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# Truncus Arteriosus Type 1 With Prenatal Diagnosis: A Case Report

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**Abstract**

**Objective:** Truncus arteriosus complicates approximately 0.01 of 1000 live births. In this case report we discussed truncus arteriosus Type 1 case detected in utero with echocardiographic findings.

**Case:** A 31 year old pregnant woman at 26 weeks of gestation referred to our clinic. She hasn't got a consenginous history.According to the fetal echocardiographic findings we diagnosed truncus arteriosus Type 1.

**Conclusion:** The big part of the congenital heart diseaseses occurs in pregnancies with no risc factors. If we visualized the subaortic ventricular septal defect in the fetal heart, we can detected the associated abnormalities; such as truncus arteriosus.

**Keywords:** Truncus arteriosus, congenital heart anomalies, fetal echocardiography.

### *Prenatal tanı alan trunkus arteriozus tip 1: olgu sunumu*

**Amaç:** İkinci trimesterdeki taramada tanısı konan, nadir görülen konjenital kalp hastalıklarından biri olan truncus arteriyozus olgusu sunulmuştur.

**Olgu:** 31 yaşında, gebeliğin 26. haftasında kliniğimize başvuran, akraba evliliği hikayesi olmayan hastanın yapılan fetal ekokardiografisinde Tip 1 truncus arteriyozus tespit edildi.

**Sonuç:** Konjenital kalp hastalıklarının çok büyük bir kısmı risk faktörü içermeyen gebeliklerde meydana gelmektedir. Detaylı bir muayene ile subaortik ventriküler defektin izlenmesi altta yatan truncus arteriyozusun yakalanmasına yardımcı olacaktır.

**Anahtar Sözcükler:** Trunkus arteriyozus, konjenital kalp anomalileri, fetal ekokardiyografi.

## Introduction

The incidence rate of congenital cardiac disease is 4-11/1000 live birth and this rate constitutes the most frequent congenital anomaly group among cardiac diseases. The most frequent cause of early neonatal deaths associated with congenital anomaly is cardiac diseases.[1,2] truncus arteriosus is characterized as single great artery coming out of heart and its incidence rate is 0.01 among 1000 live births.[3] Truncus feeds systemic, coronary and pulmonary circulations. Echocardiographic finding is diagnosed by detecting also pulmonary arteries coming out from a single great artery wider than aorta and overlapping on ventricles.[3,4]

Like in other conotruncal anomalies, truncus arteriosus cannot perform in utero cardiac decompensation; however, decompensation may occur in first days of life. The surgical fixation is quite complex since it is needed to turn dysplastic truncal valve into aorta and to make

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a connection from right ventricle to pulmonary arteries. Even after a successful surgery, 10 years of survival rate is less than 80%.[3]

A major part of congenital cardiac anomalies occurs in pregnancies which do not include risk factors.[5,6] In our article, the case of truncus arteriosus Type 1 with prenatal diagnosis was discussed in accordance with the literature.

## Cases

31 years-old G2 P1 pregnant without any consanguineous history admitted to our clinic at her 26th gestational week. No systemic disease was found in her anamnesis. Nothing was found in her family history except Type 2 Diabetes Mellitus of her mother. In the examination performed on her 12th gestational week, her nuchal transparency was measured as 2.5 mm. However, biochemical marker data of 1st and 2nd trimesters could not be found. In the detailed systemic examination performed by ultrasonography, no anomaly was observed except subaortic ventricular septal defect. Fetal growth was normal. Amniotic fluid volume was found normal. No notching was detected on bilateral uterine arteries. In the fetal echocardiography of the patient, heart was observed within left hemithorax and cardiac apex was observed as staying leftward. Atrial situs solitus was in existence. Heart chambers were in normal width and global heart contractions were detected as good. Atrioventricular concordance was full. Mitral tricuspid valve was normal. Intraventricualr septum developed; however, there was wide ventricular septal defect at outlet localization. A single great vessel (truncal vessel) output was observed from ventricles.

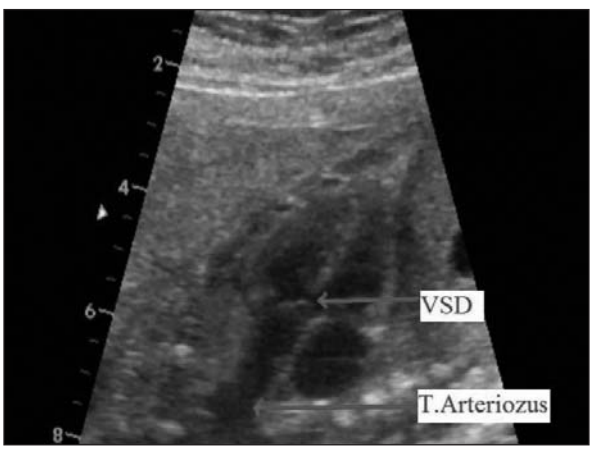


**Figure 1.** Truncus arteriosus and ventricular septal defect.

Though truncal vessel was slightly "overriding" the ventricular septal defect, it was coming out of right ventricular mainly and 80% of it was dextropositioned. Truncal vessel flow rate was found as 175 cm/sec. The pulmonary artery was just coming out of the distal of truncal valve as a single root. The pulmonary was separated into right and left branches and no stenosis was found on artery output. Truncal arcus was on rightward position and major vessels were observed, aorta coarctation or interruption. The finding was diagnosed as truncus arteriosus Type 1 according to the diagnoses mentioned above.

Genetic consultation was given to the family and karyotype analysis was suggested. Fetal blood sampling was performed to the patient in the same session. In the karyotype analysis performed on fetal blood, no numerical and structural anomalies were observed and chromosome formation was found as normal. In the examination performed by fluorescence in situ hybridization (FISH) on fetal blood, 22q11 micro-deletion was not found. Genetic consultation was given to the family about prognosis. The family stated that they wanted to maintain the pregnancy. The patient was called for control one week later. When the patient admitted to our clinic due to the reducing baby movements five days later, no fetal cardiac motions was detected. It was accepted as stillbirth and the patient was hospitalized in our department for delivery. Following the delivery induction, 1100 gr girl fetus was labored. In the postmortem examination, it was observed that pulmonary artery and aorta came out of right ventricle as a single root and thoracic aorta was descending rightwards on the high ventricular

septal defect (VSD) surface. No ductus arteriosus (Botalli) was observed. Prenatal diagnosis was confirmed.

## Discussion

Prenatal diagnosis of congenital cardiac diseases is very significant since they create the most frequent anomaly at live births.[7,8] While prenatal evaluation of all other organ systems of fetus was improved rapidly through high standards and prevalent practices, it is hard to say the same for prenatal ultrasonographic evaluation of fetal heart despite the efforts which had began much earlier. In the study of Jaeggi et al., only 15% of all heart anomalies could be recognized at prenatal period.[9] While this rate was 30% in anomalies which could be recognized by cross-section of four chambers, prenatal diagnosis rate was only 6.7% in conotruncal anomalies associated with major vessel outputs.[9] The detection rate of heart anomalies was found as 26% in the study of Tegnander et al. by imaging four chamber in fetal heart anomaly scanning.[10] Bromler et al. showed that this rate could increase to 83% by including major vessels into the evaluation in addition to four chamber imaging.[11] Including major vessels into evaluation in fetal echocardiography does not only increase to intrauterine detection rate of congenital cardiac diseases but also provides to diagnose properly and effective intervention at postnatal period by consecutive examination technique.[5,10,11]

According to the classical classification, truncus arteriosus cases are gathered under three major groups. At truncus arteriosus Type 1, the major pulmonary comes out of the location superior to artery truncal valve, the left posterolateral truncus and branches off to right and left pulmonary artery branches. In Type 2, right and left pulmonary artery branches (without the major pulmonary artery) come out of the same location as in Type 1 but separately. In Type 3, the outlet location is upper and separate from lateral. In this type, coronary artery anomalies are often.[12]

In our case, major pulmonary artery came out of the location superior to artery truncal valve and then branched off into right and left pulmonary artery branches. According to this definition, our case was found compatible with truncus arteriosus Type 1.

There are aortic arches located in the right side in 15-30% of truncus arteriosus cases.[13] Aortic arch was also located in the right side in our case.

It is known that there is ductus arteriosus agenesis in 50-75% of cases with truncus arteriosus.[14,15] In our case, ductus arteriosus which could not be detected in fetal echocardiography was also not found in post-mortem examination.

It may be difficult to distinguish truncus arteriosus from Fallot tetralogy with pulmonary atresia in utero.[3] Final diagnosis is established by observing pulmonary artery leaving truncus. In pulmonary atresia cases with VSD, only aorta is followed instead of truncus and pulmonary artery is not followed.

There is also a relationship between congenital cardiac anomalies and structural chromosome anomalies. It is known that 22% of cardiac anomalies in fetus were seen together with aneuploid.[16] The most frequent karyotype anomalies are Trisomy 18, 13, 21 and Monosomy X.[16] On the other hand, it was found in a study performed on 11-14 weeks old fetuses that 72.9% of fetuses with fetal achocardiography abnormality had karyotype anomalies.[17]

Beside major structural chromosome anomalies, it was found that conotruncal anomalies progressed together with 22q11 micro-deletion (Di George syndrome; it is together with thymic aplasia, hypocalcemia, abnormal face shape and mental retardation). 22q11 micro-deletion was found in 35% of cases with truncus arteriosus.[12] In the examination performed on our case by FISH, no deletion was found and the karyotype analysis was reported as normal.

An opportunity will be obtained by fetal echocardiography for giving a full briefing to parents about prognosis and risks at 18th-22nd

gestational week and parent will be able to benefit the opportunity of termination on time in cases with severely poor prognosis. An overwhelming majority of congenital cardiac anomalies, as seen in our case, is formed of pregnancies not including risk factor. In truncus arteriosus cases, generally the view of normal four chambers is observed. In case of observing subaortic VSD in detailed examination during examining major vessels, truncus arteriosus diagnosis can be established. In fetuses with congenital cardiac diseases, performing fetal karyotype analysis and FISH will allow prenatal diagnosis of syndromes displaying association. In cases which are desired to continue pregnancy, genetic consultancy should be given for chromosome anomaly that can be followed with 14% frequency and for 22q11 deletion and Di George syndrome that can be followed with 35% frequency. As seen in our case, congenital cardiac disease may also be responsible for fetal death cases. Performing postmortem examination will provide to establish the most proper diagnosis in stillborn fetuses that are not diagnosed as antenatal congenital cardiac disease.

## Conclusion

An overwhelming majority of congenital cardiac diseases occur in pregnancies which do not have risk factors. By a detailed examination, observing sub-aortic ventricular defect would help to detect truncus arteriosus. Performing postmortem examination will provide to establish the most proper diagnosis in fetal death cases that are not diagnosed as antenatal congenital cardiac disease.

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