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## CASE REPORT

# Diagnosis of Pentalogy of Cantrell with Craniorachischisis at 11 weeks on 2 Dimensional Ultrasound - A rare case report

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## ABSTRACT

Pentalogy of Cantrell (PC) is an extremely rare congenital malformation, with estimated incidence from 5.5 to 7.9 per million live births. Diagnosed in second trimester or beyond with first trimester detection in hand full of cases and 2 case reports at 11 weeks in literature. An unusual combination of PC with craniorachischisis, has been reported by a few at 18-26 weeks with one at 12 weeks. Ours is the first ever presented case report diagnosed as early as 11 weeks. This patient was diagnosed with PC and craniorachischisis at 11 weeks by 2D ultrasound who underwent pregnancy termination. Presence of a throracoabdominal defect at such early gestation should alert sonologists to include PC in differentials. This rare combination of dorsal and ventral midline defects occurring at a similar embryologic timeline could point to a common inciting event that needs to be further investigated.

**Keywords:** pentalogy of cantrell, cantrell syndrome, craniorachischisis, first trimester ultrasound.

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## Introduction

First described by Cantrell in 1958, pentalogy of Cantrell (PC) is a constellation of five defects that includes lower sternum defect, a midline supraumblical thoracoabdominal wall defect, a defect of anterior diaphragm, a defect of diaphragmatic pericardium and congenital cardiac anomalies[1].

Although case reports of such patterns date back to early 1700s, it was Cantrell in 1958 who unified this pattern into a clinical syndrome, proposed its pathophysiology and clinical management, majority of which holds true even 6 decades later barring certain advances in postnatal approaches that improved survival in certain subtypes.

**Case:**

A 28 years old third gravida with nil significant previous obstetric history, presented to us for routine antenatal care. Pregnancy was from a non consanguineous marriage without a history of congenital defects or chromosomal anomalies or medical disorders. She was on folic acid supplementation. Aneuploidy scan at 11 1/7 weeks done using Voluson P8 showed a crown rump length (CRL) of 30 mm corresponding to 10 weeks and nuchal translucency (NT) could not be measured, abdominal contents freely floating in the amniotic fluid outside the abdominal cavity. Heart was located outside the thoracic cavity with good cardiac activity as shown in Fig. 1. Diaphragm appeared to be slightly elevated. Better and precise delineation of intracardiac, diaphragmatic and sternal defects would require scan at a later gestation. Cranial vault was not formed and a vertebral bony defect was noted posteriorly with exposed neural tissue extending up to the lumbar area. A comparison of the neural tube defect in the antenatal scan and the expelled fetus are shown in Fig. 2. The findings were a peculiar combination of dorsal and ventral midline defects including ectopia cordis, omphalocoele/gastroschisis, eventuation and craniorachischisis. A presumptive diagnosis of PC with craniorachischisis was made.

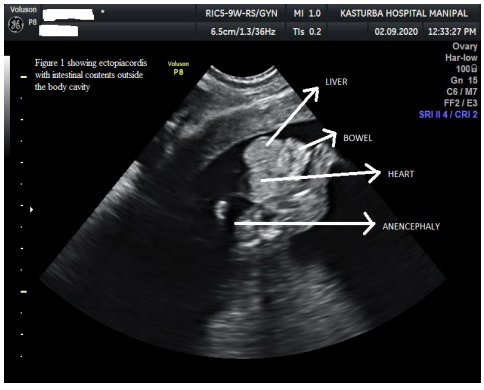


**Fig. 1.** Image showing abdominal contents and heart outside the body cavity.

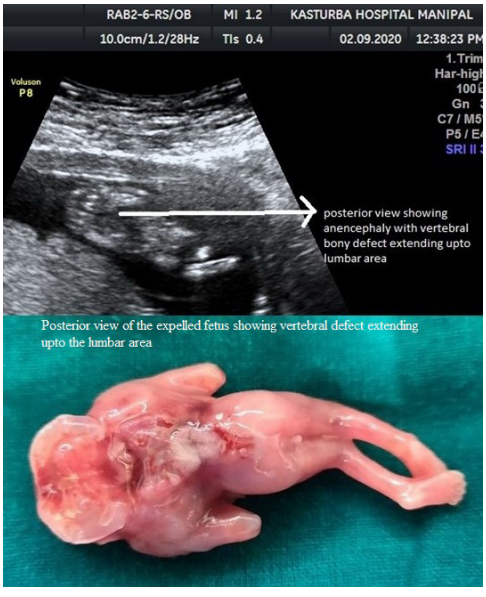


**Fig. 2.** Comparison of the vertebral defect in the antenatal scan and after expulsion of the fetus.

Though diagnosis of anterior abdominal wall defect cannot be made before 12 weeks of gestation, we suspected a probable gastroschisis since it was also associated with ectopia cordis and dorsal midline defects. PC has a varied presentation and the prognosis depends greatly on the severity of condition, especially the ectopia cordis, intracardiac defects, extent of thoracoabdominal defect and its associated anomalies. The associated craniorachischisis in this

patient is a lethal condition which was explained to the couple in detail who opted for pregnancy termination. The couple deferred genetic evaluation, karyotype analysis and autopsy. Termination of pregnancy was done at 12 weeks using 400 micrograms of vaginal misoprostol tablets every 3 hour for 3 doses (which is half the usual dose as per the International Federation of Gynecology and Obstetrics protocol as this patient is a case of previous low transverse cesarean section). A dead male fetus of 130 g was expelled after 3rd dose. Gross examination of the fetus showed anencephaly with prominent orbits and absence of posterior vertebral elements with splaying of the lamina and exposed spinal cord involving up to lumbar vertebrae, suggestive of craniorachischisis totalis. There was a lower sternal defect with the heart noted outside chest wall and upper abdominal wall defect with exposed stomach and intestines without a covering membrane suggestive of gastroschisis. These findings are shown in Fig. 3, 4. Upper and lower limbs, anus and external genitalia appeared normal.

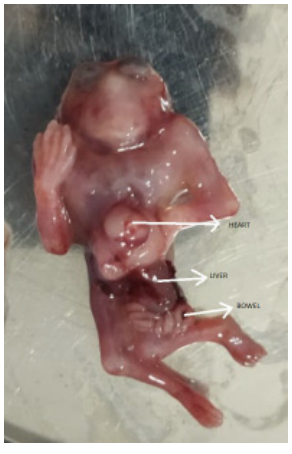


**Fig. 3.** Anterior view showing anencephaly with protruding orbits with heart, liver and bowel outside the body cavity.

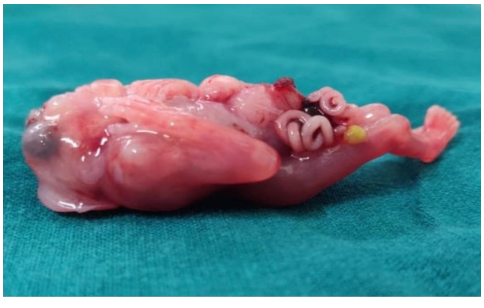

**Fig. 4.** Lateral view showing anenchephaly and thoracoabdominal defect.

## Discussion

Around 250 cases of PC have been reported to date with entire spectrum occurring rarely[2]. The pathogenesis originally described by Cantrell (1958) occurs between 14 and 18 days of embryonic life. First the defects in anterior diaphragm, inferior pericardium and heart occur due to anomalous development of septum transversum and its adjacent somatic and splanchnic mesoderm. Second, abdominal wall defects occur due to defective attachment of abdominal

musculature to the improperly formed primordial sternum[1]. Craniorachischisis is the open cranial defect (anencephaly/exencephaly) continuous with a complete spinal dysraphism and represents a complete failure of neurulation resulting from dysmorphogenesis occurring no later than 20-22 days after conception, which coincides with occurrence of PC on the embryologic timeline[3]. It is proposed that time point of embryogenetic insult decides extent and severity of malformation. Hence this explains wide spectrum of PC and associated neural tube defects (NTD) reported so far, including ours which describes one of the severe types of NTDs where the inciting event must have probably occurred very early in the timeline of embryogenesis. Co-occurrence of such ventral and dorsal midline defects suggests possibility of common genetic and environmental factors behind their coexistence that has to be addressed in future research.

Any genetic factors and etiological causes related to PC are unknown. Though a possibility of familial inheritance and association with chromosomal anomalies like trisomy 18, 13 and Turners syndrome has been suggested, cases have been sporadic in occurrence mostly[4].

Toyama in 1972 proposed a classification that grouped various cases with less than 5 defects and incomplete presentations as subtypes of Cantrell syndrome[5]. Recent studies have stressed that strict classification is not as important as a thorough description and understanding of congenital abnormalities in utero. Similarly, Coleman et al studied PC and two closely related syndromes with overlapping features namely OEIS (omphalocoele, exstrophy, imperforate anus, spina bifida) and limb-body wall complex (LBWC) as a spectrum of diseases resulting from improper closure of lateral and craniocaudal folds. They concluded that degree of pulmonary hypoplasia was far more indicative of prognosis than any specific classification system per se. In counseling families, planning delivery and management, a descriptive diagnosis is more valuable than trying to line characteristics up with a specific syndrome or a subtype within syndrome[6].

Diagnosis of PC in first trimester was first described by Bennett, using 2D sonography at 12 weeks[7]. Subsequently few more cases at 12-13 weeks have been published[8]. Earliest diagnosis at 11 weeks was done in two case reports apart from ours which described an associated single umbilical artery and called it "hexology"[9] and the other had associated limb defect and lardoscoliosis[10] which were not seen in our case. However our patient had an associated craniorachischisis additionally. A large retrospective analysis over a period of 9 years done on 100,997 pregnancies to evaluate performance of first trimester scan on detection of non-chromosomal anomalies showed fetal anomalies in 1.7% pregnancies with 27.6% of the anomalies being detected during 11 to 13 weeks gestation while 53.8% detected on the second trimester scan and 18.6% detected in the third trimester or postnatally. There were only 2 reported cases of PC in this study both of which were detected in first trimester scan. However, there was no associated craniorachischisis in these cases[11].

Another retrospective analysis was done on 6,366 twin pregnancies between 2002 and 2019 to analyze the performance of 11 to 13 weeks scan in detection of fetal anomalies which had one case of isolated PC without craniorachischisis that was detected in first trimester. The prevalence of this anomaly in twin gestation was said to be 1 in 12,732 compared to 1 in 50,499 among singleton pregnancies[12].

A systematic review on 67 total reported cases of PC diagnosed in the first trimester of pregnancy (published from January 1980 to July 2019) suggested that ultrasound findings of a coexistent omphalocele and ectopia cordis at 11 to 13 weeks scan is highly suggestive of PC[13].

PC in association with craniorachischisis was reported at 12 weeks using 3D ultrasound[14]. Present case report is first of its kind in reporting PC in combination with craniorachischisis by 2D ultrasound at 11 weeks. Early diagnosis in first trimester will assist in early pregnancy termination thereby limiting the physical and emotional anguish to the couple.

We conclude that though the intricate details of

the syndrome may not be appreciable at such early gestation, presence of a thoraco-abdominal defect should alert the sonologists to include PC in differentials and look for associated anomalies that can improve patient care.

## Potential conflicts of interest

The authors declare no conflicts of interest.

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