Case Report

# Prenatally diagnosed case of tricuspid valve dysplasia: A case report with review of the literature

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## Abstract

We present a case of fetal tricuspid valve dysplasia (TVD) and pulmonary atresia, diagnosed during a routine obstetric ultrasound scan. Serial fetal echocardiographic evaluations revealed progressively augmented prodigious thickening of the tricuspid valvular and subvalvular structures, which eventually extensively obliterated the right ventricle cavity. Thickened dysplastic valve displayed a "cotton-wool" appearance. Unusual configurations of three vessels in the three-vessel view were also observed on a consecutive gray scale and color Doppler scans. During pregnancy, the fetus exhibited satisfactory growth parameters, and complications of progressive hemodynamic compromise associated with TVD and pulmonary atresia such as grievous hydrops or arrhythmia did not develop till 39 weeks of gravidity.

**Key words:** Dysplasia; non-immune hydrops; pulmonary atresia; tricuspid annulus; tricuspid regurgitation; tricuspid valve

## Introduction

Tricuspid valve dysplasia (TVD) is a rare cardiac malformation, infrequently missed on prenatal sonography because of characteristic echocardiographic findings such as thickened but normally inserted tricuspid valve (TV), tricuspid regurgitation, and asymmetric cardiomegaly.[1,2] The anomaly has high perinatal mortality due to the hostile hemodynamic status which is primarily related to large TV annulus, and inadequate trans-atrial blood flow. We describe a case of TVD, in which massive thickening of the TV and atypical configuration of the three vessels in the three-vessel view were observed. A review of the literature is also provided to enhance the understanding of this rarely reported cardiac anomaly. This case is notable because unusual findings were observed during serial fetal echocardiographic examination.

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## Case Report

A routine fetal sonographic scan of a 26-year-old third gravida showed a singleton fetus of 25 weeks' gestation. A cardiac evaluation revealed significant tricuspid regurgitation and asymmetric cardiomegaly due to the enlarged right atrium [Figure 1]. The TV was irregular, thickened, and echogenic although normally inserted [Figure 1]. The foramen ovale was enlarged with the right to left blood



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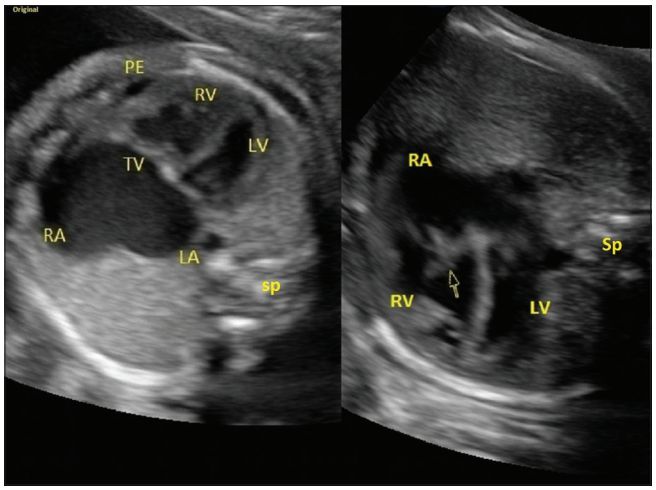


**Figure 1:** Four-chamber view at 25 weeks of pregnancy: Thickened normally inserted tricuspid valve (TV) with enlarged right atrium (RA). Thickened subvalvular structures (arrow: right panel). LA: Left atrium, RV: Right ventricle, LV: Left ventricle, PE: Pericardial effusion, Sp: Spine

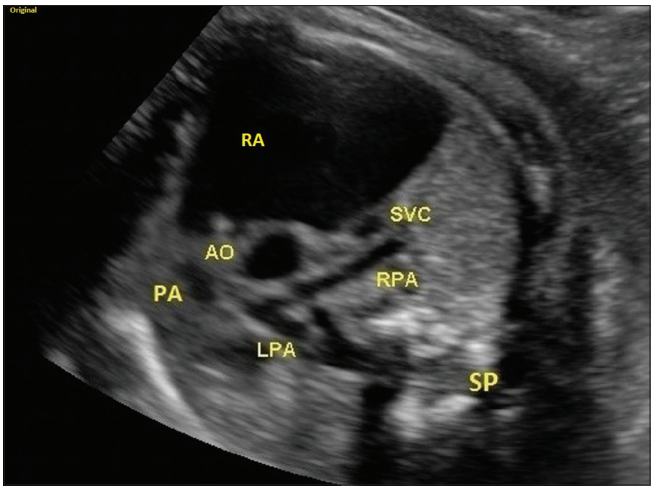


**Figure 2:** Three-vessel view: showing normal arrangement of three vessels with very small pulmonary artery (PA) and its branches. RA: Enlarged right atrium, AO: Ascending aorta, SVC: Superior vena cava, LPA: Left pulmonary artery, RPA: Right pulmonary artery

flow across it. Pulmonary artery and its two branches were very small in diameter [Figure 2]. Pulmonary valve and ductus arteriosus were not visualized in a gray scale scan. Pulsed and color Doppler examination manifested no antegrade or retrograde flow in the pulmonary artery as well as in the ductus arteriosus. The cardiothoracic ratio was 0.51 with left-axis deviation of about 90°.There was minimal pericardial effusion. Based on sonographic findings, a diagnosis of TVD with pulmonary atresia was made and prognosis of the condition was explained to the couple.

On follow-up scan at 29 weeks, pulmonary artery and its branches, as well as the ductus arteriosus, were not identified. In the three-vessel view, only two vessels aorta and superior vena cava were visualized [Figure 3]. Aorta was malpositioned and showed "I" sign [Figure 3]. Successive echocardiographic evaluations manifested markedly thickened TV without any notable dilation of the tricuspid orifice [Figures 4 and 5]. Pulmonary regurgitation and reverse flow in the ductus arteriosus were observed at 32 weeks of gestation [Figure 6]. Although the regurgitation aggravated with advancing gestation [Figure 6], no complications related to the reversed blood flow developed till term. The family did not return for a follow-up, but informed that the neonate survived for four days after a normal vaginal delivery. Although postnatal confirmation was not possible, the prenatal diagnosis of TVD was obvious.

## Discussion

Congenital causes of TV deformations include Ebstein's anomaly (EA), unguarded tricuspid orifice, Uhl's anomaly, and TVD.[3,4] All these anomalies may expedite to the same hemodynamic burden, such as tricuspid regurgitation and its pathophysiological sequelae.[5] In fetuses, severe regurgitation may accelerate cardiomegaly, secondary lung hypoplasia, non-immune hydrops, and cardiac arrhythmia,[6] leading to high perinatal mortality.[2,6,7]

EA is characterized by the defect in TV delamination with various degrees of downward displacement of septal leaflets of the dysplastic valve and atrialization of the proximal right ventricle.[5,8,9] In unguarded tricuspid orifice, the septal cusps, chordae tendineae, and papillary muscles are absent,[3] causing low-velocity to-and-fro flow of blood across the markedly dilated TV annulus with identical systolic and diastolic velocities.[3,7] Uhl's anomaly of the right ventricle is characterized by a partial or complete absence of the myocardium of the right ventricle and is usually associated with either dysplastic TV[10,11] or may coexist with the unguarded tricuspid orifice.[12] TVD is a primary defect in TV leaflets, chordae tendineae, and papillary muscles due to myxoid degeneration,[7] resulting in thickened and deformed valve. The damaged valve does not close properly during ventricular systole leading to tricuspid insufficiency with a variable degree of regurgitation.[7,13]

Prenatally, TVD is diagnosed when normally delaminated, echogenic,[1] thickened, and redundant[14] TV is associated with tricuspid regurgitation and right atrial enlargement.[6] Typically, the systolic regurgitant jet is seen originating at the level of TV orifice,[7] differentiating TVD from other anomalies.

In this case, progressively increased thickening of valvular and subvalvular structures of TV was observed, which eventually extensively obliterated the right ventricle cavity. However, the TV annulus did not enlarge notably with advancing gestation and no dreadful hydrops or arrhythmia developed till term. Rapid and progressively worsening hemodynamic compromise is usually associated

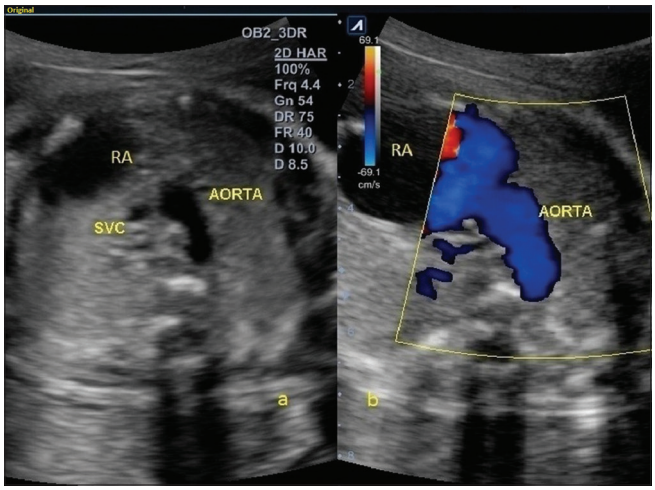


**Figure 3:** Gray scale and color Doppler three-vessel view at 29 weeks of pregnancy. Only two vessels aorta and superior vena cava (SVC) are visualized. Unorthodox malpositioned "I" shaped aorta. Ductus arteriosus and pulmonary artery are not seen. RA: Right atrium

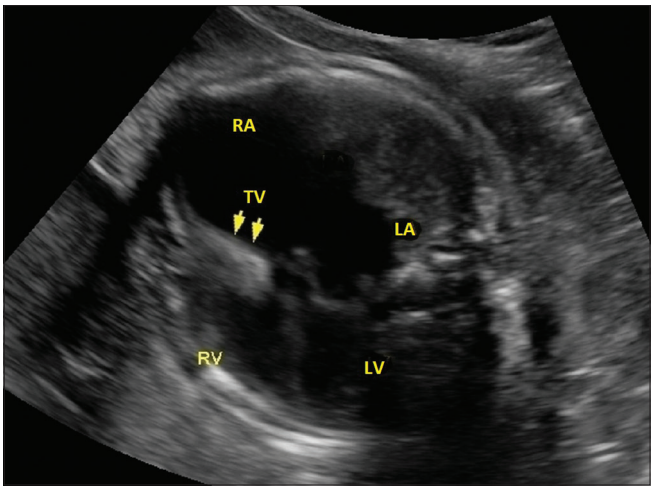


**Figure 4:** Dysplastic tricuspid valve (TV) at 32 weeks. RA: Right atrium, RV: Right ventricle, LA: Left atrium, LV: Left ventricle

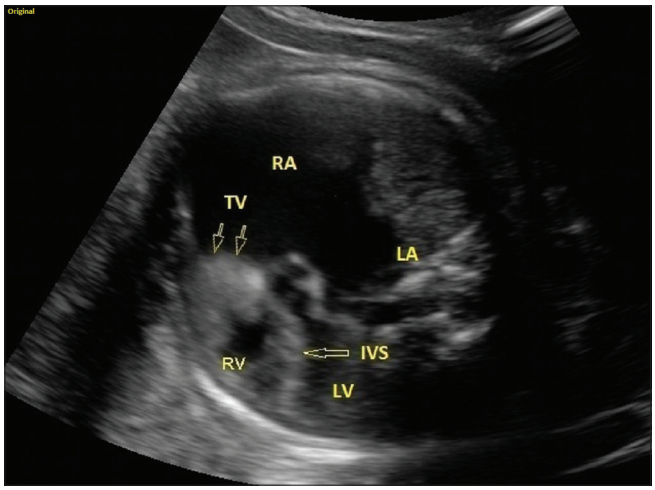


**Figure 5:** Thickened dysplastic tricuspid valve (TV) at 39 weeks: "Cotton wool appearance". RA: Enlarged right atrium, IVS: Interventricular septum, RV: Obliterated right ventricle, LA: Left atrium, LV: Left ventricle

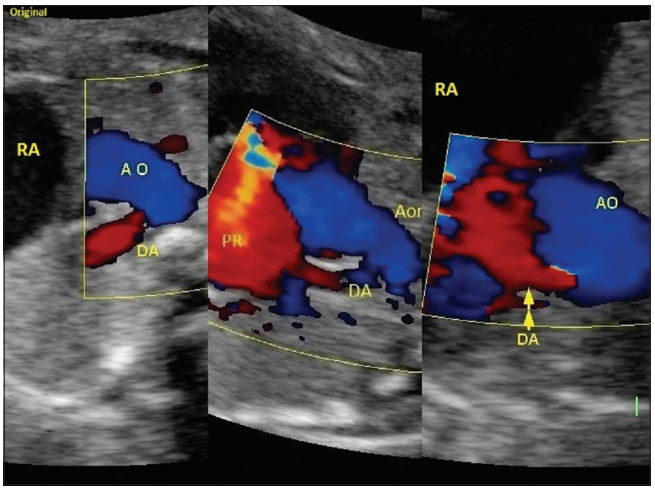


**Figure 6:** Color Doppler three-vessel views at 32 weeks (left panel), 35 weeks (middle panel), and 39 weeks (right panel) showing pulmonary regurgitation (PR) and retrograde flow in the ductus arteriosus (DA). Note the changing angle between the aorta (AO) and the DA with advancing gestation and worsening pulmonary regurgitation. RA: Right atrium. PA: Pulmonary artery

with a large annulus,[15] which causes severe tricuspid regurgitation, functional atresia of the pulmonary valve, retrograde flow in the ductus arteriosus, and pulmonary regurgitation.[15] The regurgitation completes circular shunt,[1,6] causing short left ventricular ejection time and increased myocardial performance index (MPI),[14] culminating in low organ perfusion and fetal distress. A larger TV annulus Z-score >5.6 predicts poor outcome and is associated with the progressive disease among initially low-risk fetuses.[15] Survival of the fetus is also related to adequate trans-atrial communication, which plays a determinant role in the potential increase in volume load on the left heart and prevents major circulatory insufficiency.[16] The diameter of the foramen ovale, which is a marker of trans-atrial blood flow, was adequate in this case. Foramen ovale/atrial septal length ratio <0.36 is associated with the poor end result.[17]

TVD frequently coexists with functional or organic intrauterine critical right ventricle outflow obstruction and has been observed prenatally and postnatally in a large percentage of cases.[7,18,19] The functional atresia occurs because the right ventricle pressure is not enough to overcome the pulmonary artery pressure owing to tricuspid insufficiency and regurgitation. In this form of obstruction, the pulmonary valve is present but remains immobile, the pulmonary artery is of normal caliber with forwarding flow through it.[7,19,20] In organic obstruction, pulmonary valve is atretic, the pulmonary artery is small in size, and there is no detectable forward flow in the vessel.[7,19,20] Although there are many findings suggesting functional or organic atresia in prenatal echocardiography, it is difficult to distinguish these two forms before birth.[7,19,21] This is due to the fact that failure to identify the main pulmonary artery

or valve on a single examination is not specific to organic obstruction.[21] Similarly, a normal caliber main pulmonary artery does not exclude the diagnosis of the atresia then as well.[19] However, detection of pulmonary regurgitation,[21] reverse flow in the ductus arteriosus [Figure 6], and a ductus-dependent pulmonary circulation is a strong indication of the presence of severe pulmonary stenosis or atresia.[22] Prenatal diagnosis of pulmonary atresia is of particular importance because the affected neonate may require urgent administration of prostaglandins.[21] It also warrants avoidance of maternal administration of drugs which may cause constrictive effects on the ductus arteriosus.[22]

Tricuspid insufficiency and regurgitation may also occur in a normal TV when the pulmonary atresia is associated with an intact ventricular septum,[21,22] or as a consequence of an intrauterine insult causing right ventricular dysfunction or papillary muscle ischemia.[23]

Although the pulmonary artery and its two branches were very small in diameter in the three-vessel view at 25 weeks of gestation, the spatial arrangement of the three vessels was normal. However, in subsequent scans, only two vessels aorta and superior vena cava were visualized and the right ventricular outflow tract was not demonstrable. The ascending aorta was malpositioned and showed "I" sign which is characteristically seen in anomalies like transposition of great arteries. Non-visualization of the pulmonary artery and its branches is by virtue of pulmonary atresia where the arteries are so small to identified in gray scale ultrasound. This results in only two vessels to be seen in the three-vessel view. Small pulmonary arteries are hemodynamically disadvantageous and predict poor lung viability in the presence of small lung volumes.[24] However, such arteries are now not considered as a contraindication for Fontan circulation.[25]

The unusual configuration of three vessels in the three-vessel view, and non-visualization of ductus arteriosus may be attributed to worsening cardiomegaly, pulmonary atresia, and also to the dynamic nature of fetal development.[7] The ductus arteriorsus becomes invisible due to an acute inferior angle between the arterial duct and aorta.[23] The appearance of the duct and angle at which it joins the aorta [Figure 6] may provide clues about the timing of the development of pulmonary atresia and concomitant retrograde ductal flow.[26]

In most fetuses diagnosed as TVD, a progressive hemodynamic compromise was observed and favorable hemodynamic status in the second trimester did not ensure a favorable status in the third trimester.[15] For such cases, many fetal risk factors for adverse hemodynamic condition and perinatal mortality have been reported. Independent predictors of mortality at the time of diagnosis are: TV annulus diameter Z-score >5.6,[15] presence of pulmonary regurgitation on the first echocardiogram,[27] absence of antegrade flow across the pulmonary valve,[15] MPI,[28] cardiothoracic area ratio >0.48,[15] and pericardial effusion.[19] The presence of pulmonary regurgitation, in particular, signifies circular shunt physiology, which often culminates in mortality.[6] Left ventricle MPI is also a good predictor of the outcome because it represents global myocardial performance.[28] Higher values of MPI reflect worse function and poor outcome.[29]

The outcome of the discommodious hemodynamic status related to TV disease is extremely poor, intrauterine fetal death is frequently observed, and the postnatal mortality rate among patients with prenatal diagnosis is very high.[5,9] The poor end result is because of long-standing lung compression, which causes secondary lung hypoplasia and non-immune hydrops.[19,24] Volumetric assessment of fetal lung hypoplasia can be done using ultrasonography[30] and MRI.[29,31,32] Knowledge of the relative fetal lung volume assists in the confirmation and quantification of fetal pulmonary hypoplasia and may help in planning a reconstructive surgery such as Fontan procedure.

Two-dimensional echocardiography is the primary technique used to diagnose congenital heart diseases and associated complications. Fetal MRI, with novel computational techniques,[33] has the potential to increase the prenatal detection rates of these cardiac aberrations. Presently, MRI is used to detect vascular disorders such as conotruncal anomalies but its use to detect intracardiac malformations is unclear.[34] Nevertheless, the modality is valuable in prenatal diagnosis of associated extracardiac complications and/or anomalies which may have serious implications on the fetal or neonatal survival.

Precise diagnosis of congenital anomalies helps in adequate parental counseling and the planning of perinatal management. In recent years, the outcome of TVD has improved due to better detection of less severe pathology and advances in the surgical management of severe disease.[35]

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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### Conflicts of interest

There are no conflicts of interest.

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