

A Case of Trisomy 18 Presenting With Findings of Increased NT and Heart Anomaly on 12 Weeks of Gestation

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Abstract

Objective: To emphasize that major cardiac anomalies can be diagnosed at the end of first trimester by well experienced trained perinatologists.

Cases: Early fetal echocardiography was performed with suspicion of possible cardiac anomaly to a 26 year old lady in 12 weeks of gestation with a history of G1P0. She was referred to our clinic because of increase in the nuchal thickness (83 mm). Cardiac situs and position was normal thus the atrioventricular concordance, however; ventriculoarterial discordance and both great arteries originating from the morphologic right ventricle was clearly demonstrated. CVS for karyotyping was performed after informed consent of the family. The result obtained after 24 days was Trisomy 18. Echocardiographic evaluation at this week revealed cardiac malposition due to cystic adenomatoid malformation and pulmonary stenosis additional to the early findings. Fetus was terminated vaginally by misoprostol induction. Prenatal findings were verified with postmortem autopsy.

Conclusion: Well trained sonographer operating appropriate high frequency ultrasonography can diagnose major cardiac anomalies by the end of first trimester.

Keywords: Congenital heart defect, trisomy 18, nuchal translucency, pregnancy.

12 gebelik haftasında artmış NT ve kalp anomalisi bulguları ile olan trizomi 18 olgusu

Amaç: İlk trimester sonunda majör kalp anomalilerinin fetal tıpla ilgilenen tecrübeli ellerde tespit edilebileceğini göstermek.

Olgu: 26 yaşında G1P0 öyküsü ile, gebeliğin 12+3 haftasında, nukal kalınlıkta artış nedeniyle (83mm) referans edilen olguya kalp anomalisi riski nedeniyle erken fetal ekokardiografi uygulandı. Kalbin situsunun solitus ve pozisyonunun normal olduğu, atrioventriküler bağlantısının konkordant olmakla beraber, ventrikuloarteriel bağlantısının diskordant bağlantı tipinde olduğu gözlemlendi ve her iki büyük arterin morfolojik olarak sağ ventrikülü işaret eden yapıdan orjin aldığı tespit edildi. Ailenin de onayı ile, karyotip tayini için CVS uygulandı. 24 gün sonra elde edilen kültür sonucu; Trizomi 18 idi. Kalpte bu haftada yapılan değerlendirmede, erken dönemdeki bulgulara ek olarak kalpte malpozisyon (kistik adenomatoid malformasyona bağlı) ve pulmoner stenosis izlendi. Fetüs, misoprostol indüksiyonu ile vajinal yoldan termine edildi. Prenatal dönemde elde edilen bulgular, postmortem otopsi ile doğrulandı.

Sonuç: Yüksek frekanslı uygun ultrasonografi cihazı ve tecrübeli operatör ile, ilk trimester sonunda majör kalp anomalilerinin tanısını koyabilmek mümkün olabilmektedir.

Anahtar Sözcükler: Konjenital kalp defekti, trizomi 18, nukal kalınlık, gebelik.

Introduction

Congenital heart defects are the most frequent among major congenital abnormalities and they affect 8/1000 of the newborns.¹ In early pregnancy (first and early second trimester), it is demonstrated that increased nuchal thickness can accompany congenital heart diseases as well as chromosomal abnormalities.² Detailed fetal echocardiography is strongly recommended to patients whose nuchal thickness is increased (≥ 3.5 mm) between the 11th and 14th weeks of pregnancy.^{3,18} Although it is reported that the standard fetal echocardiography time is between the 18th and 22nd weeks of pregnancy, the publications reporting that major cardiac anomalies could be defined in earlier gestational weeks are becoming more frequent.^{4,6,16,17} The systematic and sequential evaluation of the fetal heart provides the diagnosis of major and minor cardiac abnormalities in the prenatal period.⁷ It is demonstrated that the fetal echocardiography performed by experienced hands during the first or early second trimester is efficient in detecting the major cardiac anomalies and also that it has high negative predictive values.⁸ Double outlet right ventricle (DORV) is a conotruncal abnormality in which both the pulmonary artery and the aorta (at least 50%) originate from the right ventricle and is mostly followed by intra and extra-cardiac anomalies with poor prognosis.⁹ DORV is a rare form of congenital diseases with a frequency of 0.033-0.09/1000 among the fetus through newborn. It is a complex abnormality which is caused by the incomplete spiral and inter-ventricular septum formation of the heart. There are four major types; among those, side-by-side relation of the aorta and the pulmonary artery, which is presented in our case, and a subaortic ventricular septal defect (VSD) are observed almost in 50% of the cases.¹⁰ Cardiac anomalies

are frequently accompanied by extra-cardiac anomalies (i.e.; Abdominal wall defects, cleft lip and palate, cystic adenomatoid malformation).¹¹⁻¹³ The purpose of this case report is to evaluate the success of the clinical application of detailed ultrasonography and fetal echocardiography with sufficient experience during the early period of pregnancy by autopsy findings.

Case

In the abdominal ultrasonography, performed by ALOKA 4000 Prosound (Aloka Co., Ltd., Tokyo) 3-7 MHz convex probe, of a 26 year-old patient referred to our clinic with history of G1P0 in the 12 week because of increased nuchal thickness, it is observed that the nuchal thickness was 8.3 mm, there is minimal ascites in thorax, omphalocele, a ventricular septal defect and abnormal view of three vessels (Figures 1, 2, 3). Atrio-ventricular concordance and non-concordant ventriculo-arterial relation is observed in the evaluation made by the sequential segmental analysis of the heart. In addition, a subaortic VSD and an aorta originating from the right ventricle overriding the VSD "side-by-side" the pulmonary artery is also



Figure 1. Artmış nokal kalınlık.

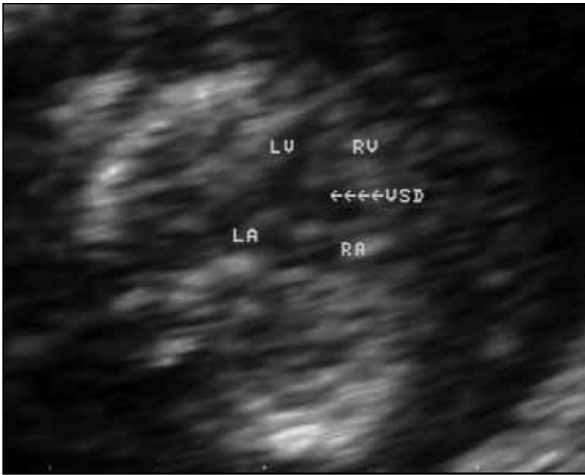


Figure 2. Ventrikuler septal defekt.

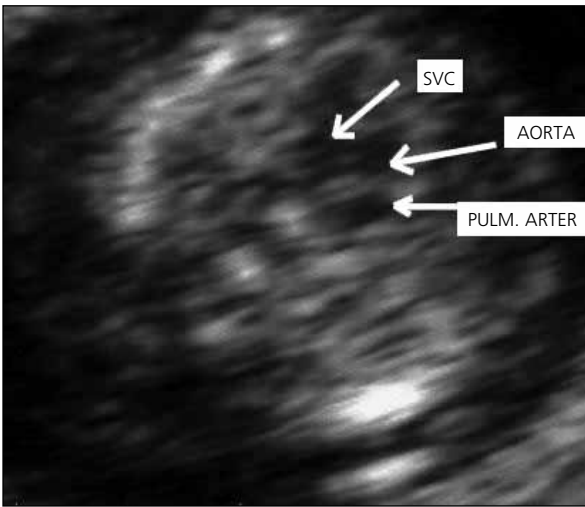


Figure 3. Anormal damar çıkışı.

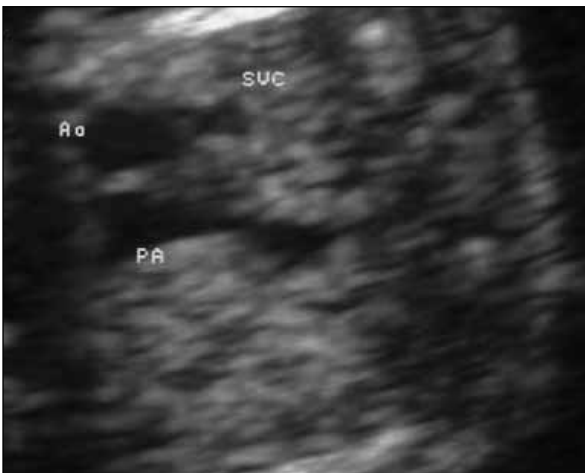


Figure 4. Anormal damar çıkışı.

observed (Figure 6).^{4,7,14} The diagnosis was DORV + VSD. After consultation with the family, it is decided to perform a CVS (Chorionic Villus Sampling) for karyotyping. Following the procedure, Trisomy 18 is defined as a result of long term culturing after 24 days. Besides the previous ultrasonography findings (abnormal three-vessel view, Figure 4), during the ultrasonographic evaluation on the 16th week of the pregnancy, cleft lip-palate in the alveolar fetus, cystic adenomatoid malformation in the lungs (microcystic type) is observed. In the fetal echocardiography, in addition to the first trimester findings, it is seen that the right side of the heart is deviated because of the cystic adenomatoid malformation in the lungs and also a pulmonary stenosis (increased blood flow speed and narrowed pulmonary artery outlet) (Figure 5). Also it is observed that the nuchal thickness seen in the first trimester is resolved. Following the consent of the family, the fetus is terminated vaginally by misoprostol induction. The post-mortem autopsy findings confirmed the findings of the prenatal period.

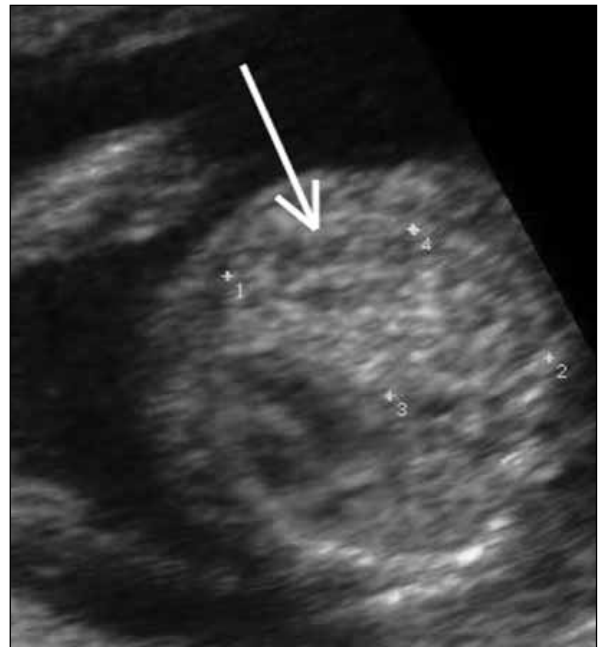
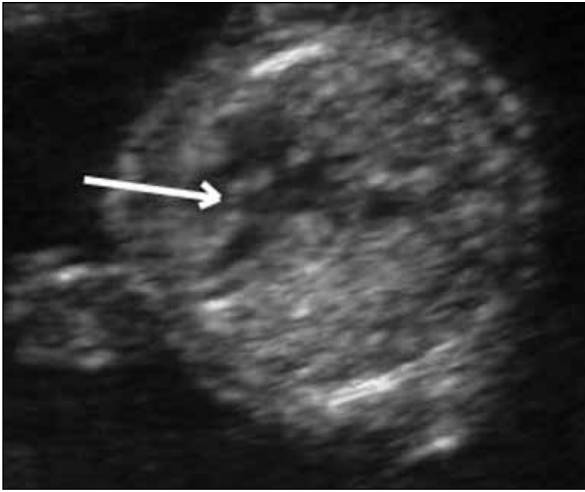


Figure 5. Daralmış pulmoner arter çıkışı.



Resim 6. Fetal kalbin sağına doğru (beyaz okla gösterilen) overriding (kayma) yapmış aorta.

Discussion

Although the fetal echocardiography performed in the mid-trimester (18th-22nd week) is mostly preferred in the diagnosis of the congenital heart diseases, the publications about the diagnosis of the major cardiac anomalies in earlier weeks of the pregnancy (11th-16th week) are becoming more frequent.^{15,16} Haak et al. demonstrated that the possibility of viewing all structures in the heart by the fetal echocardiography between the 12+0 and 12+6 days of the pregnancy is 60% and that rate rises to 92% between the 13+0 and 13+6 days, so that it is possible to make efficient echocardiography in this period for diagnosis.¹⁷ Also, Hyett et al. demonstrated that the possibility of observing cardiac anomaly with increased nuchal thickness during the first trimester and the early second trimester (between the 11th and the 14th gestational weeks) is higher and in cases which the nuchal thickness is ≥ 5.5 mm, the possibility of observing cardiac anomaly is 233/1000.¹⁸ In our case, early fetal echocardiography is performed when the patient is referred on her 12+3 week with increased nuchal thickness. As a result of the evaluation, in addition to the abnormal four-chamber view (VSD), the over-

riding aorta to the right ventricle is observed. Also the three-vessel view was not normal either. The diagnosis was DORV+VSD because the aorta originated morphologically from the right ventricle. It is possible to define many major conotruncal abnormalities by unifying four-chamber and three-vessel + tracheae views through the colored mapping method.¹⁹ Although abnormal three-vessel and tracheae views can not specify the exact diagnosis, it seems it is very helpful in referring the cases to a specialized centre. In our case, during the fetal echocardiography in the early period, a proper three-vessel view was not observed. After a detailed inspection of the outlet vessels, DORV is diagnosed. It is important to remember that this method can misdiagnose abnormalities such as transposition, stenoses in the outlet vessels and VSD. So, for an effective detection, the great arteries should be evaluated through the long and short axes. The key point in diagnosis of DORV is origination of the pulmonary artery and the aorta from the right ventricle. Mostly the view of the long axes of the great arteries will help to diagnose. As in our case, the perpendicular view of the great arteries is not observed in the most common DORV types where aorta (right) and pulmonary artery (left) are situated side-by-side. Although this situation is usually confused with concordant type of transposition, the differential diagnosis is made by defining the origins of the great arteries. The localization of the VSD should be evaluated when the outlet of the great arteries are defined. This is mainly important for the cases that will be operated during the post-natal period. Correct localization is possible by the evaluation of the short axis of the great arteries. Following the evaluation of our case, it is observed that the VSD is subaortic. VSD is almost always present in DORV cases. The most common additional intra-cardiac abnormality is pulmonary stenosis. In our case, this was diag-

nosed on the 16th week of the pregnancy because the pulmonary artery radius was narrow according to the gestational week and also increased blood flow was observed. DORV is a complex heart abnormality which is mostly accompanied by extra-cardiac abnormalities and chromosomal irregularities. Kim et al. detected chromosomal abnormalities in 21%, situs and extra-cardiac abnormalities in 35% of DORV cases.⁹ In our case, the karyotype result was Trisomy 18 and it was accompanied by cystic adenomatoid malformation and alveolar type cleft lip-palate. The extra-cardiac anomalies detected in our case are findings of Trisomy 18 and cardiac anomaly is observed about in 98-99% of this chromosomal abnormality.

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