CASE REPORT

Congenital absence of aortic and pulmonary valve in a fetus with severe heart failure

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Abstract
A case of congenital absence of both aortic and pulmonary valves with severe heart failure detected prenatally by cross-sectional and pulsed and colour Doppler echocardiography is reported in small for gestational age male fetus in 17th week of gestation. Additional double outlet right ventricle, hypoplastic left ventricle, and ventricular septal defect, as well as multiple extracardiac anomalies, were found by prenatal echocardiographic investigation and confirmed by necropsy examination. Retrograde diastolic Doppler waveforms retrieved from pulmonary artery, aorta, and umbilical arteries revealed massive insufficiency throughout both the great arteries, which eliminated diastolic placental perfusion, documented by absent anterograde diastolic flow in the umbilical vein. These prenatal echocardiographic findings may contribute to an understanding of the mechanism of rapid and progressive heart failure and growth retardation in the fetus. Severe cardiac failure may explain why congenital aplasia of both the aortic and the pulmonary valves has not been described postnatally, and only two fetal cases revealed by necropsy have been published.

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Congenital absence of both semilunar valves in separated great arteries is an exceedingly rare heart malformation. The only reported examples are described by Hartwig et al., who found congenital absence of semilunar valve leaflets in complex congenital heart defects in two fetal necropsy examinations. Here we describe a case of severe heart failure in a fetus with absence of both aortic and pulmonary valves, revealed by fetal echocardiographic examination in the 18th week of gestation.

Case report
A 33 year-old healthy gravida 2, para 1 was referred for detailed fetal cardiovascular evaluation at week 17 + 4 of gestation for a small for gestational age fetus with hydrocephaly, hydrothorax, ascites, and heart malformation, suspected from cardiomegaly and abnormal

Doppler flow patterns retrieved from the ascending aorta (A) and main pulmonary artery (B) show the systolic antegrade (s) and diastolic retrograde (D) flows. Simultaneous Doppler from the umbilical artery and vein (C) indicates reverse diastolic arterial wave form (arrows). Absent prograde diastolic flow in umbilical vein (D) (arrows) is better documented separately.

Figure 1 Cross sectional echocardiography showing the origin of the aorta (AO) from the right ventricle (RV). Ventricular septal defect (arrow) was seen close to the aorta (A). Pulmonary artery (PA) also originates from the right ventricle (B) and no semilunar leaflets were visualised. A pericardial effusion (PE) behind the right ventricle was evident.
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Figure 3 Double outlet right ventricle with d-malposed side by side great arteries at necropsy.

Figure 4 Aorta (ao) viewed through right ventricular (rv) approach. Shallow ventriculo-aortic endothelial junction with no evidence of semilunar leaflets (arrows). Coronary ostia in normal position (asterisk).

Figure 5 Pulmonary artery (pa) originates from the right ventricle (rv) with several attachments of dysplastic tricuspid valve (arrow). Only a small endothelial narrowing with aplasia of semilunar leaflets was seen at the ventriculo-pulmonary junction (arrow).

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Congenital absence of the aortic valve is an extremely rare heart malformation. Only seven cases have been reported so far. The absent aortic valve is associated with other structural heart malformations in all instances. In all but one patient the diagnosis was made postnatally. Bierman and colleagues described the antenatal and postnatal echocardiographic features of congenital absence of the aortic valve. They also found retrograde diastolic waveforms in the thoracic aorta, but they did not perform Doppler examination of the umbilical vessels. All seven patients in their report died of heart failure without surgical intervention.
The only report of congenital absence of both the aortic and the pulmonary valve was by Hartwig and associates, from a necropsy examination of two fetuses which died in utero before the 18th week of pregnancy. Both were small for gestational age and had complex congenital defects and additional extracardiac malformations (table).

The authors assumed that both the fetuses died of cardiac failure. Our fetal findings confirm this suggestion. Doppler flow patterns retrieved from the aorta, pulmonary artery, and umbilical vessels showed gross regurgitation throughout both great arteries which eliminated diastolic placental perfusion and resulted in severe heart failure and fetal growth retardation. Severe cardiac failure causing early fetal death may explain why congenital aplasia of both the aortic and the pulmonary valve is not described postnatally.

In the absence of an aortic valve the regurgitation volume is obviously smaller and therefore does not cause early fetal heart failure, although in the only published case of fatally diagnosed aplasia of the aortic valve ascites was noted. In cases with absence of the pulmonary valve, a hypoplastic anulus or right ventricular outflow tract, together with ventricular hypertrophy, will usually reduce retrograde diastolic blood volume. In absent pulmonary valve with arterial duct agenesis retrograde aortic run-off is not possible.

The pathogenesis of the absence of both semilunar valves is unknown. All three cases described were male, as were a further seven reported cases of absent aortic valve. One can speculate that X linked recessive inheritance or other X linked factors may play a role in the maldevelopment of the endocardial cushion tissue at the ventriculo-aortic junction. Unfortunately, none of these 10 cases underwent chromosomal evaluation.

Note added in proof: During the preparation of this paper, we found a report of two other fetuses with absent aortic and pulmonary valve: Mysybar S, et al. Heart Vessels 1994;9:49–55.

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