The echocardiographic diagnosis of totally anomalous pulmonary venous connection in the fetus

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Abstract

Background—Infants with isolated totally anomalous pulmonary venous return often present severely decompensated, such that they are at high risk for surgical repair. On the other hand, if surgical repair can be safely accomplished, the outlook is usually good. Thus prenatal diagnosis would be expected to improve the prognosis for the affected child.

Objective—To describe the features of isolated totally anomalous pulmonary venous drainage in the fetus.

Design—Four fetuses with isolated totally anomalous pulmonary venous connection were identified and the echocardiographic images reviewed. Measurements of the atrial and ventricular chambers and both great arteries were made and compared with normal values.

Setting—Referral centre for fetal echocardiography.

Results—There were two cases of drainage to the coronary sinus, one to the right superior vena cava, and one to the inferior vena cava. Right heart dilatation relative to left heart structures was a feature of two cases early on, and became evident in some ratios late in pregnancy in the remaining two.

Conclusions—Ventricular and great arterial disproportion in the fetus can indicate a diagnosis of totally anomalous pulmonary venous connection above the diaphragm. However, in the presence of an atrial septal defect or with infradiaphragmatic drainage, right heart dilatation may not occur until late in pregnancy. The diagnosis of totally anomalous pulmonary venous drainage in fetal life can only be reliably excluded by direct examination of pulmonary venous blood flow entering the left atrium on colour or pulsed flow mapping.

Keywords: congenital heart disease; fetus; total anomalous pulmonary venous drainage; echocardiography

In cardiac malformations that are associated with decompensation soon after birth, the diagnosis of structural heart disease in fetal life improves the outlook for morbidity during postnatal management and has also been shown to lower the mortality. Neonates can be maintained in optimal status before surgical repair by the use of prostaglandin treatment, ventilation, and interventional catheterisation procedures where appropriate. Although totally anomalous pulmonary venous drainage is a rare lesion in its isolated form, occurring only about one in 17 000 live births, it is one of the few emergencies left in paediatric cardiological practice. This is because neonates with obstructed pulmonary venous return can present with severe decompensation soon after birth, and unlike other forms of congenital heart disease, maintaining ductal patency using prostaglandins is of little help. On the other hand, the subset of patients with anomalous connection to the coronary sinus does not tend to present urgently, as the drainage to this site is rarely obstructed.

In the neonate, totally anomalous pulmonary venous drainage is notoriously misdiagnosed as persistent pulmonary hypertension—the clinical picture is similar and even the echocardiographic findings overlap. The echocardiogram typically shows right heart dilatation, evidence of raised pulmonary artery pressure, and a right to left shunt at the atrial septum. The key to the diagnosis is the lack of pulmonary venous blood flow to the left atrium and the finding of an anomalous venous channel draining above the heart to the coronary sinus, or alternatively below the diaphragm.

The management involves early surgical repair, which can be accomplished at low risk in neonates who are in good haemodynamic condition. In addition, the long term prognosis is usually good in this condition after complete repair. Thus the condition is one which would benefit particularly from prenatal diagnosis in order to avoid early decompensation and neonatal transfer, and to optimise surgical results. In a previous publication, two cases of isolated totally anomalous pulmonary venous drainage were detected in the fetus, but the diagnostic features of these cases were not described. The aim of this study was to review the echocardiographic findings in the cases we have seen in fetal life in order to find features suggestive of this diagnosis which might allow identification of this important condition more often prenatally.

Methods

The combined database of fetuses with congenital heart malformations in Guy’s Hospital dating from 1980 to 1998 and in New York Presbyterian Hospital between 1993 and the end of 1998 was searched for the diagnosis of isolated totally anomalous pulmonary venous drainage. Of 2370 fetuses with congenital heart disease, four were identified with this lesion.
Cases with totally anomalous pulmonary venous connection, identified in the setting of isomerism of the right atrial appendages and associated with additional intracardiac malformations, were excluded from this analysis. All cases were seen between 1986 and 1991. To our knowledge, no case was missed in our series, and postnatal follow up is obtained in over 90% of patients studied. The videotapes of each case at each study were reviewed. The atrial and ventricular dimensions, the aorta, and the pulmonary artery were measured directly where possible, or ratios of these structures were used where calibration was not possible on old videotapes.

Results

Of the four cases, anomalous drainage was to the coronary sinus in two, supracardiac to the right superior vena cava in one, and below the diaphragm to the inferior vena cava in one. In one of the cases with drainage to the coronary sinus, the correct diagnosis was made prospectively in fetal life. In one case of drainage to the coronary sinus and in the case with supracardiac drainage, coarctation of the aorta was suspected from the fetal echocardiogram and the correct diagnoses were made after birth. In the case with infradiaphragmatic drainage, the diagnosis was made at necropsy.

CASE 1

This fetus was seen at 20, 22, and 26 weeks of gestation because of totally anomalous pulmonary venous drainage in one previous infant of this mother and an atrioventricular septal defect in another offspring. The mother was a known carrier of a balanced translocation. There was pronounced disproportion of the ventricles and great arteries seen on the first scan, with the transverse arch smaller than the arterial duct. The pulmonary veins were seen on cross sectional imaging to be connecting to a dilated coronary sinus (fig 1) and draining to the right atrium. Colour flow mapping was not available at the time. Only ratios were obtainable from the initial two tapes, but they showed abnormal mitral valve to tricuspid valve (MV:TV), left ventricle to right ventricle (LV:RV), and aorta to pulmonary artery (Ao:PA) ratios. The right atrium was over twice the size of the left. On the study at 26 weeks, the aortic and left ventricular widths were below the fifth centile, whereas the pulmonary artery, tricuspid valve orifice, right ventricular, and right atrial widths were above the 95th centiles, giving very abnormal MV:TV, LV:RV, and Ao:PA ratios (figs 2 and 3). The right atrium to left atrium ratio was > 2. Multiple malformations were found in this fetus in association with an additional chromosomal fragment. Surgical repair took place at three months but the child succumbed during the first year as a result of the extracardiac anomalies.

CASE 2

This fetus was examined at 20, 24, 30, and 36 weeks of gestation because of an abnormal four chamber view on obstetric scanning. The heart lay centrally in the chest, the pulmonary veins appeared to drain normally on cross sectional imaging (fig 4), and there was a dilated coronary sinus. Colour flow mapping was only used
at the 24 week study but did not evaluate the pulmonary veins directly. At 20 and 24 weeks, the chamber and great artery measurements were within the normal range. At 30 and 36 weeks, the aortic and left ventricular measurements were below the fifth centile, whereas the pulmonary artery and right ventricle were still within normal limits. This gave mildly abnormal MV:TV, LV:RV, and Ao:PA ratios (figs 2 and 3). The transverse arch was smaller than the duct. A small but convincing pericardial effusion developed by 24 weeks and persisted. The right atrium was equal to the left in early pregnancy and slightly dilated towards the end of pregnancy, but there was a widely patent foramen ovale. A coarctation lesion was suspected but anomalous pulmonary venous drainage was considered a possible differential diagnosis. Totally anomalous pulmonary venous drainage to the coronary sinus was found at birth and the infant underwent successful surgical repair.

CASE 3
This patient was examined at 17, 27, and 32 weeks of gestation because of a history of a previous child with totally anomalous pulmonary venous drainage. The study was considered normal at 17 weeks, and the LV:RV and Ao:PA ratios were normal (fig 5). The pulmonary veins appeared to drain to the left atrium on cross sectional scanning, although colour flow mapping was not available. At 27 weeks, the cardiac study still appeared normal but there was major hydramnios. Restudy at 32 weeks still showed hydramnios and small but convincing bilateral pleural and pericardial effusions. The atrial chambers at each study were within the normal range for gestation and equal in size. The right ventricle was at the 95th centile for size, the left ventricle below the 5th, the pulmonary artery was above the 95th centile, and the aorta was on the 5th centile. This gave mildly abnormal LV:RV, MV:TV, and Ao:PA ratios (figs 2 and 3). Colour flow mapping, although used, did not give adequate penetration owing to the hydramnios, with the fetus lying between 15–18 cm from the transducer. The fetus was acutely distressed at delivery at 34 weeks’ gestation and died within one hour of birth despite all resuscitative efforts. Necropsy showed totally anomalous pulmonary venous drainage to a hypoplastic channel which drained below the diaphragm.

CASE 4
This patient was seen at 19, 28, and 34 weeks of gestation because of a previous child with...
the hypoplastic left heart syndrome. Disproportion of the ventricles and great arteries was seen at the first two studies, with abnormal LV:RV, MV:TV, and Ao:PA ratios (direct measurements were not obtainable). The right atrium was more than twice the size of the left atrium. In retrospect, the superior vena cava was prominent in all views and on the three vessel view was larger in size than the aorta. At 34 weeks’ gestation, the left atrium, left ventricle, and aorta were below the 5th centile for size and the right atrium and pulmonary artery were above the 95th centile. The right ventricular measurements were at the upper limit of normal. This gave abnormal MV:TV, LV:RV, and Ao:PA ratios (fig 6A). The transverse arch was about half the size of the duct (fig 6B). Colour flow mapping was used but the pulmonary veins were not demonstrated. A coarctation lesion was suspected. Delivery took place in the cardiac centre and obstructed totally anomalous pulmonary venous drainage to the superior vena cava was found postnatally. The neonate died at surgical repair. Necropsy was declined.

Discussion

During a detailed or targeted fetal echocardiogram in a high risk patient, it is our policy that it is essential to identify at least one pulmonary vein on direct examination draining to the left atrium (fig 7), and this can nearly always be achieved. In practice, both the right and left pulmonary veins are usually seen simultaneously. The use of newer technologies such as power Doppler can aid the detection of the pulmonary veins. However, the proximity of the upper veins to the pulmonary arteries can cause confusion, as power Doppler does not show direction of flow. Pragmatically, as it is only totally anomalous drainage that is life threatening, identification of one normally draining vein is sufficient to exclude this diagnosis. As in postnatal life, the cross sectional image alone may be misleading, and flow must be seen entering the left atrium on colour flow mapping in every case. The identification of the pulmonary veins on colour was not our routine policy until the early 1990s, although colour flow mapping had been available to us in some cases from 1986. Two of the four cases were detected early because of disproportion, noted particularly in the ventricular size. In contrast, dilatation of right heart structures did not occur until late in pregnancy in the remaining two cases. However, at this time in gestation some disproportion of right heart structures relative to the left can occur in the normal fetus. Interestingly, three of the four cases described were referred for a history of congenital heart disease in a previous child, which in two cases was anomalous pulmonary venous drainage. This contrasts with data from the Baltimore-Washington infant study, where only three of 60 neonates with this diagnosis had a family history.

In the human fetus, more than 20% of the combined ventricular output passes to the lungs and thus returns to the left atrium in the pulmonary veins, increasing to 25% in the last 10 weeks of pregnancy. This represents about 50% of left ventricular flow. If all the pulmonary venous return is to the coronary sinus or a supracardiac vein, this additional volume of flow will be to the right atrium, right ventricle, and pulmonary artery. As a result, the right heart will be dilated and the left heart smaller than normal, causing abnormal LV:RV and Ao:PA ratios (figs 2 and 3). However, in two cases—one to the coronary sinus and one to below the diaphragm—there was no abnormality in these ratios until late in pregnancy. This can be explained partly by increasing lung blood flow later in pregnancy but also perhaps by the large atrial septal defect in case 2, allowing some of the excess venous return to the right atrium to reach the left atrium. In case 3, the pulmonary venous blood drained below the diaphragm but this flow will be predominantly directed from the inferior vena cava across the foramen ovale to the left ventricle, resulting in the equal ventricular sizes that were found in early pregnancy. Additionally, if pulmonary venous return is obstructed, this will tend to increase pulmonary resistance, diverting blood away from the branch pulmonary arteries into the duct, reducing the volume of pulmonary venous return. Thus the most severe forms of obstructed pulmonary venous return may be less obvious from indirect echocardiographic signs in utero than the less obstructed forms.

Reduction in the volume of pulmonary flow prenatally may be part of the cause of peripheral pulmonary venous stenosis or widespread venous hypoplasia, which can be a fatal condition. Perinatal death is the most severe cases of obstructed totally anomalous pulmonary venous drainage. This may have caused the pleural effusions which were prominent in case 3, although the associated hydramnios—which was pronounced—is less easy to explain. If the veins are not found to be draining normally during cardiac evaluation in utero, an ascending or descending venous channel should be sought. However, this may be small and difficult to find in the presence of obstruction and consequently diminished pulmonary blood flow. A site of turbulence in the upper thorax or upper abdomen at the site of obstruction to the venous pathway might be a clue to the site of drainage in supracardiac or
infracardiac drainage, respectively, where this diagnosis is suspected.

The normal pattern of pulmonary venous flow on pulsed Doppler has recently been described2 13 and the presence of a normal pattern appears to confirm a normal connection to the left atrium. In contrast, an abnormal flow pattern led to the detection of infradiaphragmatic anomalous pulmonary venous drainage in the setting of isomerism of the right atrial appendages.7 9 However, where there is a suspected diagnosis of an asplenia syndrome, the site of pulmonary venous drainage should always be sought, as anomalous drainage is a common associated feature which can be identified in the fetus.7 Recognition of this complicating aspect of the diagnosis is important in this condition, but it is particularly in the isolated form of total anomalous pulmonary venous drainage—where there can be a good prognosis if surgical repair can safely be accomplished—where there would be a real advantage in prenatal diagnosis.

Although three of these four cases did not survive, one died of extracardiac anomalies after successful repair of the anomalous veins, and one (case 4), although obstructed, is likely to have survived in the modern surgical era, especially if the diagnosis was anticipated. A fatal outcome in many cases with obstructed veins below the diaphragm is now preventable, but it is unlikely that our case (case 3) could have been saved because of the anatomical hypoplasia present throughout the pulmonary venous channels and demonstrated at necropsy.

This admittedly small series of cases does show some important findings. Indirect evidence of abnormality such as ventricular hypoplasia present throughout the pulmonary venous channels and demonstrated at necropsy.

Doppler visualisation of pulmonary vein flow into the left atrium and a normal pulsed flow pattern should be documented in order to exclude this important diagnosis. Where a diagnosis of coarctation of the aorta is suspected in the fetus, an alternative diagnosis of totally anomalous pulmonary venous drainage should be specifically excluded. Lastly, where the diagnosis of totally anomalous pulmonary venous connection is made correctly, amniocentesis should be considered because, although this is rarely associated with chromosomal anomalies, an unbalanced translocation did occur in one of our cases and led to eventual demise.

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