



# Cantrell's syndrome revisited: a case with increased nuchal fold diagnosed in the first trimester

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## Abstract

**Objective:** Our aim was to evaluate the characteristics of a case of pentalogy of Cantrell diagnosed at our obstetrics department.

**Case:** A 25-year-old pregnant patient, unaware of date of last menstrual period, applied for first trimester anomaly scans. A transabdominal ultrasound evaluation revealed a fetus of 11 weeks, 5 days by CRL measurements. The fetus had a septated cystic hygroma, thickest measurement 6 mm, and accompanying wide defect of the fetal abdominal wall. A large part of the liver and heart were extracorporeal in the area of the defect. The patient was thought to suffer Cantrell's syndrome due to fetal omphalocele and ectopia cordis.

**Conclusion:** In cases of first trimester ultrasonography identifying omphalocele, pentalogy of Cantrell should be kept in mind and ultrasonographic scans should be deepened to carefully determine whether the heart is contained within the omphalocele sac.

**Key words:** Cantrell's syndrome, cardiac malformation, omphalocele.

## Artmış ense kalınlığı ile birlikte ilk trimesterde tanı konulmuş olgu nedeniyle Cantrell sendromunun gözden geçirilmesi

**Amaç:** Bu çalışmada obstetrik kliniğimizde tanı konulan bir Cantrell pentalojisi olgusunun tartışılması amaçlanmaktadır.

**Olgu:** Son adet tarihini bilmeyen 25 yaşındaki gebe birinci trimester anomali taraması için başvurdu. Transabdominal ultrasonografik değerlendirmede CRL ölçümüne göre 11 hafta 5 gün olarak ölçülen fetusta en kalın yerde 6 mm ölçülen septalı kistik higroma ve eşlik eden fetal karın ön duvarında geniş bir defektin bulunduğu görüldü. Karaciğer ve kalbin büyük bir kısmının bu defekt düzeyinde ekstrakorporeal yerleşimli olduğu saptandı. Hastada fetal omfalosel ve ektopia kordis bulguları ile Cantrell sendromu düşünüldü.

**Sonuç:** Ultrasonografik olarak birinci trimesterde omfalosel saptanan olgularda Cantrell pentalojisi olasılığı akla gelmeli ve ultrasonografik tarama derinleştirilmeli omfalosel kesesi içerisinde kalp olup olmadığı dikkatlice değerlendirilmelidir.

**Anahtar sözcükler:** Cantrell sendromu, kardiyak malformasyon, omfalosel.

## Introduction

Cantrell's syndrome (or pentalogy of Cantrell), a rare thoraco-abdominal developmental disorder, was first described by Cantrell et al. in 1958.<sup>[1]</sup> The syndrome includes defects of the abdominal wall, pericardium, diaphragm, sternum and heart. Anomalies of the lower sternum and midline abdominal wall defects with accompanying diaphragm and frontal pericardiac defects are the characteristics of the syndrome.<sup>[2]</sup>

Differential diagnosis should be made keeping in mind ectopia cordis, simple omphalocele and amniotic band syndrome.<sup>[3]</sup> Cardiac anomalies, which are useful in determining prognosis, are included in the anomalies of the disorder. In the literature case reports of major cardiac anomalies such as atrial septal defects, ventricular septal defects and tetralogy of Fallot are frequently reported.<sup>[4]</sup> To our knowledge, early diagnosis of Cantrell's syndrome is rare with only a few cases of

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diagnosis in the first trimester, and there are a few reports about diagnosis of Cantrell's syndrome which is including increased nuchal fold in first trimester.

This article presents the case report of a prenatal, early diagnosis of Cantrell's syndrome with increased nuchal fold, and aims to discuss Cantrell's syndrome together with a review of the literature.

## Case Report

A 25-year-old pregnant patient, unaware of date of last menstrual period, applied for first trimester anomaly scans. The patient, married to a third degree relative, was pregnant for the second time, the first ending in abortion (2 months). The patient was not taking folic acid due to hyperemesis gravidarum.

A transabdominal ultrasound evaluation revealed a fetus of 11 weeks, 5 days by CRL measurements. The fetus had a septated cystic hygroma, thickest measurement 6 mm, and accompanying wide defect of the fetal abdominal wall. A large part of the liver and heart were extracorporeal in the area of the defect (**Figs. 1 and 2**). The patient was thought to suffer Cantrell's syndrome due to fetal omphalocele and ectopia cordis. With perinatology council decision, taking account of the patient and her partners' wishes and permission, a medical

evacuation was performed. Monitoring after the termination showed no abnormal results and the patient was discharged.

## Discussion

Pentalogy of Cantrell is a syndrome characterized by full or partial ectopia cordis, together with omphalocele, congenital heart disease and defects of the pericardium, lower sternum and anterior diaphragm. Pentalogy of Cantrell is a rare syndrome seen in 1/65,000 to 1/200,000 births. Male to female ratio is 1:1.<sup>[5]</sup> Though the pathogeny is not fully understood, it's thought that pentalogy of Cantrell results from faulty mesoderm cell development, differentiation and migration in the first weeks of embryonic life. There is no family predisposition known. Isolated cases of Cantrell's syndrome with trisomy 18 and trisomy 21 are known.<sup>[6]</sup> Literature search showed that even without all parameters, diagnosis of Cantrell's syndrome based on identification of ectopia cordis and omphalocele can be made in the 1st trimester. In this case, two major ultrasonographic results were used to make the diagnosis.

Early diagnosis in the first trimester using nuchal translucency measurements is thought to identify chromosomal anomalies as well as cardiac anomalies which



**Fig. 1.** Transabdominal axial ultrasound image showing omphalocele sac containing heart and liver.



**Fig. 2.** Transabdominal sagittal ultrasound image showing septated cystic hygroma and heart located outside the thorax.

cause significant increased NT.<sup>[7]</sup> Cystic hygroma identified in the first trimester, which can accompany Cantrell's syndrome, may be a noticeable, early symptom. While cases of Cantrell's syndrome babies born when pregnancy is not terminated generally have a negative prognosis with cardiac anomalies, it is basically the degrees of cardiac anomaly and accompanying other extra cardiac anomalies which determine the prognosis. Very few cases survive after corrective surgery.<sup>[8]</sup>

### Conclusion

In cases ultrasonographically diagnosed to have omphacele during first trimester, pentalogy of Cantrell should be kept in mind and ultrasonographic scans should be deepened to carefully determine whether the heart is contained within the omphalocele sac. Early diagnosis in pregnancy weeks 10-14 can be aided by first trimester scanning tests such as NT measurements which can help to identify chromosomal anomalies as well as cardiac anomalies and thus can be a warning sign for diagnosis of Cantrell's syndrome.

**Conflicts of Interest:** No conflicts declared.

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