

Absence of the Aortic Valve: Antenatal and Postnatal Two-Dimensional and Doppler Echocardiographic Features

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Antenatal and postnatal two-dimensional and gated pulsed Doppler echocardiography, beginning at 32 weeks' gestation, were used to evaluate the cardiovascular contribution to nonimmunologic hydrops in a gravida 1, para 0 nineteen year old woman. Antenatal two-dimensional imaging demonstrated a hypoplastic left heart variant of complete common atrioventricular (AV) canal. Antenatal gated pulsed Doppler examination of the fetal thoracoabdominal aorta revealed atypical pandiastolic

retrograde flow. Postnatal noninvasive examination at 36 weeks' gestational age was unchanged from the prenatal study.

Postmortem examination confirmed the noninvasive anatomic findings and revealed polysplenia. The mechanism of pandiastolic retrograde flow was attributable to "absence of the aortic valve," a previously unreported congenital cardiac anomaly, which resulted in severe antenatal and postnatal aortic regurgitation.

Two-dimensional echocardiography permits direct visualization and rapid identification of simple and complex cardiac anomalies in the newborn (1-5). Integration of this modality with gated pulsed Doppler echocardiography extends comprehensive noninvasive diagnostic cardiology to the fetus. This case report summarizes the combined use of both ultrasound techniques in the antepartum recognition of a unique constellation of intra- and extracardiac anomalies.

Case Report

The patient, a 19 year old woman, gravida 1, para 0, had routine obstetrical ultrasound examination at 32 weeks' gestation for evaluation of severe polyhydramnios. This study revealed fetal ascites, hepatomegaly and an abnormal cardiac configuration. The patient was referred for evaluation of fetal cardiovascular anatomy using combined two-dimensional, gated pulsed Doppler echocardiography.

Fetal echocardiography. Subxiphoid equivalent projections (1-5) were generated using an Advanced Technology Laboratory Mark 500 cardiac imager with 3.0 and

5.0 MHz in-line scan heads. Atrial and ventricular long- and short-axis projections displayed a common atrium related to a large anterior and hypoplastic posterior atrioventricular (AV) valve (Fig. 1a). The continuity of the AV valves across the plane of the ventricular septum suggested an extensive endocardial cushion anomaly. The anterior morphologic right ventricle was considerably larger than the posterior left ventricle that was akinetic, thick walled and morphologic (Fig. 1b). A single, large semilunar root arose from the right ventricular outflow tract. Subxiphoid equivalent parasagittal projections revealed two parallel vertical vessels in the posterior mediastinum that traversed the diaphragm and continued caudally in the retroperitoneal space (Fig. 2a). Gated pulsed Doppler examination of the smaller, posterior, vertical channel revealed cranially directed, nonpulsatile venous flow. Examination of the anterior vertical channel revealed a caudally directed arterial waveform (Fig. 2b). The profile of the systolic waveform in the arterial channel, that is, the thoracoabdominal aorta, was normal. The diastolic component, however, was unusual in its variability with position of the sample volume in the thoracic aorta. A normal antegrade diastolic flow pattern was recorded with the sample volume in the thoracic aorta adjacent to the ductal/aorta confluence. In contrast, pandiastolic retrograde flow was present with the sample volume in the more distal supraceliac abdominal aorta.

Neonatal presentation. Spontaneous labor occurred at 37 weeks' gestation and resulted in the premature delivery

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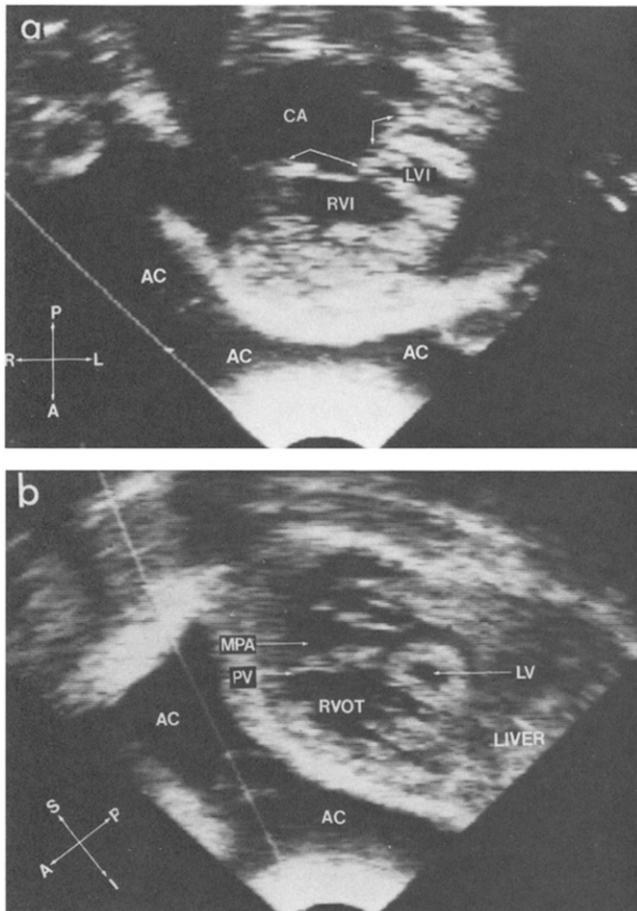


Figure 1. Fetal two-dimensional echocardiogram. **a**, Atrial long-axis view demonstrating a common atrium (CA), a large anterior tricuspid and hypoplastic posterior mitral valve (arrows). **b**, Short-axis view through the right ventricular outflow tract (RVOT) and mid-equatorial plane of the left ventricle (LV). The right ventricular outflow is continuous with the pulmonary semilunar root. The posterior, thick walled hypoplastic left ventricle contains dense, circumferential endocardial signal. A = anterior; AC = amniotic cavity; I = inferior; L = left; LVI = left ventricular inflow; MPA = main pulmonary artery; P = posterior; PV = pulmonary valve; R = right; RVI = right ventricular inflow; S = superior.

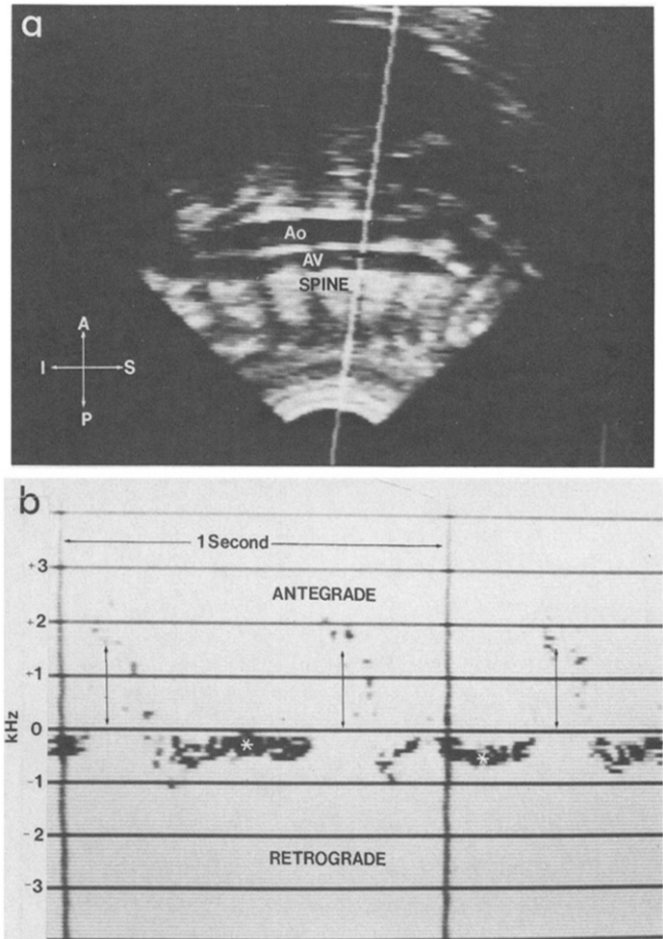


Figure 2. a, Fetal echocardiogram with the echocardiographic beam parallel to the sagittal plane of the trunk demonstrating the vertical vascular channels in the posterior thorax and upper abdomen. **b**, Frequency spectral display with sample volume positioned in distal thoracic aorta. *Retrograde diastolic flow revealed by the broad band negative Doppler frequency display. Ao = aorta; AV = azygous vein; SPINE = vertebral bodies; other abbreviations as before.

of a 2,590 g male infant. The Apgar scores were 6 at 1 minute and 7 at 5 minutes. Physical examination in the delivery suite revealed an acyanotic infant in moderate respiratory distress with grunting and retractions. The pulse was 112 with a respiratory rate of 58. No bruits were audible over the calvarium or abdomen. The thorax was symmetric with an active left precordium. A grade 2/6 systolic and 2/4 diastolic murmur were present at the left midsternal border. An S_3 gallop was noted. The abdomen was distended with the hepatic margin beyond the level of the umbilicus in the right midclavicular line. The pulses of the upper and lower limbs were symmetric and bounding. The chest roentgenogram revealed cardiomegaly and symmetric pulmonary vascular congestion. The electrocardiogram demonstrated

sinus tachycardia with a left superior frontal axis and right ventricular predominance.

Neonatal echocardiogram. The repeat two-dimensional echocardiogram demonstrated viscerotrial situs solitus, d-ventricular looping and a double-outlet right ventricle with a transitional form of the Rastelli type A complete common AV canal (6). The anterior AV valve was large and the posterior valve was hypoplastic with a cleft in its anterior leaflet (Fig. 3). A small ventricular septal defect was noted with a dense matrix of septal chordae inserting on its crest. The posterior left ventricle had a small lumen with marked thickening of the free wall (Fig. 4). Although two great vessels arose from the right ventricle, only the left-sided root had mobile semilunar cusps (Fig. 5). An immobile, unrestrictive ridge was present on the proximal endocardial surface of the "valveless" great vessel. A gated

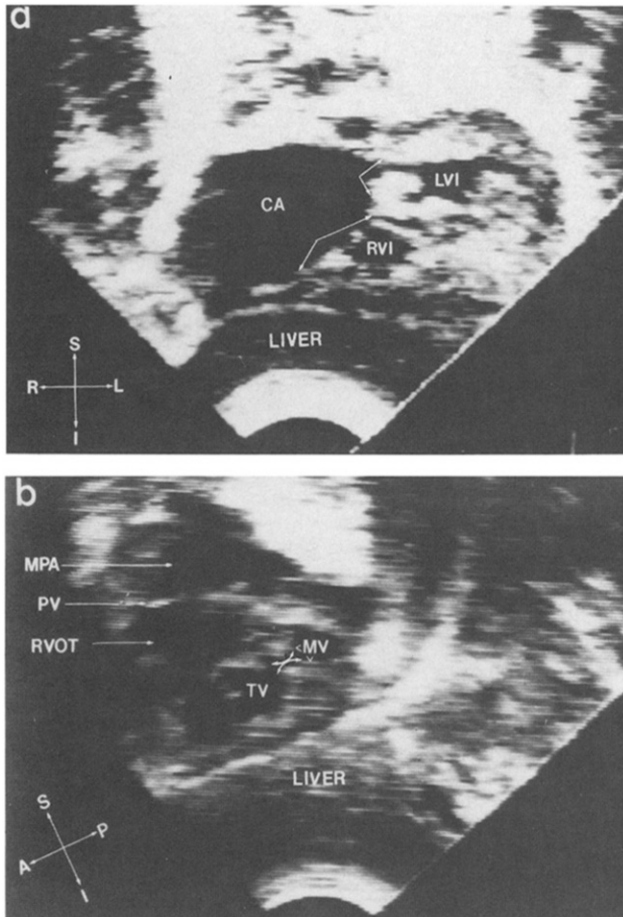


Figure 3. Neonatal two-dimensional echocardiogram. **a**, Atrial long-axis projection demonstrating a common atrium (CA) and right and left ventricular inflow (LVI) segments (↙ ↘). **b**, Short-axis projection through ventricular inflow segments demonstrating cleft of anterior mitral leaflet and ventricular septal defect. The interventricular communication is contiguous with the mitral and tricuspid anuli. An endocardial cushion type ventricular septal defect is shown by the **arrows**. MV = mitral valve; **arrows** indicate superior and inferior endocardial cushion contributions; other abbreviations as before.

pulsed Doppler examination of the thoracoabdominal aorta was repeated and again a relatively normal systolic waveform was noted. In contrast to the antenatal examination, pandiastolic retrograde flow was displayed throughout the distal thoracic aorta (Fig. 6).

Cardiac catheterization. Cardiac catheterization was performed at 18 hours of age with biplane cineangiography of the right ventricle and proximal thoracic aorta. Ventriculography suggested a single, large anterior ventricle with no posterior chamber. Aortography revealed a double aortic arch with an intact right cervical but interrupted left segment. The ductus arteriosus was patent. Severe semi-lunar valve regurgitation rapidly opacified the large anterior ventricle.

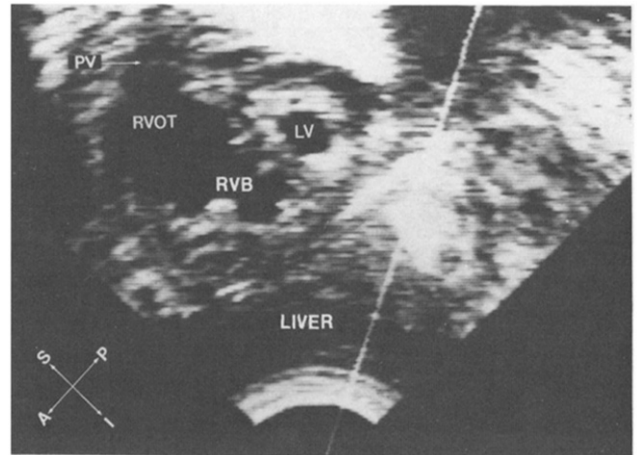
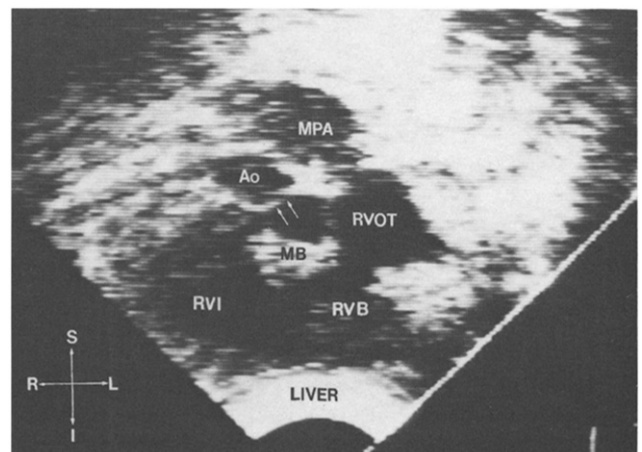


Figure 4. Neonatal two-dimensional short-axis echocardiogram in projection through the right ventricular outflow tract (RVOT) and midsagittal plane of the left ventricle demonstrating a thick walled left ventricle with dense endocardial signal. RVB = right ventricular body; other abbreviations as before.

The infant remained in intractable congestive heart failure and died at 20 hours of age.

Postmortem examination. Examination revealed viscerotransverse situs solitus, d-ventricular looping and a double-outlet right ventricle. The posterior ventricle was hypoplastic and a Rastelli type A complete, common AV canal with a small ventricular septal component was present. The mitral valve was hypoplastic but patent with a cleft in its anterior leaflet (Fig. 7). The interatrial communication resulted in a common atrial chamber. The interventricular septal defect in the endocardial cushion segment was small with a dense matrix of thickened chordae inserting on its crest. Both great vessels were totally related to the right ventricle; however, the outflow tract was partitioned by a

Figure 5. Neonatal coronal right ventricular projection of two-dimensional echocardiogram demonstrating dual origin of both great vessels from the right ventricle. **Arrows** indicate aortic endothelial ridges. MB = muscle bundle; other abbreviations as before.



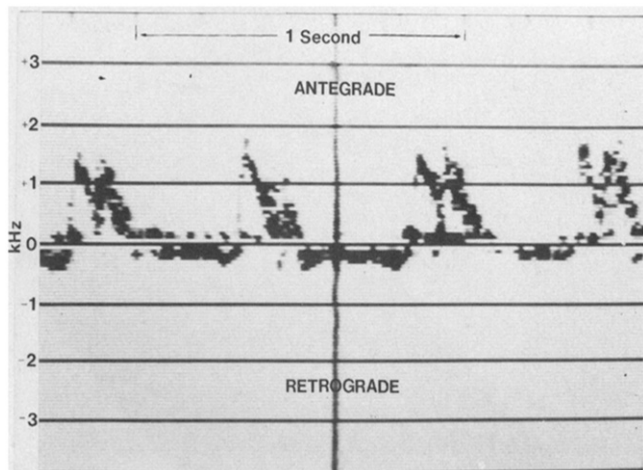


Figure 6. Spectral display of the postpartum gated pulsed Doppler study recorded in the thoracoabdominal aorta. Diastolic retrograde flow is still evident as a negative Doppler shift. Abbreviations as before.

broad, nonobstructive muscle bundle. The pulmonary valve was anterior and leftward. The dilated main pulmonary artery was continuous with the descending aorta across the ductus arteriosus. The juxtaductal aorta was narrowed but not restrictive. The proximal ascending aorta arose from the right ventricle to the right of the pulmonary root.

The striking feature of the cardiac anatomy was the absence of cusps in the aortic root (Fig. 8). The aortic endothelium below the two coronary ostia formed shallow, nonobstructive flat ridges with no apparent functional organization. The remainder of the postmortem examination was remarkable for interruption of the suprarenal vena cava with azygous continuation of venous drainage to the left

Figure 7. Hypoplastic left ventricle at autopsy viewed through an apical incision revealing the thickened posterior free wall, pale endocardium and small mitral valve with cleft in its anterior leaflet; **asterisk** indicates the ventricular septal defect; **IVS** = interventricular septum; **LVFW** = left ventricular free wall; **arrows** show superior and inferior components of anterior mitral leaflet.

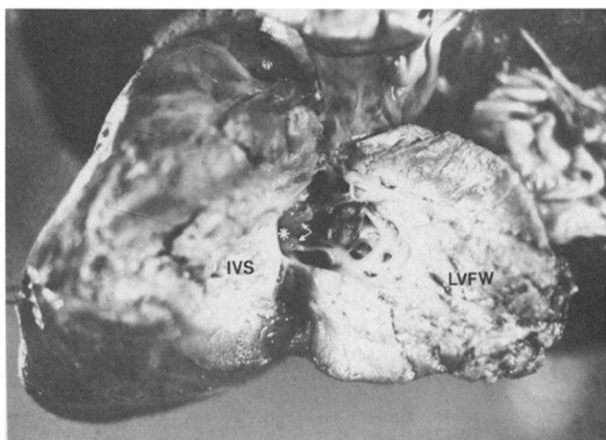


Figure 8. Proximal ascending aorta (AO) with vertical incision extending across ventricular outflow tract. Two coronary ostia (**black arrows**) are present immediately distal to shallow endothelial ridges (**white arrows**). MB = muscle band; MPA = main pulmonary artery.

superior vena cava. In addition, a double aortic arch was present with hypoplasia of its left segment. The right superior vena cava was absent and the left superior vena cava drained anomalously to the right atrium.

The spleen was represented by multiple discrete nodules in the left upper quadrant of the abdomen. An unobstructed midgut malrotation was present.

Discussion

This case illustrates the utility of gated pulsed Doppler study in examining complex cardiovascular anomalies during diagnostic fetal echocardiography. The profound cardiovascular problems of neonates with complex congenital heart disease are usually not manifest during gestation. Fetal congestive heart failure presenting as nonimmunologic hydrops has been reported with sustained fetal arrhythmias (7,8), arteriovenous malformations, Ebstein's anomaly of the tricuspid valve, classic hypoplastic left heart syndrome and endocardial fibroelastosis (9,10). Antenatal presentation, however, of these lesions is unusual. Major intra- and extracardiac anomalies are palliated by in utero patency of the ductus arteriosus and shunting across the foramen ovale.

Antenatal and postpartum two-dimensional echocardiography. Standard two-dimensional echocardiographic subxiphoid projections, or their equivalents, permit detailed antepartum studies. In this case, atrial long-axis projections revealed the common atrium resulting from absence of the endocardial cushion, septum primum/secundum and sinus venosus portions of the interatrial septum. The presence of a complete, common AV canal was suggested by the confluence of the interatrial defect with the posterior interventricular septal communication. In addition to the interventricular septal defect, ventricular short-axis projec-

tions revealed the cleft of the anterior mitral leaflet, hypoplasia of the left ventricular inflow and a thick walled, akinetic left ventricular body. Postpartum coronal right ventricular projections revealed the double-outlet configuration of the right ventricle that was not apparent on antepartum examinations.

Integration of antenatal two-dimensional and Doppler echocardiography. Integration of parasagittal subxiphoid projections and gated pulsed Doppler ultrasound permitted detailed examination of thoracoabdominal vasculature. Interruption of the inferior vena cava was suggested by the absence of infrahepatic/suprarenal caval continuity (11). The gated pulsed Doppler examination differentiated the azygous vein from other vessels found in the posterior mediastinum and retroperitoneum. In addition to the normal thoracoabdominal aorta, a posterior vertical channel may be found in the posterior mediastinum and retroperitoneal space when anomalous infradiaphragmatic pulmonary venous return to the portal system is present. Anatomic imaging in the fetus may not discriminate between these vascular structures. Targeted gated pulsed Doppler ultrasound, however, identifies the vascular organization by systolic waveform and direction of flow. In the fetus, the Doppler systolic spectral waveform discriminates between arterial and central venous vessels. The direction of flow differentiates between cranially directed anomalous systemic venous return to the atria, that is, azygous vein, and caudally directed anomalous pulmonary venous drainage to the portal system.

Doppler findings on fetal aortic flow. In this case, gated pulsed Doppler spectral data also revealed a major adulteration of fetal aortic flow. The placenta is a low resistance network in the fetal systemic arterial system (12). The reduced resistance is normally reflected by the increase of peak diastolic frequencies during passive antegrade diastolic aortic blood flow. In this case, however, fetal juxtaductal antegrade diastolic blood flow contrasted with the retrograde diastolic flow in the distal thoracoabdominal aorta. The diastolic retrograde flow reflected the free aortic regurgitation with absence of aortic cusps.

In principle, this antenatal aortic waveform with diastolic retrograde flow is not unique to the anomaly summarized in this report. Similar findings in the distal thoracic aorta of the fetus would be anticipated with absence of the pulmonary valve with patency of the ductus arteriosus or with isolated cerebral arteriovenous malformation. In both of these lesions, the systemic arterial network experiences a

low resistance diastolic "sink." Patency of the ductus arteriosus in the absence of a competent pulmonary valve results in systemic diastolic regurgitation. Similarly, direct communication between the cerebral arterial and venous circulation permits "regurgitation" of distal thoracic aorta blood volume toward the transverse aortic arch.

Conclusion. The organization of the fetal cardiovascular system is a major adaptation of the fetal placental unit. Antenatal cardiac evaluation of the fetus must take into consideration these modifications and their impact on anatomic anomalies of the cardiovascular system. Integration of two-dimensional echocardiographic definition of anatomy and targeted gated pulsed Doppler display of flow permits detailed examination of the most complex fetal cardiovascular anomalies as in this case of double-outlet right ventricle, complete AV canal and absent aortic valve.

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