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Brief Report

Congenital absence of aortic valvar leaflets: a rare variant of the hypoplastic left heart syndrome

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Abstract Congenital absence of aortic valvar leaflets is a rare and fatal variant of the hypoplastic left heart syndrome. We describe a recent patient seen at our institution with this lesion, illustrating a combined echocardiographic and angiographic approach that delineates both anatomy and physiology. The early mortality experienced in previous reports, as well as unsuccessful surgical palliation in our case, should promote further discussion regarding the optimal treatment.

Keywords: Aortic valvar leaflet aplasia; aortic insufficiency; hypoplastic left heart syndrome

HYPOPLASTIC LEFT HEART SYNDROME represents a spectrum of congenital cardiac defects, which predominantly include severe left-sided obstructive lesions such as mitral and aortic valvar atresia. Congenital absence of aortic valvar leaflets is an extremely rare complication of this syndrome.[1–6] We describe here the first neonate with such congenital aplasia of the aortic valvar leaflets in whom, as far as we know, palliative surgery was performed. In addition, we discuss the pathophysiology of congenital absence of the aortic valvar leaflets.

Case report

A twelve hour old neonate delivered at term was referred to our institution after presenting with cyanosis. His cardiovascular examination was remarkable for diminished peripheral and carotid pulses, a hyperdynamic right ventricular impulse, a single second heart sound, a gallop, and a grade II/VI medium frequency systolic murmur heard at the mid-left sternal border. A chest radiogram showed moderate cardiomegaly, with normal pulmonary vascularity. An electrocardiogram was remarkable for right atrial enlargement, severe right ventricular hypertrophy with strain, and poor R wave progression in the precordial leads.

An echocardiogram demonstrated a diminutive left ventricle and mitral valve, with continuous valvar insufficiency. The aortic valvar leaflets were absent, with free aortic valvar insufficiency (Fig. 1). There was a mildly hypoplastic aortic arch, which predominantly filled retrogradely via a large patent arterial duct. The left ventricle was hypertrophied, and numerous small fistulous communications extended from the cavity to the coronary arteries. There was the suggestion of endocardial fibroelastosis within the left ventricle. The proximal left coronary artery was not well seen.

Cardiac catheterization confirmed the echocardiographic findings. Hemodynamic evaluation was remarkable for a markedly elevated left ventricular end-diastolic pressure of 46 mmHg, which was equal to the ascending aortic diastolic pressure. Severe left atrial hypertension did not improve following an atrial balloon septostomy. An ascending aortogram demonstrated free aortic valvar insufficiency, while lack of antegrade flow in the left coronary artery suggested a severe stenosis at its origin. The proximal stem of the left coronary artery, however, filled retrogradely via multiple fistulous communications located within the left ventricular wall (Fig. 2a).

The patient was supported with an infusion of prostanglandins and mechanical ventilation until seven days of age, when he underwent a Norwood

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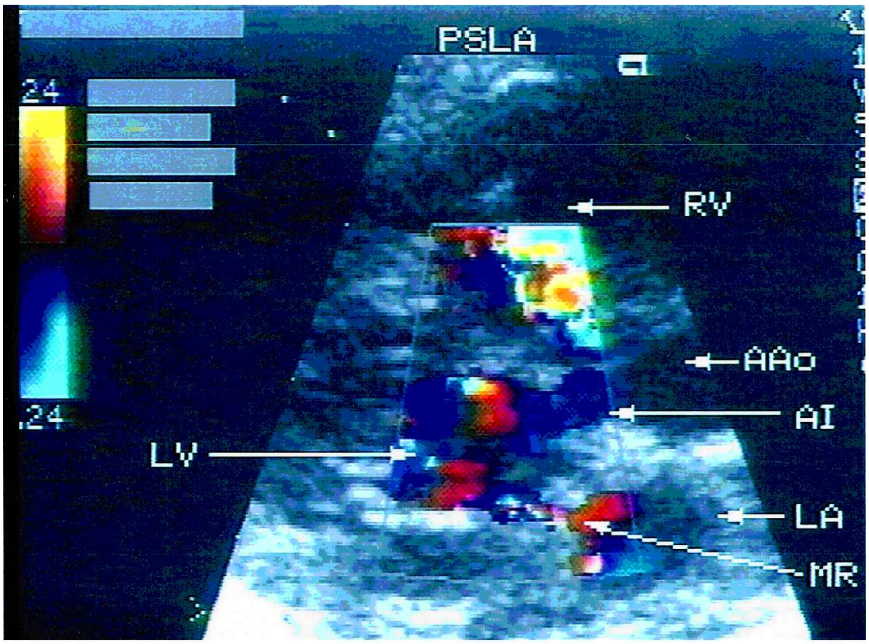

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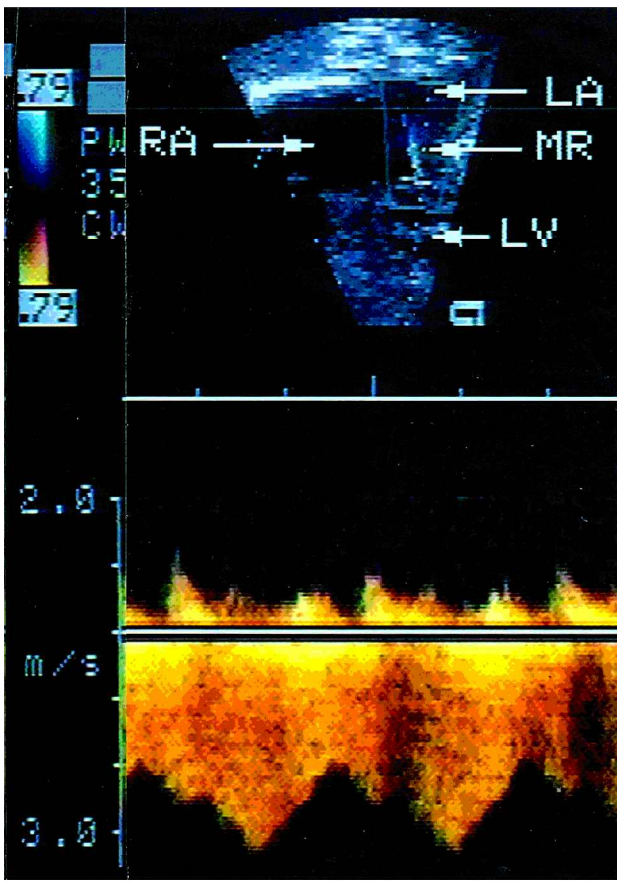

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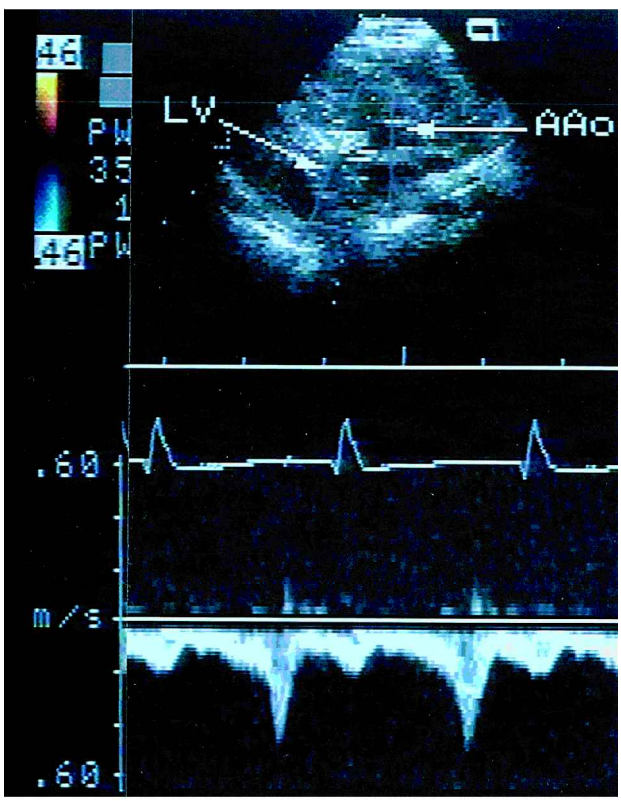

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Figure 1.
Echocardiographic features of aortic valvar leaflet aplasia. (a) This parasternal long axis image demonstrates free aortic valvar insufficiency and mitral valvar insufficiency. The left ventricular chamber is diminutive. (b) Pulse Doppler at the level of the mitral valve in this apical 4-chamber view shows continuous mitral valvar insufficiency. (c) Pulse Doppler within the left ventricular outflow tract shows a paucity of antegrade flow through the left ventricular outflow tract, and free aortic valvar insufficiency with late acceleration during diastole. Abbreviations: AI = aortic insufficiency; AAo = ascending aorta; LA = left atrium; LV = left ventricle; MR = mitral insufficiency; PSLA = parasternal long axis; RA = right atrium; RV = right ventricle.

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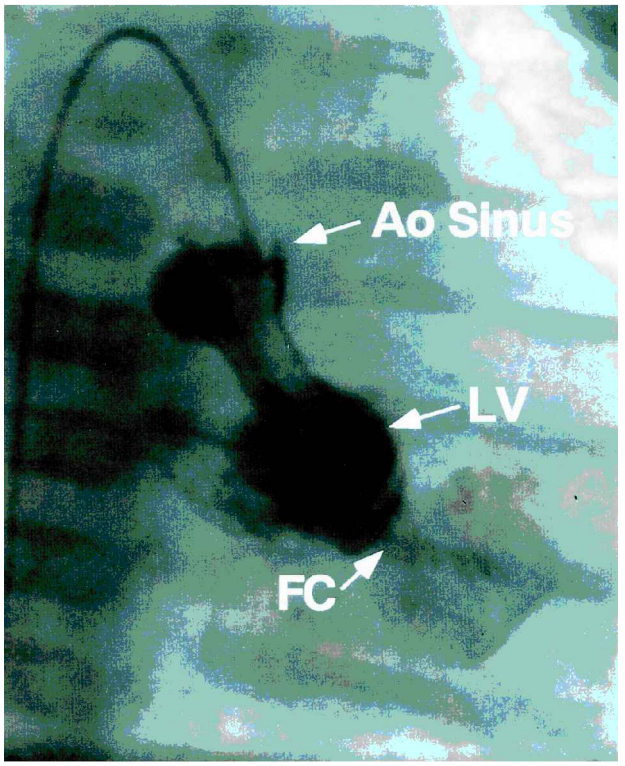

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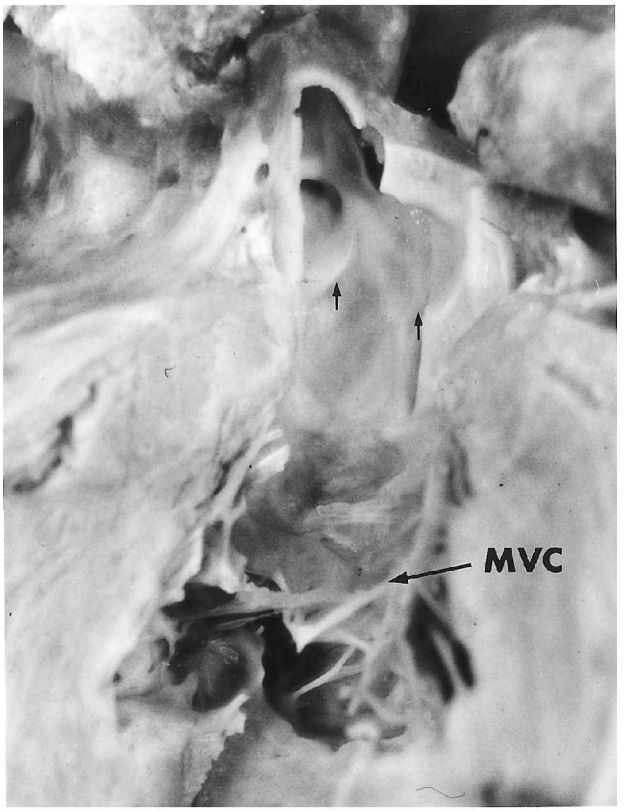

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Figure 2.
Angiographic and anatomic features of agenesis of the aortic valvar leaflets. (a) This ascending aortogram shows absence of aortic valvar leaflets, free aortic valvar insufficiency, a diminutive left ventricular chamber, and left ventricular hypertrophy. The right coronary artery appears normal but there is no filling of the left coronary artery. Subsequent images demonstrated retrograde filling of the left coronary artery via multiple left ventricular fistulous communications (FC) to the coronary arteries. (b) Examination of the opened left ventricle confirms the absence of aortic valvar leaflet tissue, dysplastic mitral valvar cords, a diminutive left ventricular cavity, and marked left ventricular hypertrophy. Abbreviations: Ao Sinus = aortic sinus of Valsalva; FC = fistulous communications; LV = left ventricle; MVC = mitral valve cords.

procedure, placement of a right modified Blalock-Taussig shunt, closure of the mitral valve, and atrial septectomy. Absence of the aortic valvar leaflets was confirmed intraoperatively. The left ventricular outflow tract was left patent to maintain perfusion of the left ventricular myocardium via the fistulous communications with the coronary arteries. Postoperatively, he remained critically ill, with evidence of multisystem organ failure secondary to low cardiac output. Repeat cardiac catheterization on the twentieth postoperative day demonstrated severe biventricular dysfunction, free native aortic valvar insufficiency, no native insufficiency of the pulmonary valve, and no residual mitral valvar insufficiency. Support was withdrawn shortly thereafter.

A limited autopsy confirmed aplasia of the aortic valvar leaflets. There were only tiny intimal ridges at the level of the ventriculo-arterial junction guarding shallow sinuses of Valsalva. The orifice of the left coronary artery was not obstructed anatomically. In addition, there was marked left ventricular hypertrophy, moderate left ventricular endocardial fibroelastosis, and subendocardial fibrosis. The diminutive mitral valve, which was completely oversewn, had several dysplastic cords that attached abnormally to the ventricular septum (Fig. 2b).

Discussion

Congenital absence of aortic valvar leaflets is an extremely rare, and thus far fatal, form of the hypoplastic left heart syndrome (Table 1). The cases described have been associated with other hemodynamically significant cardiac lesions. The rare incidence of this condition among newborns may be, in part, a consequence of poorly tolerated fetal hemodynamics, resulting in spontaneous abortion. Recent reports of fetal death secondary to absence of

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Table 1. Summary of cases.

Case	Presentation	Associated anomalies	Age at death	Reference
1	Cyanosis/ Respiratory distress	DORV, hypoplastic MV, dysplastic TV, "diminutive LV cavity"	2 days	1
2	Nonimmune hydrops, Respiratory distress	DORV, complete AVSD, small LV with markedly thickened free wall, hypoplastic MV, polysplenia	20 hrs	2
3	Cyanosis/tachypnea	Hypoplastic MV, hypoplastic LV, EFE, hypoplastic AAo	8 hrs	6
4	Cyanosis/ Respiratory distress	MV atresia, severe LV, EFE, "bizarre trabeculation" of hypertrophied LV, dysplastic TV & PV	4 hrs	4
5	Cyanosis	Hypoplastic LV with thickened walls, hypoplastic MV with insufficiency	16 hrs	5
6	Cyanosis	EFE, "spongy architecture" of myocardium, pulmonary venous stenosis, abnormal MV	20 hrs	3
7	Cyanosis/ Respiratory distress	Hypoplastic LV, dysplastic MV with severe Insufficiency, EFE, hypoplastic LA, PDA, Cath: LV =AAo pressures = 45/25 mmHg	24 hrs	3
8	Cyanosis	MV atresia, Ebstein's malformation of TV, TAPVC, "spongy" LV wall, PAD, Cath: LV = AAo pressures = 60/40 mmHg	6 days	3
9	Cyanosis	Diminutive MV with continuous MR hypoplastic LV, coronary fistulous communications, EFE, PAD, dysplastic TV, Cath: LV = AAo pressures = 58/46 mmHg	28 days	our case

Abbreviations: AAo = ascending aorta; AVSD = atrioventricular septal defect; Cath = cardiac catheterization; DORV = double outlet right ventricle; EFE = endocardial fibroelastosis; LV = left ventricle; MR = mitral valvar insufficiency; MV = mitral valve; PAD = patent arterial duct; PV = pulmonary valve; TAPVC = totally anomalous pulmonary venous connection; TV = tricuspid valve

the leaflets of both the aortic and pulmonary valves may represent an extreme variant.[7–8]

The etiology of congenital absence of aortic valvar leaflets remains unknown. Proposed etiologic factors include a deficiency of primitive valvar tissue, abnormal ventriculo-arterial septation, and early destruction of the valvar leaflets. There may be a genetic component, as all reported cases are males.[3]

The pathophysiology of the condition is intriguing. As the leaflets of the aortic valve are absent, free aortic valvar insufficiency leads to a persistent severely elevated left ventricular end-diastolic pressure. In fact, the left ventricular end-diastolic pressure has been equal to the ascending aortic diastolic pressure in all affected patients in whom a cardiac catheterization was performed. Consequently, myocardial perfusion is compromised secondary to decreased coronary arterial perfusion pressure. The elevated left ventricular end-diastolic pressure may also promote the development of fistulous communications with the coronary arteries. The abnormal flow seen in the proximal left coronary artery in our patient represented a functional, rather than anatomic, obstruction of coronary blood flow. Increased myocardial oxygen consumption due to left ventricular hypertrophy, in combination with compromised coronary perfusion, places the myocardium at risk for ischaemic injury.

The elevated left ventricular end-diastolic pressure may also inhibit normal development of the mitral valve by hampering the opening of its leaflets. The mitral valve has been either atretic or hypoplastic in all reported cases with congenital absence of the aortic valvar leaflets.[1–6] Moreover, the finding of continuous mitral valvar insufficiency in our patient, which has not been previously associated with congenital absence of aortic valvar leaflets, reflects the overwhelming hemodynamic stress on the mitral valve caused by the elevated left ventricular end-diastolic pressure. Abnormal development of the tendinous cords supporting the mitral valve may result from altered flow of blood within the left ventricle secondary to combined aortic and mitral valvar insufficiency. Thus, the marked elevated left ventricular end-diastolic pressure occurring secondary to free aortic valvar insufficiency results in a cascade of developmental perturbations, including altered coronary arterial perfusion, reduced myocardial perfusion, and diminished mitral valvar and left ventricular growth.

Surgical palliation may be considered in the management of neonates with congenital absence of aortic valvar leaflets. Meticulous delineation of associated lesions, using echocardiography and cardiac catheterization, is essential for guiding appropriate surgical management. It has yet to be determined, however, whether surgical intervention

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will alter the dismal prognosis among these patients. Cardiac transplantation may not be a feasible option, as the hemodynamics would likely be poorly tolerated while waiting for an available donor. Improved outcome may necessitate prenatal diagnosis and fetal intervention, such as ligation of the left ventricular outflow tract, in order to minimize the development of myocardial ischemia and dysfunction. When a neonate is diagnosed with hypoplastic left heart syndrome, the presence of continuous mitral valvar insufficiency, fistulous connections between the left ventricle and the coronary arteries, or severe aortic valvar insufficiency, should prompt further evaluation to rule out congenital absence of the aortic valvar leaflets.

References

1. Toews WH, Lortscher RH, Kelminson LL. Double outlet right ventricle with absent aortic valve. Chest 1975; 68: 381–382.
2. Bierman FZ, Yeh MN, Swersky S, Martin E, Wigger JH, Fox H. Absence of the aortic valve: antenatal and postnatal two-dimensional and doppler echocardiographic features. J Am Coll Cardiol 1984; 3: 833–837.
3. Rossi MB, Ho SY, Tasker RC. Absent aortic valve leaflets. Int J Cardiol 1986; 11: 235–237.
4. Niwa K, Ikeda F, Miyamoto H, Nakajima H, Ando M. Absent aortic valve with normally related great arteries. Heart Vessels 1987; 3: 104–107.
5. Lin AE, Chin AJ. Absent aortic valve: a complex anomaly. Pediatr Cardiol 1990; 11: 195–198.
6. Parikh SR, Hurwitz RA, Caldwell RL, Waller B. Absent aortic valve in hypoplastic left heart syndrome. Am Heart J 1990; 119: 977–978.
7. Hartwig NG, Vermeij-Keers C, De Vries HE, Gittenberger-De Groot, AC. Aplasia of semilunar valve leaflets: Two case reports and developmental aspects. Pediatr Cardiol 1991; 12: 114–117.
8. Marek J, Skovranek J, Povysilova V. Congenital absence of aortic and pulmonic valve in a fetus with severe heart failure. Heart 1996; 75: 98–100.