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Pentalogy Of Cantrell In One Fetus Of A Twin Pregnancy Diagnosed At First Trimester

Birinci trimesterde tanı alan bir ikiz gebeliğin tek eşinde Cantrell Pentalojisi

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ÖZET

Cantrell Pentalojisi, insidansı 1/100.000 olarak bildirilen ve beş karakteristik bulgu ile kendini gösteren nadir bir konjenital anomalidir. Bulguları: ektopia kordis ve intrakardiyak anomaliler, sternum alt kısmında defekt, supraumblikal torakoabdominal duvar defekti, diafram ön kısmında defekt ve perikardiyumun diafragmatik kısmında defekt olarak sıralanabilir. Sendromun etyolojisi tam olarak belirlemiş değildir. Burada ikiz olarak başlayan gebelikte erken dönemde tanı koyduğumuz Cantrell Pentalojisi vakasını sunuyoruz. 34 yaşında diamniyotik di koryonik ikiz gebelik ön tanısıyla başvuran hastanın ultrasonografisinde fetuslardan birinde kardiyak aktivite saptanamamış ve diğer fetusun da ense saydamlığı (NT) 5.38 mm olarak ölçülmüştü. Bu fetüsün detaylı ultrasonografik incelemesi geniş omfalosel ve karın duvarı defektinin üst komşuluğunda regüler fetal kalp atımının fetal toraksın dışında izlendiğini ortaya koyması üzerine terminasyon kararı alındı. Terminasyon sonrası detaylı anatomik inceleme bulguları Cantrell Pentalojisini işaret etmekte idi. Bu nadir anomalinin ilk trimesterde erken tanısı ile ilgili az miktarda vaka takdimi mevcuttur. Bu denli ağır komponentler içeren anomalilerin erken prenatal tanısı perinatoloji pratiğinde önemli bir unsurdur.

Anahtar Kelimeler: Cantrell pentaloji, anomali, prenatal tanı

ABSTRACT

Pentalogy of Cantrell is a rare congenital anomaly, with an incidence of 1/100.000 and has five characteristic findings: Ectopia cordis and intracardiac anomalies; defect of lower sternum; midline supraumbilical thoraco-abdominal wall defect; anterior diaphragmatic defect; and defect of diaphragmatic part of pericardium. The exact aetiology of the syndrome is still unknown. Here we report an early diagnosed pentalogy of Cantrell in a twin pregnancy. A 34-year-old patient with a diamniotic and dichorionic twin pregnancy in which one of the fetuses had no heart activity and other fetus had a nuchal translucency of 5.38 mm. Ultrasonographic examination revealed a large omphalocele and above the defective abdominal wall regular fetal heart rate was seen out of the fetal thorax. After termination of the pregnancy anatomic examination revealed findings meeting the criteria of Pentalogy of Cantrell. Since its early detection is found in a few cases we report a very early detection of a rare abnormality. Early detection of an entity with a poor prognosis is extremely important in the prenatal medicine practice.

Key Words: Cantrell pentalogy, anomaly, prenatal diagnosis

Introduction

Anterior abdominal wall defects are of a large spectrum of fetal anomalies and usually appear as a part of complex syndromes. Isolated ectopia cordis is a rare congenital anomaly, in which the fetal heart is partially or completely located outside the thoracic cavity. The condition is extremely rare and its estimated prevalence is 5.5–7.9 per million live births (1). It is usually associated with pentalogy of Cantrell (2).

Pentalogy of Cantrell is a rare congenital anomaly, first described by Cantrell et al in 1958, in 5 cases with five characteristic findings: Ectopia cordis and intracardiac anomalies; defect of lower sternum; midline supra umbilical thoraco-abdominal wall defect; anterior diaphragmatic defect; and defect of diaphragmatic part of pericardium that results in relation between pericardial cavity and peritoneum (3). Pentalogy of Cantrell is a rare condition with an incidence of 1/100.000 (4). There are cases with the complete syndrome and incomplete variants. Although the exact etiology is unknown, there are case reports of the entity associated with chromosomal abnormalities and other congenital defects in the literature (5). As the prognosis depends on the severity of the abnormality, and is usually determined as poor; antenatal diagnosis is crucial for the parental informing and the decision making about the continuing pregnancy. Here we report an early diagnosed pentalogy of Cantrell in a twin pregnancy.

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Case Report

A 34-year-old patient with a diamniotic and dichorionic twin pregnancy, was 17 weeks and 3 days according to the last menstrual period admitted to the outpatient clinic for a routine first trimester control. It was her fourth pregnancy; she had two spontaneous labors and one dilatation curettage. The patient and her husband were neither relatives, nor any family history of chromosomal abnormalities, congenital anomalies or birth defects and she did not experience any of them in previous pregnancies. The patient did not have any chronic health problems and any complications related with the pregnancy. Her current pregnancy was a spontaneous multiple pregnancy.

Ultrasonographic assessment revealed that one of the fetuses had no heart activity and other fetus had a nuchal translucency of 5.38 mm (Figure 1).The fetus was measured as 12 weeks and 4 days according to crown- rump length. Further examination of the living fetus revealed ectopia cordis and anterior abdominal wall defect. Abdominal wall defect was a large omphalocele and above the defective abdominal wall regular fetal heart rate was seen clearly out of the fetal thorax (Figure 2).

Figure 1: Ultrasonographic measurement of nuchal translucency (5.38 mm).

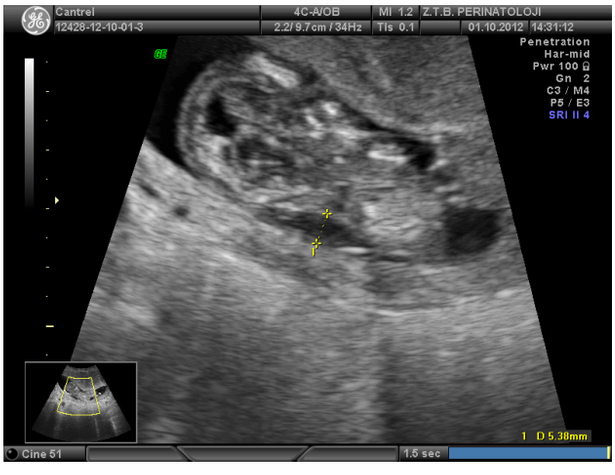
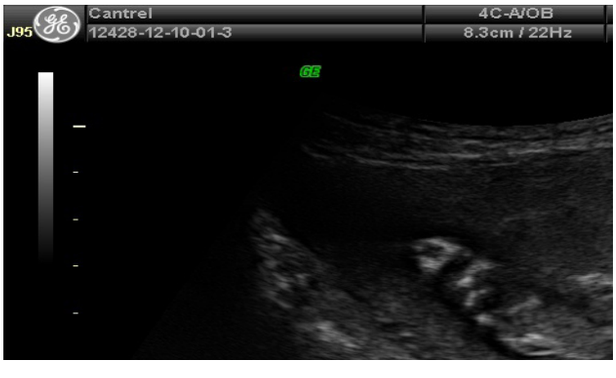


Figure 2: Fetal heart seen out of the thorax at the ultrasound examination



After detailed information about the findings, possible diagnosis and poor prognosis of the entity, family decided the elective termination of the pregnancy. Pregnancy was terminated and a postabortal complete anatomical examination was performed.

There was an anterior defect at the abdominal wall located just above the umbilicus (Figure 3) and the heart, liver, intestines were all placed out of the abdominal cavity from this defect (Figure 4). The edema located at the neck of the fetus was clearly visible (Figure 5).

Figure 3: Fetal omphalocele detected during detailed anatomic examination

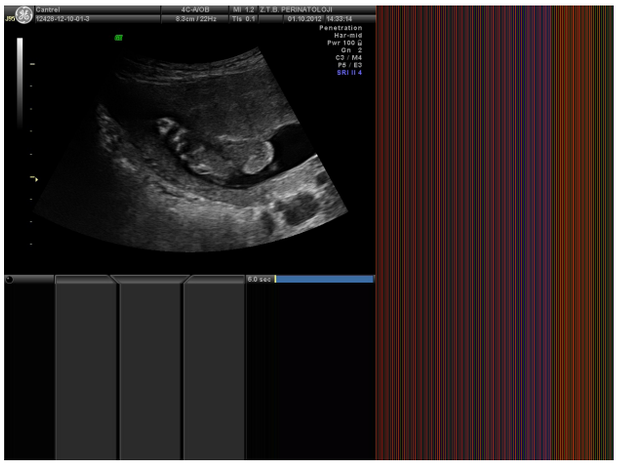


Figure 4: Fetal heart and liver seen out the thorax and abdominal wall

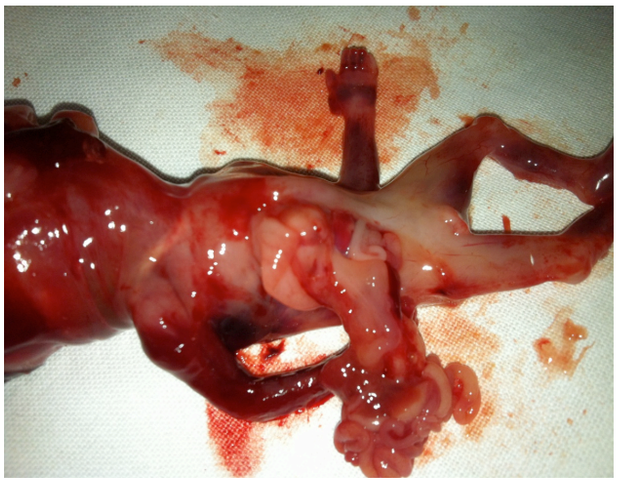
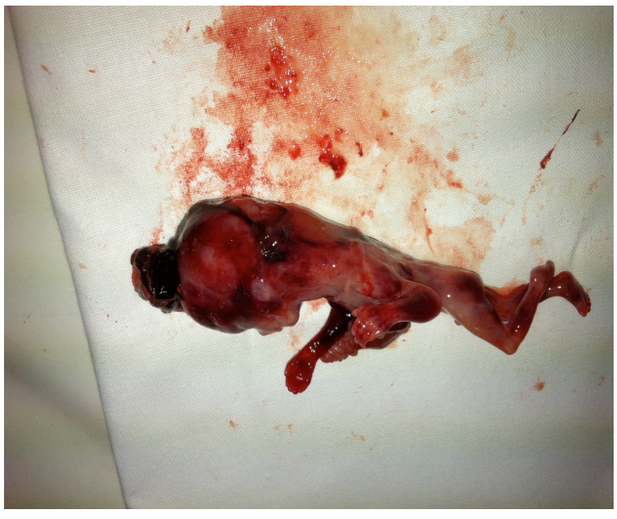


Figure 5: Edema observed at the neck of the fetus



Discussion

Full Pentalogy of Cantrell is very rare, but incomplete forms with combination of two or three defects were reported more frequently. Common intra-cardiac anomalies seen with Pentalogy of Cantrell are ventricular septal defect, ostium secundum atrial septal defect, pulmonary stenosis or atresia and Tetralogy of Fallot (6).The exact aetiology of the syndrome is still unknown. Cantrell offered a developmental failure in lateral mesoderm during day 14-18 as a reason for indecision of transverse septum of diaphragm, therefore thought to involve failure of the lateral body folds to fuse in the thoracic region. Failure of the transverse septum to develop, as well as abnormal development of the myocardium, cause diaphragmatic and cardiac defect (7,8).

Because of the poor prognosis even with the treatment after birth; early prenatal detection is really important. Midline abdominal wall defect detected together with other abnormalities especially with ectopia cordis; Pentalogy of Cantrell should be considered as the most possible diagnosis. Both 2D and 3D examination can be performed but 3D is not necessary for the diagnosis (9).

In the present case, increased measurement of nuchal translucency (NT) was the key factor which was the alerting signal for seeking other possible abnormal findings. Increased fetal nuchal translucency can be an early result of cardiac malformation and increased mediastinal pressure because of the diaphragmatic herniation or omphalocele. This finding reveals the importance of the first trimester screening and NT examination for determination of any congenital abnormalities or chromosomal disorders (10).

Since its early detection is found in a few cases we report a very early detection of a rare abnormality. Early detection of an entity with a poor prognosis is extremely important in the prenatal medicine practice.

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