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Brief Report

Prenatal diagnosis of tetralogy of Fallot with obstructed supracardiac totally anomalous pulmonary venous connection

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Abstract We describe our experience with prenatal diagnosis of tetralogy of Fallot with supracardiac totally anomalous pulmonary venous connection. We also suspected obstruction in the ascending vertical vein as it crossed the right bronchus and coursed superiorly to join the right superior caval vein. This finding was confirmed on postnatal echocardiography, and at autopsy.

Keywords: Fetal cardiology; cyanotic congenital cardiac disease; paediatric cardiology

The association of totally anomalous pulmonary venous connection with tetralogy of Fallot is rare, with a few cases of this combination reported subsequent to echocardiography, cardiac catheterization, at surgery or at autopsy.[1–5] The combination has been corrected surgically with success in infancy, as well in older children.[6–8] As far as fetal diagnosis is concerned, however, totally anomalous pulmonary venous connection is well recognized in association with complex malformations such as the heterotaxic syndromes, especially right isomerism.[9] To the best of our knowledge, nonetheless, totally anomalous pulmonary venous connection in association with tetralogy of Fallot has yet to be diagnosed prenatally. In this report, we describe our prenatal findings of obstructed supracardiac totally anomalous pulmonary venous connection to the right superior caval vein at 21 weeks gestation in association with tetralogy of Fallot.

Case report

A 32-year-old mother of six children was referred for expert fetal echocardiography, a ventricular septal defect having been detected at 21 weeks gestation on routine sonographic screening. Multiple cysts had also been seen in the right kidney. Fetal echocardiography at our centre showed a large perimembranous ventricular septal defect, with the aorta overriding the crest of the ventricular septum (Fig. 1a). There was anterior, superior, and leftward deviation of the muscular outlet septum producing subpulmonary obstruction, along with mild hypoplasia of the pulmonary trunk and the right and left pulmonary arteries. The peak velocity measured across the pulmonary valve was 99 centimetres per second, suggestive of mild obstruction. The peak velocity across the aortic valve was 70 centimetres per second. The ascending aorta was dilated, being measured at 4.8 millimetres diameter, and there was a left-sided aortic arch, with antegrade flow through a left-sided arterial duct. In addition, an abnormal pulmonary venous confluence was visualized behind the left atrium (Fig. 1b). From this confluence, a vein coursed cranially and vertically to the right and posterior to the right pulmonary artery and the right bronchus, joining the right superior caval vein at its junction with the right atrium. Doppler interrogation revealed turbulence in the vertical vein as it crossed the right bronchus, and pulsed Doppler interrogation showed an increased velocity of flow, at 1.3 metres per second, with phasic variation (Fig. 1c,d). The pulsed Doppler pattern in the

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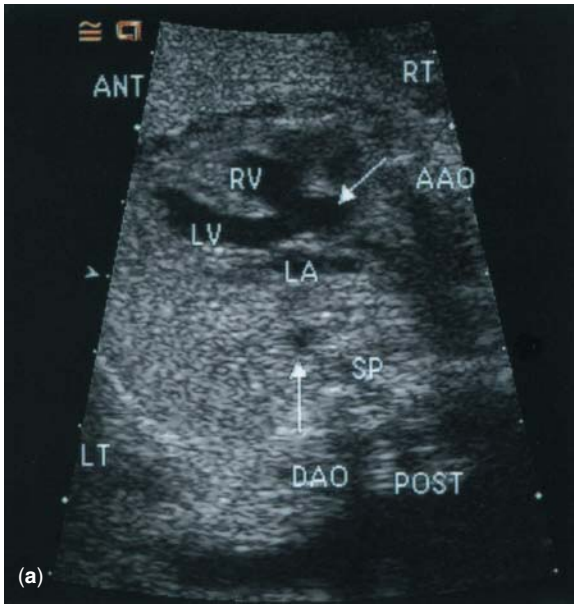

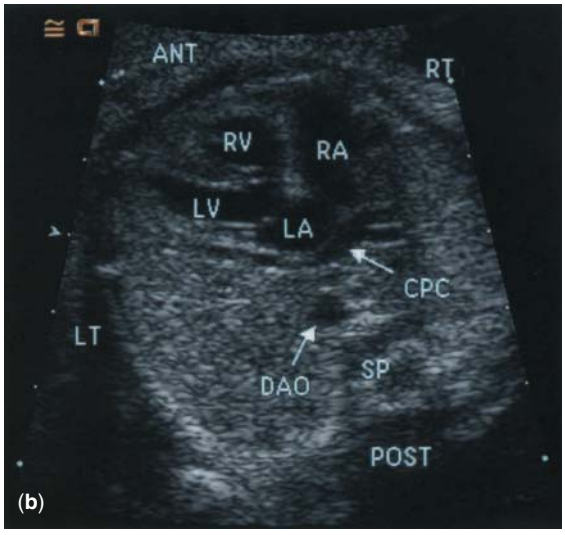

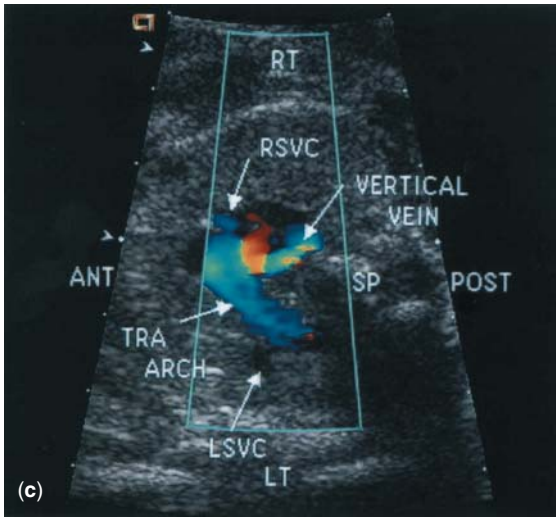

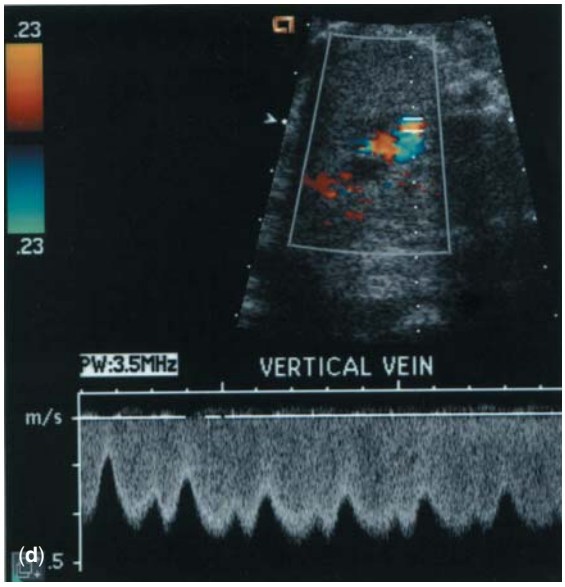

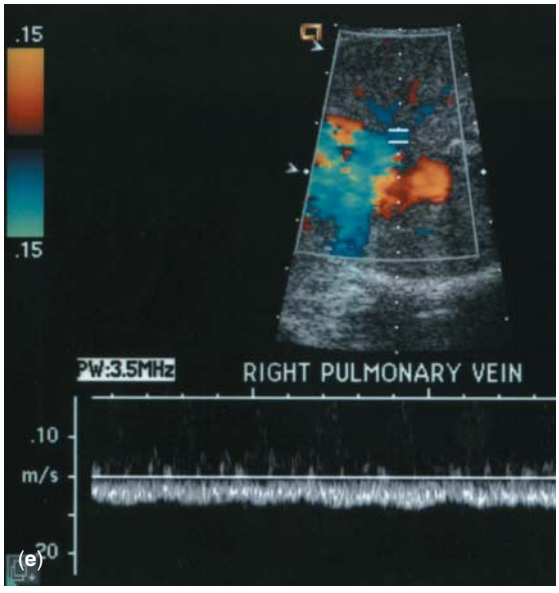

Figure 1.
*(**a**) The long axis view of the left ventricle shows a perimembranous ventricular septal defect with the aorta overriding the crest of the ventricular septum. (**b**) The four-chamber view shows the pulmonary venous confluence behind the left atrium. Note there is an increased space between the left atrium and the descending aorta. (**c**) The cross-sectional view of the upper mediastinum with colour flow Doppler shows the transverse aortic arch, the left superior caval vein, and the dilated ascending vertical vein posteriorly joining the posterolateral aspect of the right superior caval vein. There was turbulence noted in the vertical vein as it crossed the right bronchus. (**d**) Pulsed Doppler at the site of turbulence in the vertical vein shows a high velocity of 1.3 metres per second with phasic flow. (**e**) The pulsed Doppler in the right pulmonary vein shows a damped pattern of flow, suggestive of obstruction in the pulmonary venous pathways. RT: right; LT: left; ANT: anterior; POST: posterior; SP: spine; RA: right atrium; LA: left atrium; LV: left ventricle; RV: right ventricle; CPC: pulmonary venous confluence; AAO: ascending aorta; DAO: descending aorta; TRA ARCH: transverse arch; RSVC: right superior caval vein; LSVC: left superior caval vein.*

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individual pulmonary veins was damped, with a peak velocity of 4.6 centimetres per second in the right pulmonary vein (Fig. 1e), and 7 centimetres per second in the left pulmonary vein, suggestive of obstruction in the pulmonary venous pathway. There was normal biventricular function, and no atrioventricular valvar regurgitation. The coronary sinus was enlarged and drained a persistent left superior caval vein.

Due to the presence of complex cardiac disease, as well as the renal anomaly, the prognosis was felt to be poor. Amniocentesis revealed a normal karyotype, while the screen for 22q11 microdeletion was negative. Pregnancy was not terminated, and follow-up fetal echocardiograms were performed at four-week intervals.

At 32 weeks gestation, following a spontaneous labour, the mother delivered a male infant weighing 2.2 kilograms. Apgar scores of 1, 5, 6 and 6 were recorded at one, five, ten and fifteen minutes, respectively. The neonate was intubated and ventilated, with initial pulse oximeter readings of 82% on 60% oxygen. He had a seizure on the first day of life, and was started on phenobarbital. Subsequently he was weaned to room air, with the pulse oximeter then reading 82% on the second day of life. On examination, he had dysmorphic facial features. All pulses were normally palpable, the praecordium was normal, the first heart sound was normal, and the second heart sound was single. There was no murmur heard on auscultation. The postnatal echocardiogram confirmed the diagnosis of tetralogy of Fallot with obstructed totally anomalous pulmonary venous connections to the right superior caval vein, with a peak gradient of 14 to 16 millimetres of mercury measured in the ascending vertical vein. There was abnormal pulsed Doppler pattern in the individual pulmonary veins. The infant developed renal failure with progressive deterioration in his clinical condition. He died after 14 days.

Autopsy findings confirmed the presence of tetralogy of Fallot with totally anomalous pulmonary venous connection to the right superior caval vein, with obstruction in the vertical vein as it coursed behind the right bronchus and the right pulmonary artery to join the right superior caval vein. There was mild hypoplasia of the pulmonary trunk and right and left pulmonary arteries. There was also persistence of the left superior caval vein, which drained through an enlarged coronary sinus into the right atrium. The non-cardiac anomalies consist of bilateral cystic renal dysplasia and hypoxic-ischemic encephalopathy of the brain.

Discussion

Tetralogy of Fallot is the most common form of cyanotic congenital cardiac disease. On routine prenatal sonographic evaluation, the four-chamber view is normal in fetuses with this malformation. It can be identified on the five chamber view, however, or on the long axis view of the left ventricle, where the crest of the ventricular septum beneath the large subaortic ventricular septal defect is seen to be overridden by the aorta. The three-vessel view also will be abnormal. Although the pulmonary arterial diameters may be normal for gestational age, the ratio of the aortic and pulmonary arterial diameters will be abnormal. It may also be possible to recognize the malaligned muscular outlet septum narrowing the entrance to the right ventricular outflow tract, albeit that a significant gradient across the right ventricular outflow tract may not be detected prenatally.

The largest series reporting the association of anomalous pulmonary venous connection with tetralogy of Fallot came from Redington et al.,[1] who found the anomalous pulmonary veins in 7 of 1183 patients with tetralogy of Fallot. Of their seven patients, three had totally anomalous connections, but the pulmonary venous connections were only partially anomalous in the other four. Among their patients with totally anomalous connection, one was supracardiac, one drained to the coronary sinus, and the other showed a mixed supracardiac and infracardiac pattern of drainage. The diagnosis had been established by cross-sectional echocardiography in one, preoperative or postoperative angiocardiography in five, and at necropsy in one.

The decreased flow of blood to the lungs in the setting of tetralogy of Fallot makes preoperative diagnosis of associated totally anomalous pulmonary venous connection difficult. Patients with obstruction in the pulmonary venous pathways, however, can become symptomatic early. In contemplating surgery in such patients, complete repair of both lesions is recommended. Palliative surgery, like construction of a modified Blalock–Taussig shunt, would augment the flow of blood to the lungs, and increase the pulmonary venous congestion in the presence of an obstructed pulmonary venous connection.

The prognosis of tetralogy of Fallot itself is worse when diagnosed prenatally in association with other non-cardiac or chromosomal anomalies. In our case, management was complicated by prematurity, the obstructed totally anomalous pulmonary venous connection, and renal failure. With improvement in ultrasonic equipment, the experience of the operators, and careful meticulous cardiac segmental analysis, we were able to reach the correct cardiac diagnosis during fetal life, albeit that the outcome proved fatal.

In summary, therefore, we report the first case of tetalogy of Fallot associated with obstructed supracardiac totally anomalous pulmonary venous connection diagnosed prenatally. Interrogation of the

individual pulmonary veins using pulsed and colour flow Doppler, as well as in the ascending vertical vein, was very helpful in reaching the correct diagnosis.

References

1. Redington AN, Raine J, Shinebourne EA, Rigby ML. Tetralogy of Fallot with anomalous pulmonary venous connections: a rare but clinically important association. Br Heart J 1990; 64: 325–328.
2. Muster AJ, Paul MH, Nikaidoh H. Tetalogy of Fallot associated with total anomalous pulmonary venous drainage. Chest 1973; 64: 323–326.
3. Gerlis LM, Fiddler GI, Pearse RG. Total anomalous pulmonary venous drainage associated with Tetralogy of Fallot: report of a case. Pediatr Cardiol 1983; 4: 297–300.
4. Bonvicini M, Picchio FM, Baccarani G, Rapezzi C, Magnani B. Tetralogy of Fallot associated with total anomalous pulmonary venous drainage. Case report. G Ital Cardiol 1980; 10: 1538–1545.
5. Gutierrez J, Perez de Leon J, de Marco E, et al. Tetralogy of Fallot associated with total anomalous pulmonary venous drainage. Pediatr Cardiol 1983; 4: 293–296.
6. Webber EM, Gillis DA, Ross DB. Tetralogy of Fallot with total anomalous pulmonary venous drainage and esophageal atresia: complete correction in infancy. Ann Thorac Surg 1996; 62: 571–573.
7. Lee ML, Wu MH, Lue HC. Infracardiac total anomalous pulmonary venous connection in tetralogy of Fallot with decreased pulmonary flow and masked pulmonary venous obstruction: report of one case. Int J Cardiol 1994; 47: 81–84.
8. Neema PK, Krishnamanohar SR, Rathod RC. Tetralogy of Fallot with total anomalous pulmonary venous connection: pathophysiology and management. J Cardiothorac Vasc Anesth 2002; 16: 211–213.
9. Valsangiacomo ER, Hornberger LK, Barrea C, Smallhorn JF, Yoo SJ. Partial and total anomalous pulmonary venous connection in the fetus: two-dimensional Doppler echocardiographic findings. Ultrasound Obstet Gynecol 2003; 22: 257–263.