A Case of Tricuspid Atresia: Prenatal Diagnosis and Postnatal Evaluation

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Abstract

Background: In this case report, we aimed to discuss prenatal diagnosis and postnatal evaluation of tricuspid atresia is a cyanotic form of congenital cardiac defect in which there is not a direct connection between the right atrium and the right ventricle.

Case: A 25-year-old pregnant woman, G2 P1, in 28th gestational week was referred to our clinic with an abnormal four-chamber-view of the fetus, additionally with a suspected VSD. Fetal echocardiography revealed situs solitus, tricuspid atresia and ventriculo-arterial concordance. A large VSD was present between the left ventricle and the rudimentary right ventricle. A decision of cordocentesis was taken by parents after giving genetic counselling. A normal karyotype, 46 XY, was obtained. Later on, parents made up their decision as termination of pregnancy due to the possible surgical complications and difficulties of palliative surgical interventions. Postmortem examination was performed and a normal situated heart, tricuspid atresia, VSD, rudimentary right ventricle, pulmonary stenosis was detected.

Conclusion: The diagnosis of tricuspid atresia is feasible with fetal echocardiography. A cooperative evaluation should be made by an obstetrician, a thoracic-cardiac surgeon and a specialist in genetics and counselling should be also given to the parents by this team.

Keywords: Tricuspid atresia, fetal echocardiography, gebelik.

Triküspid atrezili bir olgu: prenatal tanı ve postnatal değerlendirme

Amaç: Bu olgu sunumunda, konjenital kalp hastalıklarının siyanotik bir formu olan ve sağ atrium, ventrikül arasında doğruda bir bağlantı olmaması sonucu ortaya çıkan triküspid atrezisinin prenatal tanısı ve postnatal değerlendirilmesi tartışılmıştır.

Olgu: Yirmibeş yaşında, G2 P1, 28.gebelik haftasında olan ve fetüsta anormal dört kadran görüntüsüne ek olarak ventrikülo septal defekt (VSD) şüphesi ile sevk edilen hastanın yapılan fetal ekokardiografisinde; situs solitus, sağ atrium ile sağ ventrikül arasında bağlantıyı sağlayan triküspid kapağının yokluğu (tek perfore mitral kapak varlığı) ve konkordant ventrikülo-arterial iletinin olduğu gözlendi. Rudimenter sağ ventrikül ve normal morfolojide olan sol ventrikül arasında büyük bir VSD mevcuttu. Aile genetik danışma sonrası kordosenteze karar verdi. Karyotip 46, XY olarak tespit edildi. Karyotip tayinini takiben, aile postnatal operasyonları ve olası komplikasyonları düşünerek terminasyona karar verdi. Fetüse yapılan postmortem muayenede; normal anatomik pozisyonunda yerleşmiş kalp, triküspid atrezisi, VSD, rudimenter sağ ventrikül ve pulmoner stenoz tespit edildi.

Sonuç: Triküspid atrezinin fetal ekokardiografi ile doğum öncesi tanısı mümkündür. Bu olgular kadın doğum, genetik, göğüs kalp damar cerrahisi gibi alanlardan seçilmiş uzmanlardan oluşan bir ekip tarafından değerlendirilmeli ve aileye danışmanlık da bu ekip tarafından verilmelidir.

Anahtar Sözcükler: Triküspid atrezisi, fetal ekokardiografi, gebelik.

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Background

Cardiac anomalies occur in 0.4 to 1.1% of live neonates, and they comprise the most common anomaly group.[1,2] Tricuspid atresia is a cyanotic form of the congenital heart diseases, and its clinical presentation is a result of the absence of a direct connection between the right atrium and the right ventricle. Its incidence is between 1% and 2.5% among infants with congenital heart diseases.[3,4] Almost all of the cases with tricuspid atresia has ventricular septal defect (VSD), frequently accompanied with rudimentary right ventricle and pulmonary stenosis. Additionaly, 80% of the cases with tricuspid atresia have concordant ventriculoarterial connection, 5 while 20% have disconcordant connection (transposition).[6]

Prognosis is worse in the infants born with tricuspid atresia if it is not managed palliatively. Usually the infants undergo 2 to 3 procedures (Glen and Fontan operations) within the first six years following the labor. The mean survival rate is 50% at the end of 15 years following the procedure.[7]

Case

A twentyfive-year-old pregnant woman, G2 P1, at gestational week 28 who was monitored for a suspected VSD in addition to the abnormal four-chamber-view of the fetus was referred to the Obstetrics and Gynecology Department of the Medical Faculty of Kahramanmaraş Sütçü İmam University for fetal echocardiography. In a sequential analysis of the heart,[5] presence of situs solitus, lack of formation of the tricuspid valve connecting the right atrium with the right ventricle (presence of a single perforated mitral valve) and concordant ventriculo-arterial connection were observed. A large VSD was present between the rudimentary right ventricle and left ventricle, which was in normal morphology (Figure 1). No anomaly other than cardiac was observed during the systemic examination by ultrasonography. The parents made their choice for cordosynthesis upon genetic consultation. Cariotype was found 46, XY. Following that, the parents decided for the termination of pregnancy considering the postnatal operations and potential complications. A postmortem examinati-

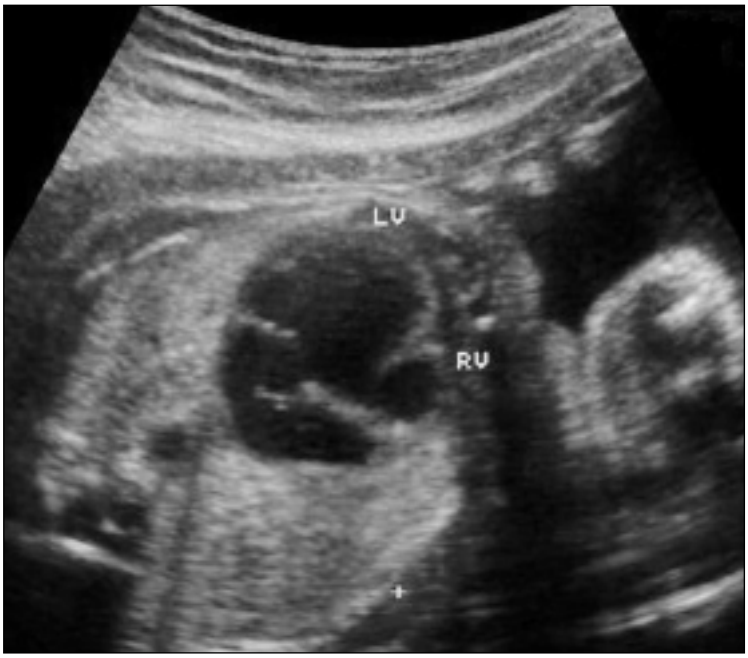

Figure 1. View of tricuspid atresia, rudimentary right ventricle, ventricular septal defect and abnormal four-chamber.

on on fetus revealed a normal anatomic position of the heart, tricuspid atresia, VSD, rudimentary right ventricle and pulmonary stenosis.

Discussion

Diagnosis of tricuspid valve anomalies is made by a standard four-chamber-view, however fetal echocardiography is required for final diagnosis. Tricuspid atresia is a cardiac anomaly which is a rare condition among other cardiac anomalies, and doesn't provide promising results in spite of the procedures where the heart is operated into a single ventricular structure anatomically during the postnatal period.[3,4,7] Contrary to other cardiac anomalies, it is very rare that tricuspid atresia is accompanied with chromosomal disorders and multiple malformation syndromes, but chromosomal evaluation should also be undertaken.[8] Incidence of concomittant non-cardiac malformations is around 10%.

No additional anomaly was observed during the prenatal ultrasonographic and postnatal evaluations in our case. In prenatal diagnosis, advanced ultrasonography (particularly facial, renal and gastrointestinal systems), fetal cariotype, family history of cardiac arrhythmia, history of congenital cardiac anomaly and history of lithium intake during pregnancy should be evaluated.[8] As a result of these evaluations, no etiology was found in our case to understand the cardiac anomaly. In differential diagnosis, pulmonary atresia accompanied with intact septum should be evaluated. Monitoring of the tricupsid regurgitation is helpful in differential diagnosis in such cases. Another diagnosis that should be considered is Double-Inlet ventricle. The family should be informed about the risk for a recurrency rate of appr. 1% in subsequent pregnancies during the genetic consultation.[8]

Conclusion

Prenatal diagnosis of tricuspid atresia can be made by fetal echocardiography. Such cases should be evaluated by a team of experts from fields such as obstetrics, genetic, and cardiovascular surgery, and counselling should be provided by that team.

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