome, but incomplete forms with combination of two or three defects were reported frequently [2]. Intracardiac anomalies that are constant portion of pentalogy of Cantrell are VSD (in 100% of cases), ASD (52%), pulmonary stenosis (33%) and Tetralogy of Fallot (20%) [1]. Also sternal fusion defect is a rare malformation and an inferior type is seen in pentalogy of Cantrell [3]. Cantrell offered a developmental failure in lateral mesoderm during day 14-18 as a reason for indecision of transverse septum of diaphragm, therefore migration of paired mesodermal

fold does not occur [1]. Failure of the transverse septum to develop, as well as abnormal development of the myocardium, cause diaphragmatic and cardiac defect, respectively [4].

Because of various phenotypes of abdominal wall defect in Cantrell's pentalogy, multiple factors is said to be responsible, including mechanical teratogens, major gene mutation, chromosomal abnormalities such as trisomy 13 and 18 and disrupted vessels defects [5]. Mutation of TAS gene which mapped at Xq25-q26.1 area is mentioned to have a role in fusion of sternum, multiple cardiac, diaphragmatic and anterior abdominal wall defects, and also additional abnormalities reported in some cases of Cantrell's pentalogy [6, 7]. Carmi *et al.*; reported some cases with encephalocele and cleft lip with or without cleft palate, in association with abdominal wall defects such as pentalogy of Cantrell [7].

In some cases of Cantrell of pentalogy, aggregation of fluid in the chest and neck cavity was reported as a result of venous congestion because of cardiac failure, increased mediastinal pressure due to diaphragmatic herniation or Omphalocele [5]. Abnormalities of the extremities are also reported in

associated with few cases of pentalogy of Cantrell [8-10]. One study reported arthrogryposis, left thumb defect and shortening of left upper limb together with exencephaly [11] Peixoto-Filho *et al.*; mentioned that clubfoot was seen in few cases [12]. Also in the presented case clubfoot was seen.

Intrauterine diagnosis of this pentalogy is impossible before 12<sup>th</sup> week of gestation, because of herniation of bowel out of abdomen is a normal event in fetal development at this time, but after that ultrasonography is a useful method even in the first trimester [12, 13]. Differential diagnosis of fetal abdominal wall defect after 12<sup>th</sup> week is Omphalocele, pentalogy of Cantrell and Gastroschisis. If midline abdominal wall defect is present together with other abnormalities specially ectopia cordis one should consider pentalogy of Cantrell [14]. Both 2D and 3D obstetric ultrasonography are recommended, but 3D ultrasonography is not necessary in first trimester [12]. Other diagnostic methods including CT-Scan and MRI can be used for confirmation [4, 15, 16]. Prognosis of pentalogy of Cantrell depends on severity of intra and extra cardiac defects, pulmonary hypoplasia, extent of abdominal wall defect, cerebral anomalies and diaphragmatic herniation. The mean survival rate without any interventional surgery is about 36 hours [3]. Studies showed that even with care monitoring in professional centers and multiple corrective surgeries, they had high morbidity and mortality rate and longtime prognosis is poor [17].

## CONCLUSION

The presented case had all portions of pentalogy of Cantrell as well as midline supraumbilical wall defect and ectopia cordis, lower sternal, pericardial and diaphragmatic defects together with bilateral deep cleft lip/palate and left side clubfoot which were reported in other case reports. These severe anomalies resulted in his death.

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