

Prenatal diagnoses of an uncommon isolated obstructed supracardiac total anomalous pulmonary venous connection

Case report and review of the literature (CARE compliant)

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Abstract

Introduction: Total anomalous pulmonary venous connection is an uncommon congenital heart disease. Four types are described based on the site of pulmonary venous drainage: supracardiac, cardiac, infradiaphragmatic, and mixed connection. In most cases of supracardiac type, the common venous confluence drains through an ascending left vertical vein into the brachiocephalic vein, right superior vena cava, and then into the right atrium. Anomalous connection of the pulmonary venous confluence directly to the right SVC, especially the obstructed form is an unusual and severe supracardiac variant. The prenatal diagnosis is challenging.

Patient concerns: We present a case report of a fetus diagnosed with TAPVC at 23 gestational weeks.

Diagnosis interventions: The 4-chamber view showed a small left atrium, with a “smooth” posterior wall and the absence of pulmonary vein connection. This is the first case of prenatally diagnosed isolated, obstructed supracardiac type with drainage directly into the right superior vena cava.

Conclusion: The obstetrician and fetal cardiologist should be cautious at the direct and indirect echocardiographic signs of this condition. A prenatal diagnose of isolated, obstructed form is important for adequate planning of delivery and postnatal surgery in a tertiary center.

Abbreviations: 2D = bi-dimensional, 3VTV = 3 vessel view, 4CV = 4 chamber view, AngioCT = computer tomography angiography, Ao = aorta, Ao arch = aortic arch, BCV = brachiocephalic vein, CC = common venous confluence, Duct art. = ductus arteriosus, IVC = inferior vena cava, LA = left atrium, PA = pulmonary artery, RA = right atrium, SVC = superior vena cava, TAPVC = total anomalous pulmonary venous connection, v = pulmonary veins.

Keywords: fetal, isolated, obstructed, supracardiac, total anomalous pulmonary venous connection

1. Introduction

Total anomalous pulmonary venous connection (TAPVC) is an uncommon congenital heart disease, with an incidence of about 0.6 to 1.2/10.000 live birth and accounting for about 0.7 to 1.5% of all cardiac defects.^[1] TAPVC is a result of the nonfusion of the confluence of the pulmonary venous drainage and the left

atrium.^[2] It is often associated with complex cardiac lesions. The prenatal diagnosis of isolated form of TAPVC is challenging.

Darling described 4 types of TAPVC based on the site of pulmonary venous drainage: supracardiac (type I), cardiac (type II), infradiaphragmatic (type III), and mixed connection (type IV). In most cases of supracardiac TAPVC, there is an anomalous connection of the common venous confluence with a persistent left superior vena cava (SVC), also named ascending left vertical vein, which further drains into the brachiocephalic vein, right SVC, and then into the right atrium.^[3] The connection of the pulmonary venous confluence directly into the right SVC is an uncommon condition. The symptoms of TAPVC vary from mild cyanosis to severe cyanosis, respiratory distress, and acidosis even in the first hours of life.^[4] These symptoms depend mostly on the degree of venous obstruction; therefore, the symptoms will increase in severity at the same time with the degree of venous obstruction.^[4] The supracardiac type of TAPVC will present pulmonary venous obstruction less frequently than the infracardiac type.^[4] The obstructive forms of TAPVC present more rapidly respiratory disorders, hypoxia, pulmonary hypertension, even cardio-respiratory failure, and distress-syndrome-like pattern.^[5] The 2-dimensional echocardiography is the one that usually establishes the diagnosis of this rare cardiac congenital condition. Obstructive TAPVC is a congenital condition that requires emergency surgical intervention. Other therapeutic measures, such as atrial septostomy, can be performed by cardiac catheterization.^[4] The surgical treatment consists in

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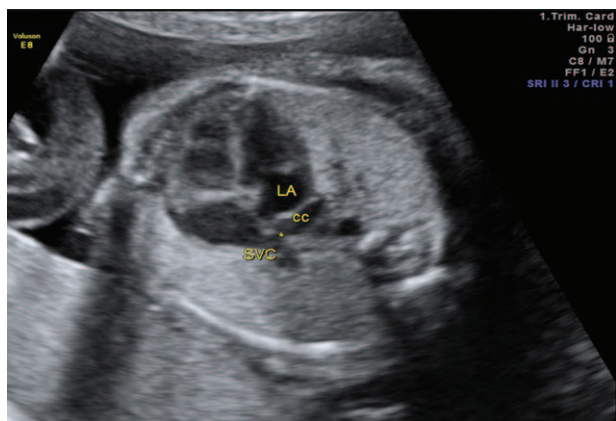


Figure 1. Prenatal echocardiography. 4C view. Small left atrium with a “smooth” posterior wall. A common venous confluence behind the left atrium. 4C view = 4 chamber view, CC = common venous confluence, LA = left atrium, SVC = superior vena cava.



Figure 3. Prenatal echocardiography – The high mediastinal view. Dilated superior vena cava due to drainage of the common venous confluence. BCV = brachiocephalic vein, SVC = superior vena cava.

ligature of the vertical vein in order to prevent the residual left-to-right shunt.^[5] Heritable cases of TAPVC have also been described in the literature; therefore, the physician must always take into consideration the referral of the family for genetic assessment and advice.^[6]

We hereby present an intrauterine diagnosed case of isolated, obstructed supracardiac TAPVC.

The informed consent was given by the patient’s parents (legal guardians) for publication of this case report.

2. Case report

2.1. Prenatal diagnostic assessment

A 26-year-old primigravid woman, without relevant history, was referred for fetal echocardiography at 26 weeks of gestation, after the second trimester fetal echocardiography screening revealed TAPVC in the fetus. Cardiac examination was carried out by a pediatric cardiologist and a gynecologist with experience in fetal echocardiography. Echocardiography was performed transabdominally. The 4-chamber view revealed a right-to-left chamber

discrepancy, a small left atrium, with a “smooth” posterior wall. Also, the 2D image raised the suspicion of absent pulmonary vein connection (Fig. 1). Low-flow color mapping identified pulmonary veins draining into a common confluence behind the small left atrium. Three vessel-trachea (3VT) view revealed a dilated right SVC, with normal innominate vein. No vertical vein was detected on 3 vessel (3V) or 3VT view (Fig. 2). Low-flow color Doppler imaging revealed a turbulent flow in right SVC at the drainage site of the common confluence (Fig. 3). On further assessment, a very narrow opening of the confluence into the right SVC was detected (Fig. 4). No other cardiac anomaly was detected.

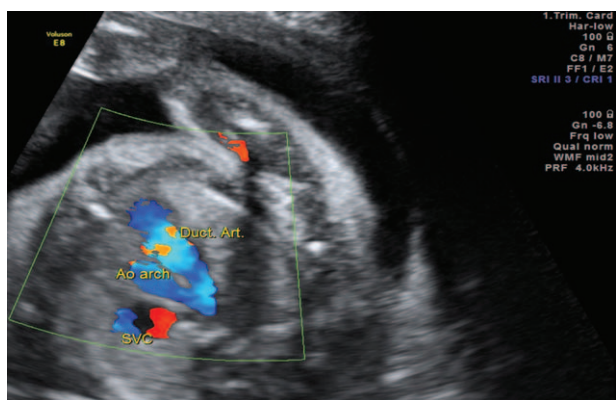


Figure 2. Prenatal echocardiography. A 3 vessel trachea view (3VT) with color Doppler flow mapping. Dilated superior vena cava (SVC) with turbulent flow. 3VTV = 3 vessel trachea view, Ao arch = aortic arch, Duct art. = ductus arteriosus, SVC = superior vena cava.

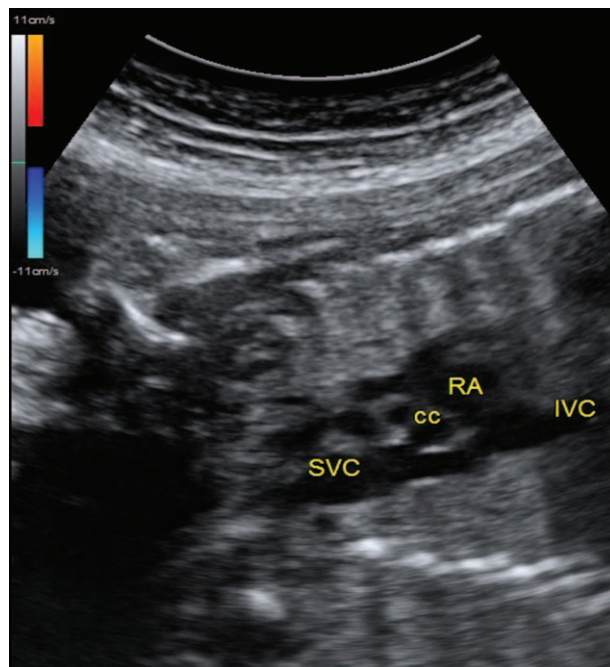


Figure 4. Prenatal echocardiography. Oblique bicav view. Dilated SVC due to the drainage of the common venous confluence through a narrow opening. CC = common venous confluence, IVC = inferior vena cava, RA = right atrium, SVC = superior vena cava.

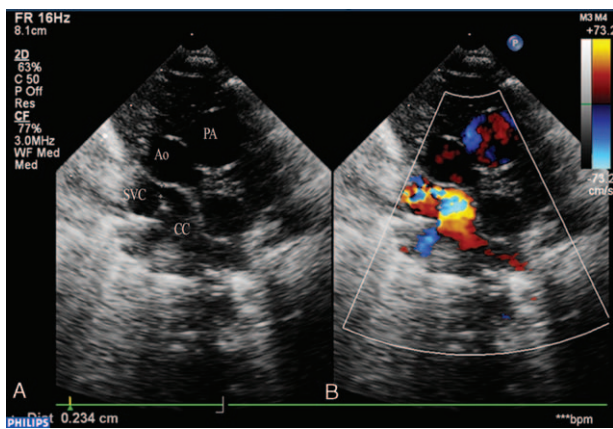


Figure 5. Postnatal echocardiography. (A) 2D image. Suprasternal oblique incidence. The common confluence of the pulmonary veins drained directly into the right SVC. The opening is very narrow (0.23 cm). (B) Color-Doppler flow mapping of the confluence opening into the right SVC showed a high turbulence suggestive for critical obstruction. 2D = bi-dimensional, Ao = aorta, CC = common venous confluence, PA = pulmonary artery, SVC = superior vena cava.

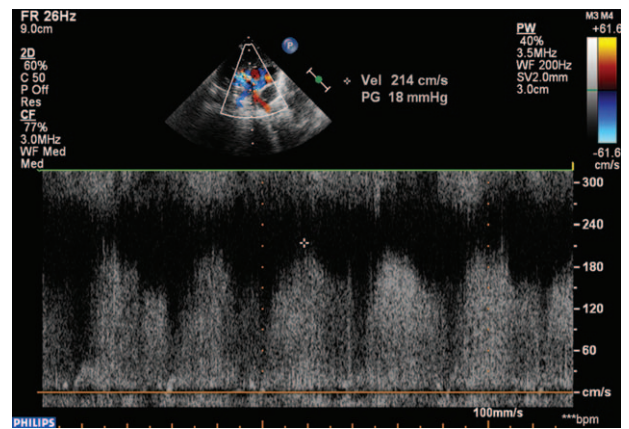


Figure 6. Postnatal echocardiography. Pulsed Doppler evaluation of the pulmonary venous confluence opening into the SVC showed a maximal velocity of 21 cm/s and maximal gradient of 18 mm Hg, suggesting obstruction at that level. SVC = superior vena cava.

2.2. Clinical findings

At 38 weeks of gestation, a male neonate was born in a tertiary cardiovascular Center by cesarean section due to cephalous-pelvic disproportion. His birth weight was 3700 g, length was 57 cm, and head girth was 34cm. The Apgar score at 1 and 5 minutes after birth was 8 and 9, respectively. He presented cyanosis, with tachypnea, a respiratory rate of 60 to 70 breath/min, suprasternal and subcostal retractions. The pulse oximeter showed 73% to 82% of room air oxygen saturation.

2.3. Postnatal diagnostic assessment

Cross-sectional echocardiographic examination performed 1 hour after birth confirmed the prenatal diagnosis. This revealed an enlarged right heart (atrium and ventricle) and ostium secundum type atrial septal defect of 6 mm, with right-to-left shunt. The 4 pulmonary veins drained into a common confluence localized behind the left atrium, which opened directly into the right SVC, without an ascending vertical vein. The color flow mapping of the confluence opening into the right SVC showed a critical obstruction (Fig. 5). Pulsed Doppler evaluation of the pulmonary venous confluence opening into the superior vena cava showed a maximal velocity of 21 cm/s and maximal gradient of 18 mm Hg, suggesting obstruction at that level (Fig. 6). The chest radiography revealed signs of increasing pulmonary venous congestion. Two hours after birth the patient was transferred to the neonatal intensive care unit. A computer tomography angiography was performed, which revealed similar findings (Figs. 7 and 8).

2.4. Therapeutic focus and assessment

The neonate underwent an emergency open heart surgery by performing a side-to-side anastomosis between the pulmonary venous confluence and left atrium. The connection of the confluence with the SVC was ligated.

2.5. Follow-up and outcomes

The postoperative recovery was complicated by pulmonary arterial hypertension, right ventricular myocardial dysfunction

with hemodynamic failure, and unfortunately the newborn died 3 days after surgery.

2.6. Discussions

Anomalous connection of the pulmonary venous confluence directly to the right SVC, especially the obstructed form is an unusual supracardiac variant. In a retrospective, collaborative study involving 19 pediatric cardiac centers from the United Kingdom, Ireland, and Sweden the incidence of TAPVC was about 7.1/100,000 live births. This incidence is higher compared to 6.8 in the Baltimore-Washington Infant Study or 5.9 in the

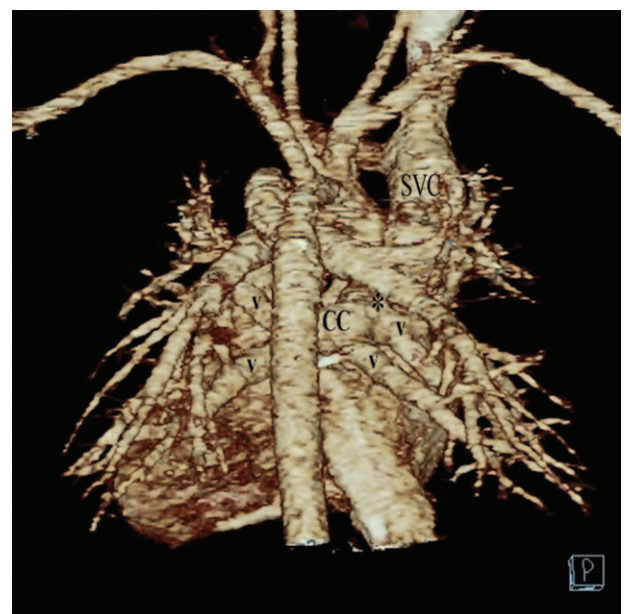


Figure 7. AngioCT of the heart (3D reconstruction). Common confluence of the 4 pulmonary veins, opening directly into the right SVC: AngioCT = computer tomography angiography, CC = common venous confluence, SVC = superior vena cava, v = pulmonary veins, * drainage site of the common venous confluence into the SVC.



Figure 8. AngioCT of the heart. Common pulmonary venous confluence opening into the right SVC: AngioCT = computer tomography angiography, CC = common pulmonary venous confluence, SVC = superior vena cava, * = drainage site of the common venous confluence into the SVC.

New England Regional Infant Cardiac Program.^[7–9] Several studies have reported case series with prenatally diagnosed supracardiac TAPVC,^[10–12] some of them with drainage direct into the right SVC.^[13,14] However, all of these cases were without obstruction at the drainage level. This is the first case of prenatally diagnosed isolated, obstructed supracardiac TAPVC with drainage directly into the right SVC, identified by us in the literature.

TAPVC is well tolerated in utero. This fact can be explained by the particularities of the fetal pulmonary circulation: low pulmonary blood flow, due to high pulmonary vascular resistance. However, Rasanen et al^[15] showed that during the pregnancy there is a slight increase of pulmonary blood flow compared with the combined cardiac output, from 13% at 20 weeks of gestation to 25% at 30 weeks of gestation, with no changes in the last semester. This pulmonary flow increase can explain the right-left ventricular discrepancy after 28 weeks of gestation.^[10] After postnatal transition, the pulmonary blood flow increases and as a result of systemic and pulmonary venous flow mixing in the right heart, the neonate will be cyanotic. If obstruction of pulmonary venous flow is associated, then pulmonary venous congestion occurs. Untreated, this can become very quickly a life threatening condition.^[16]

Prenatal diagnosis of isolated TAPVC is challenging. Some indirect and direct signs suggesting the prenatal diagnosis of isolated supracardiac TAPVC, are described in the literature. The initial sign that can become obvious in the third trimester of pregnancy is the right ventricular predominance, with increase of pulmonary blood flow. Another indirect sign detectable in 4-chamber view is a small left atrial size and “smooth” posterior aspect of it. Dilation of the SVC and detection of an additional vein (vertical vein – in typical supracardiac TAPVC) in 3 VTV is helpful. Low-flow color mapping in order to identify pulmonary veins could be helpful for diagnosis improvement. Also, abnormal pulsed Doppler flow in pulmonary veins can give

important information. Because of the low pulmonary blood flow in fetus, obstruction of the pulmonary venous confluence may be difficult to detect.^[10,14] A4C and 3VT view are of great importance and compulsory in diagnosing isolated supracardiac TAPVC beside Doppler color and pulsed Doppler flow imaging of pulmonary veins. However, in our particular supracardiac TAPVC case, there was no additional vessel in the 3 VTV, because of the direct drainage of the common pulmonary venous confluence into the right SVC. We noticed a dilated right SVC at the site of the pulmonary venous drainage in 3VTV, with a turbulent Doppler color flow at that level. Further careful assessment of the right SVC allowed identification of the drainage site of the common confluence.

TAPVC can also be diagnosed after birth, during infancy, or even later. Recently, it was described a case of a 4-month-old infant without any pathological history, admitted for perioral cyanosis and poor feeding, who was diagnosed with TAPVC, which was successfully repaired by surgical intervention.^[17] Zhang et al^[18] in their study showed that comparing with surgical or AngioCT results, the postnatal echocardiography had a sensitivity of 97.6% and a specificity of 99.9%. Unfortunately, even though we diagnosed our patient in utero, he died after surgery. On the other hand, a study performed on patients with mixed-type TAPVC, underlined the fact that the mortality during the first year after surgery is high, but those who survive past the 1st year, usually present a good prognosis, without sequelae.^[19] TAPVC can be associated with other cardiac malformations, such as in the case described by Ide et al^[20] about a neonate diagnosed with a single ventricle, right atrial isomerism, pulmonary atresia with major aortopulmonary collateral arteries, a small central pulmonary artery and a supracardiac TAPVC, who underwent an uneventful surgical repair. Even though, complications after surgical repair, such as pulmonary arterial hypertension usually appear in the first 6 months after surgery, this can appear also later on life, like in the case described by Martínez–Quintana about a teenager who developed pulmonary venous obstruction and severe secondary pulmonary arterial hypertension after a surgical intervention for TAPVC during infancy.^[21] In the case described above, severe pulmonary arterial hypertension appeared very quickly after the surgical intervention and led to the newborn’s death on the 3rd day after surgery. Other rare cases involving TAPVC were also reported in the specialty literature, such as the association between tetralogy of Fallot with anomalous left coronary artery from pulmonary artery and totally anomalous pulmonary venous connection,^[22] or the coexistence of TAPVC and persistent left superior vena cava.^[23] According to Shi et al^[24] obstructed TAPVC was discovered in 25% of newborns, the mean surgical age were about 215 days of life with 51 deaths of the 768 cases with different types of anomalous pulmonary venous connection.

Although our case had a severe obstructed form and unfortunately died after surgery, we can emphasize that a prenatal diagnosis of isolated, obstructed TAPVC is important for adequate postnatal planning of delivery and surgery in a tertiary center.

3. Conclusions

Prenatal diagnosis of isolated obstructed supracardiac TAPVC with drainage into the right SVC is possible by careful assessment of direct and indirect signs. Prenatal diagnosis of obstructed forms allows emergency treatment after birth in tertiary cardiovascular centers.

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