# First Trimester Diagnosis of Cantrell Pentalogy: A Case Report

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**ABSTRACT**

Pentalogy of Cantrell is a rare syndrome with a deficiency of anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, various congenital intracardiac abnormalities and a defect of the lower sternum.

Herein a fetus with Cantrell Pentalogy who was diagnosed in the first trimester was presented and discussed in the light of the literature.

**Keywords:** Cantrell pentalogy, Congenital cardiac abnormalities, Abdominal wall defect

Gynecol Obstet Reprod Med 2015;21:104-105

## Introduction

Pentalogy of Cantrell was described in 1958 by Cantrell. It was named as pentalogy because of the presence of five major malformations, which include a midline, upper abdominal wall abnormality, lower sternal defect, anterior diaphragmatic defect, diaphragmatic pericardial defect and congenital heart abnormalities.[1,2]

By the time the classification of the syndrome was suggested as class 1, definite diagnosis with 5 defects; class 2, diagnosis with 4 defects and class 3, incomplete expression.[3]

## Case Report

A-30-year-old woman, gravida 1, para 0, presented to our hospital's perinatology unit for a routine obstetric scan in the 11th week of gestation. Of note, her history and prenatal course were unremarkable up to this point.

On sonographic evaluation, the fetus had 7 mm nuchal translucancy, large supraumbilical omphalocele (3x5 cm approximately), ectopia cordis, diaphragmatic defects (Figure 1). After chorion villus sampling (CVS) therapeutic abortion was performed.

The fetus and placenta were submitted for postmortem examination. Autopsy revealed a male fetus with cardiac anomalies, including hypoplastic left ventricule, sternal defect, abdominal wall defect and cleft palate. CVS result was normal.

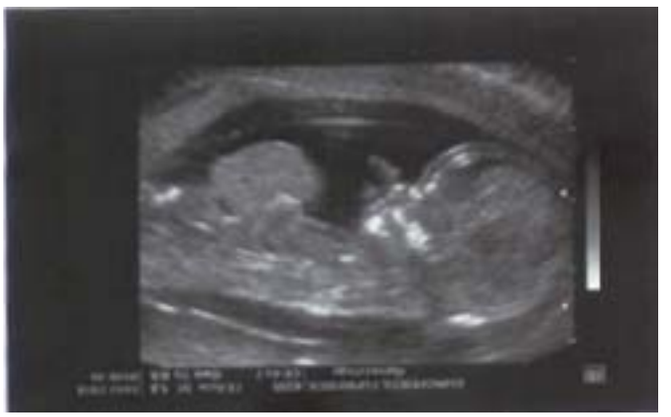

*Figure 1: Ultrasound photo of abdominal wall defect and increased nuchal translucency*

## Discussion

The pentalogy of Cantrell (PC) is a rare syndrome with an estimated incidence of 5.5 per 1 million live births.[2,4] It is described as a deficiency of anterior diaphragm, a midline supraumbilical abdominal wall defect, a defect in the diaphragmatic pericardium, various congenital intracardiac abnormalities and a defect of the lower sternum. The pathogenesis of Cantrell is not known exactly. Cantrell et al suggested an embryologic developmental failure of a segment of the lateral mesoderm.[5,6]

The etiology of PC is also not known. Most cases are sporadic and no recurrences have been reported. The is male dominance with a male to female ratio of 2.7/1.[7]

Our case was male and sporadic similar to the literature. In our case all the five compenents of the syndrome were observed. Cleft palate would not be diagnosed by ultrasound antenatally because of the difficulties of the first trimester diagnosing of the cleft palate.

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*Submitted for Publication:* *28. 03. 2014*
*Accepted for Publication:* *14. 05. 2014*

In the literature not so many cases were reported in the first trimester by using two-dimensional-ultrasound.

Peixoto-Filho et al reported two cases at 10th and 11th weeks of gestation in which the patients were preffered to terminate thier pregnancies just like in our case.[8]

Most of the cases in the literature diagnosed in the second trimester.[9,10] There are also cases which were diagnosed after birth in newborn period.[11,12]

Magadum S et al reported a case of 11-year-old with incomplete PC.[13]

In recent years 3D sonography is used widely also in diagnosing PC. It has been also suggested that magnetic resonance imaging (MRI) and prenatal fetal echocardiography provide optimal assessment of fetuses with this syndrome.[10]

However,in PC large defects are observed in cases so 2D sonography can be enough to diagnose the pentalogy just like in our case.

In conclusion when a case with omphalocele and cardiac abnormalities are observed on ultrasound, pentalogy of Cantrell should be remembered. A 2D sonography with high resolution can be enough to diagnose the pentalogy in the first trimester.

## Cantrell Pentalojisinin İlk Trimester Tanısı: Olgu Sunumu

**ÖZET**

Cantrell Pentalojisi, ön diyafram defekti, orta hat göbek üstü abdominal defekt, perikardiyum yokluğu, çeşitli intrakardiyak anomaliler ve alt sternal defekt ile görülen nadir bir sendromdur.

Burda ilk trimesterde tanı konan Cantrell Pentalojisi sunuldu ve literatür ışığında tartışıldı.

**Anahtar Kelimeler:** Cantrell pentalojisi, Konjenital kalp anomalileri, Abdominal duvar defektleri

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