Prenatal Diagnosis of Pentalogy of Cantrell with Increased Nuchal Translucency: Case Report

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Pentalogy of Cantrell is a rare sporadic syndrome with unknown etiology. We aimed to present a prenatally diagnosed case of pentalogy of Cantrell associated with increased nuchal translucency (NT). A twenty-two year old G2P0A1 pregnant woman was admitted to our clinic for routine prenatal ultrasonographic scanning at 14th weeks of gestation. Ultrasound scan revealed a NT of 7.9 mm and a large omphalocele containing the liver, bowel, and heart with ventricular septal defect. After taking the written informed consent form the parents the pregnancy was terminated. Postmortem examination confirmed the ultrasound findings and chromosome analysis revealed a normal karyotype. Increased NT measurement in first trimester of pregnancy may also bring the fetus to early attention for pentology of Cantrell if an omphalocele associates with this condition.

Key Words: Pentalogy of Cantrell, Increased nuchal translucency, Omphalocele, Prenatal diagnosis

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Introduction

Pentology of Cantrell is a rare congenital anomaly that was first described in 1958 by Cantrell et al.[1] The syndrome occures in approximately 5.5 per 1 million live births.[2] The anomaly is associated with a deficiency of the 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.[1] Although the condition is thought to be a sporadic congenital anomaly, there is a report of a family where three brothers were born consecutively with this abnormality.[3]

The survival rate for neonates born with pentalogy of Cantrell is low[4] and it is lethal without corrective or palliative operations.[5] The survival rate is higher among the cases with the absence of significant extracardiac defects.[4] Toyama[6] reported a 20% survival rate in this disorder including its variants and incomplete forms. Fernández et al.[7] reported a survival rate of 8.5% (5/59) in the complete pentology of Cantrell.

We report a case of pentalogy of Cantrell with increased nuchal tranlucency (NT) detected by ultrasonography at 14th weeks of gestation.

Case Report

A twenty-two year old G2P0A1 pregnant woman was admitted to the Kahramanmaras Sutcu Imam University Obstetrics Outpatient Clinic for routine prenatal ultrasonographic scanning at 14th weeks of gestation. Ultrasound scan revealed a CRL of 86.24 mm and an NT of 7.9 mm (Figure 1). In addition to this, a large omphalocele containing the liver, bowel, and heart was detected (Figure 2). We also detected a ventricular septal defect (VSD) at heart (Figure 3). After discussing the diagnosis of pentalogy of Cantrell and related prognosis with the parents, a chromosomal analysis (choronic villus sampling) was performed and the pregnancy was terminated at 14th weeks of gestational age after their written informed consent form had been obtained. Postmortem examination revealed an omphalocele with herniation of the bowel and liver and a midline sternal defect with ectopia cordis (Figure 4). The extremities, head and spine seemed to be normal. As the couple did not permit autopsy, to confirm the prenatal ultrasound findings was not possible. Chromosome analysis revealed a normal karyotype.

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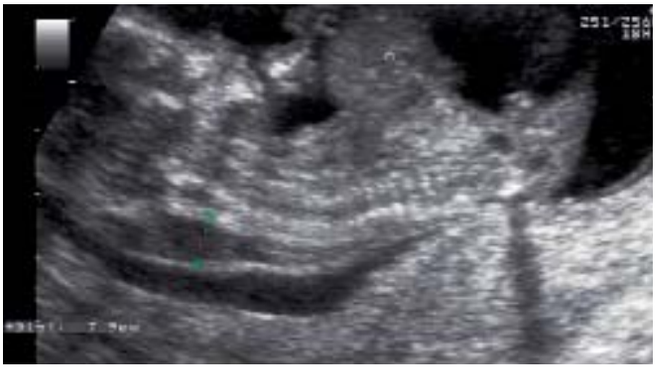

Figure 1: Transverse scan of the fetus showing increased nuchal tranlucency and omphalocele (o: omphalocele).

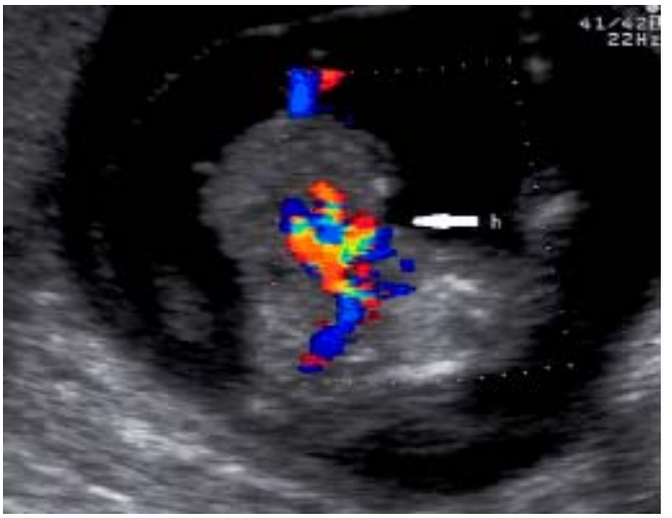

Figure 2: Doppler ultrasound view of the omphalocele containing liver, bowel and heart (h: heart).

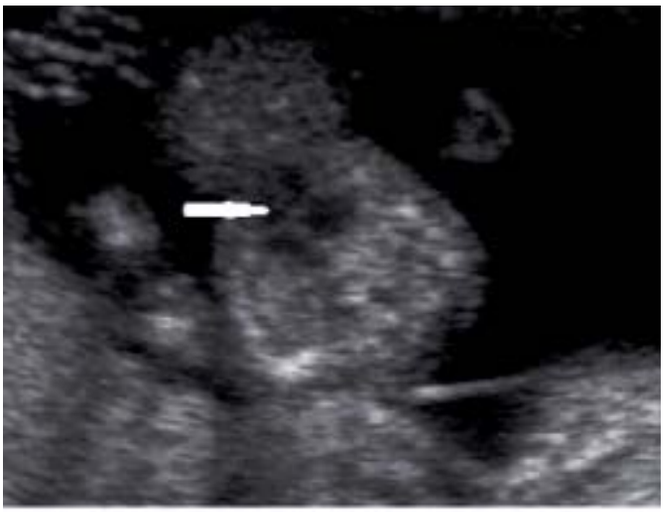

Figure 3: Ultrasound view showing the ventricular septal defect at heart (white arrow).

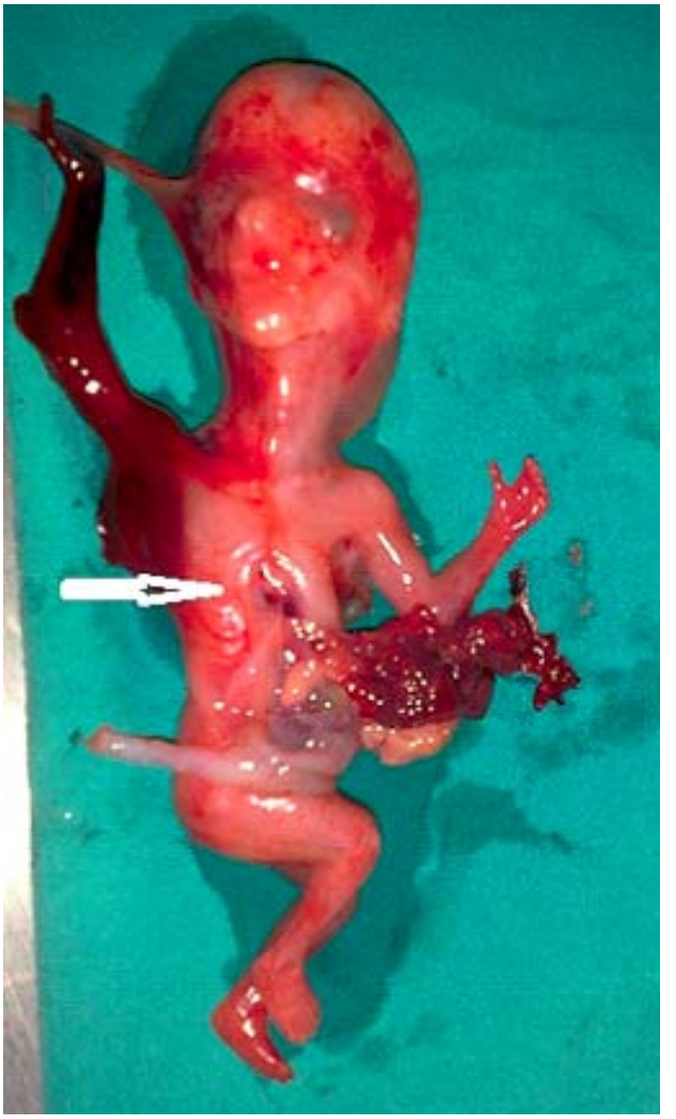

Figure 4: Anterior view of the fetus showing midline sternal defect (white arrow) and omphalocele containing liver, bowel and heart.

Discussion

Pentalogy of Cantrell is a rare entity of congenital defects in which a ventral (anterior) diaphragmatic hernia occures in association with an omphalocele.[1] The pathogenesis of pentalogy of Cantrell has not been fully elucidated. A widely-accepted theory, which was proposed by Cantrell et al. [1] stated that embryologic developmental failure of a segment of the lateral mesoderm around gestational age 14-18 days is responsible for the variety of abnormalities detected in the pentalogy of Cantrell. Consequently, the transverse septum of the diaphragm does not develop, and the ventromedial migration of the paired mesodermal folds of the upper abdomen does not occur. Organs may eviscerate through the resulting sternal and abdominal wall defects.[1]

The pentalogy of Cantrell can be diagnosed in the first trimester of pregnancy at the prenatal ultrasonographic examination.[8] Although increased fetal NT is usually associated with chromosomal aberrations such as Down syndrome, it is also predictive for fetal malformations such as cardiac anomalies or an omphalocele.[9] Increased NT very rarely associate with pentalogy of Cantrell and there were 4 cases reported in the literature.[9-11] Our case is the 5th pentalogy of Cantrell case associated with increased NT. So increased NT measurement in first trimester may also bring the fetus to early attention for pentology of Cantrell.

The abdominal wall defects in pentalogy of Cantrell include omphalocele, diastasis recti, epigastric hernia, umbilical hernia, and combined defects. The most common abdominal wall defect in cases with pentalogy of Cantrell is omphalocele.[6,12] In a fetus with omphalocele, pentalogy of Cantrell should be ruled out.[13] The abdominal wall defect in our case was an omphalocele. The embryological process of elongation of the midgut with herniation into the base of the umbilical cord is still in progress in the normal fetus at 12 weeks of gestation so the diagnosis of a ventral abdominal defect should be avoided before this week.[14] Our case was at 14th weeks of gestation, and a significant degree of protrusion of the heart outside the chest was observed.

Cardiac abnormalities are the most common anomalies associated with pentology of Cantrell. Cantrell et al.[1] stated that congenital intracardiac anomalies are consistent elements of the pentalogy, with VSD in every case (100%), atrial septal efect in 53%, pulmonary stenosis in 33%, tetralogy of Fallot in 20% and left ventricular diverticulum in 20%. In our case we detected VSD at ultrasound, but we could not confirm this finding at autopsy.

Although most cases of pentalogy of Cantrell are sporadic, expression of thoracoabdominal syndrome, pentalogy of Cantrell accepted in this category in the genetics literature, has been noted to show an X-linked dominant inheritance with lung and diaphragmatic anomalies more common in males.[3] Also it is known that concurrent structural and / or chromosomal abnormalities may complicate upto 50 to 75% of cases presenting with omphaloceles.[15,16] So, in cases of pentalogy of Cantrell parents should be counseled regarding future family planning and the possibility of a genetic link and antenatal invasive fetal testing should be offered. The karyotype analysis of our case was reported as normal. The cases with normal karyotype are sporadic, and they have a negligible recurrance risk.[17]

In conclusion, increased NT very rarely associates with pentalogy of Cantrell and there were 4 cases reported in the literature. So if increased NT measurement in first trimester associates with an omphalocele, pentalogy of Cantrell should be considered in the differential diagnosis as a cause of increased NT.

Artmış Nukal Kalınlığın Eşlik Ettiği Cantrell Pentalojisinin Prenatal Tanısı: Olgu Sunumu

Cantrell pentalojisi etyolojisi bilinmeyen nadir bir sporadik sendromdur. Biz bu olgu sunumunda, artmış nukal kalınlığın eşlik ettiği ve prenatal tanısı konmuş Cantrell Pentalojisi olgusunu sunmayı amaçladık. Yirmi-iki yaşında G2P0A1 olan gebe gebeliğinin 14. haftasında rutin prenatal ultrason taraması için kliniğimize başvurdu. Ultrasonografik incelemede 7,9 mm olan nukal kalınlık ve karaciger, barsak ve ventriküler septal defektin saptandığı kalbi de içeren büyük bir omfalosel izlendi. Anne ve babanın yazılı onayı alındıktan sonra gebelik sonlandırıldı. Postmortem incelemede ultrason bulguları doğrulandı ve kromozom analizinde normal karyotip saptandı. Gebeliğin ilk trimesterinde saptanan artmış nukal kalınlığa omfaloselin de eşlik etmesi, fetusta Cantrell pentalojisi için erken dikkat edilmesini sağlayabilir.

Anahtar Kelimeler: Cantrell pentalojisi, Artmış nukal kalınlık, Omfalosel, Prenatal tanı

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