Hypoplastic Left Heart Syndrome: Report of a Unique Survivor

MARTIN EHRLICH, MD, FREDRICK Z. BIERMAN, MD, FACC, KENT ELLIS, MD, WELTON M. GERSONY, MD, FACC

New York, New York

A remarkable patient is described, a child who has survived until the age of 7 years with hypoplastic left heart syndrome (mitral and aortic atresia) without surgical intervention. The child has led an active, normal life and, aside from minimal cyanosis, has remained asymptomatic. The unique clinical course for this patient is the result of a number of favorable hemodynamic factors that have not been previously reported in an individual patient with hypoplastic left heart syndrome and intact ventricular septum: 1) widely patent ductus arteriosus, 2) adequate retrograde coronary flow, 3) unrestricted pulmonary venous return, and 4) absence of significant vascular obstructive disease. This documentation of long-term survival in a child without surgical treatment for mitral and aortic atresia suggests that successful early palliative treatment for infants with this syndrome could also result in a favorable prognosis.

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Hypoplastic left heart syndrome refers to a closely related group of congenital cardiac anomalies characterized by underdevelopment of the left heart chambers, atresia or severe stenosis of the aortic or mitral orifice, or both, and hypoplasia of the ascending aorta (1,2). Most infants with hypoplastic left heart syndrome are well developed, have a normal birth weight and a very low incidence of extracardiac abnormalities (3). Death invariably occurs within the first month of life.

For more than a decade, surgical palliation of infants with these anomalies has been attempted (4-6). However, because survival after infancy has yet to be reported, the question as to whether a favorable prognosis is possible has remained unanswered. The longest reported survival time without operation is 3.5 years (7). In this report we present detailed noninvasive, hemodynamic and angiographic data on a patient with documented hypoplastic left heart syndrome who is alive and well without surgical intervention at 7 years of age.

Case Report

Clinical presentation. The patient is a white girl, the product of an uncomplicated pregnancy. Respiratory distress was noted 1 hour after birth and a tracheoesophageal fistula was diagnosed and successfully repaired. In the second week of life, cyanosis during crying and an intermittent gallop rhythm led to the suspicion of heart disease. The chest X-ray film revealed cardiomegaly and increased pulmonary vascular markings. The electrocardiogram showed a right superior axis, right atrial enlargement and right ventricular hypertrophy. Cardiac catheterization was performed at 2.5 weeks of age and the diagnosis of hypoplastic left heart syndrome was made. Although the parents were informed that the prognosis was poor, the child did well on digoxin and diuretic therapy, in addition to being fed through a gastrostomy tube for the first year of life. Repeat catheterization, performed at 1 year of age, revealed a hypoplastic left ventricle with mitral and aortic valve atresia, a small ascending aorta, a large patent ductus arteriosus and a descending aorta of normal caliber.

Over the succeeding 6 years, the child has remained in stable condition with only mild cyanosis and little, if any, exercise intolerance. During the first 2 years of life she was admitted to the hospital several times for respiratory infections and during these admissions she was treated with digoxin and furosemide to control symptoms attributed to congestive heart failure. However, since March 1980 (age 3 years), she has had no hospital admissions.

Initial physical examination at Columbia Presbyterian Medical Center on September 12, 1982 at 5.5 years of age revealed a well developed, slightly cyanotic girl with minimal clubbing of the fingers. The weight was 16.5 kg (12th percentile for age) and the height was 102 cm (<5th percentile). Blood pressure in the right arm and left leg was 90/60 mm Hg. There was no edema, the lungs were clear...
and the liver was not enlarged. Pulses were full and equal over the four limbs. The precordium was active with a prominent cardiac impulse. The second heart sound was single at the left upper sternal border, radiating over the entire precordium and back. There were no murmurs.

Laboratory data included hemoglobin, 14.9 g/100 ml and hematocrit, 42.2%. The electrocardiogram showed sinus rhythm with a mean frontal axis of +100°, left atrial enlargement and right ventricular hypertrophy. The ST-T wave segments were normal and the chest X-ray film showed cardiac enlargement and prominent pulmonary vascularity (Fig. 1).

Cardiac ultrasound. Two-dimensional echocardiography was performed using an Advanced Technologies Laboratory Mark 500 cardiac imager with a 3.0 MHz in-line transducer. Subxiphoid imaging was recorded in atrial and ventricular long-axis projections. Atrial long-axis projections demonstrated dilation of the right atrium and tricuspid valve anulus with a qualitatively normal-sized “left atrium” and atretic mitral valve (Fig. 2). The right upper pulmonary vein appeared to communicate with the left atrium at its septal/posterior wall junction. No interatrial communication was demonstrated. The left atrial appendage was not visualized. The coronary sinus was not dilated.

Ventricular short-axis projections revealed a posterior, hypoplastic morphologic left ventricle and anterior, dilated morphologic right ventricle. The ascending aorta terminated proximally as the coronary arteries with no semilunar valve present. A dilated right-sided superior vena cava was adjacent to a hypoplastic ascending aorta. Atrial and ventricular short-axis projections revealed similar anatomy as well as displaying continuity of the main pulmonary artery with the descending aorta by way of a large, patent ductus arteriosus.

Gated pulsed Doppler ultrasound examination of the thoracoabdominal aorta revealed pandiastolic reversal of blood flow, suggesting significant retrograde flow to the pulmonary circulation during diastole (Fig. 3).

Cardiac catheterization. A common pulmonary vein was entered by way of the left innominate vein. The right
ventricular pressure was systemic with normal end-diastolic pressure. The descending aorta was catheterized by way of the pulmonary artery and ductus arteriosus. The pulmonary artery and descending aortic blood pressures were similar: 84/47 (mean 62) and 86/49 mm Hg (mean 64), respectively; the mean pulmonary venous wedge pressure was 20 mm Hg with a right ventricular end-diastolic pressure of 4 mm Hg. The pulmonary blood flow was 12.2 liters/min per m² with a pulmonary to systemic (Qp/Qs) flow ratio of 2.4:1. Systemic arterial saturation was 91%, reflecting the markedly increased pulmonary blood flow. Despite pulmonary artery hypertension at systemic levels, the large pulmonary blood flow resulted in a calculated pulmonary vascular resistance that was only mildly elevated (3.4 U/m²).

Angiocardiographic studies were performed with selective injections into the confluence of the right pulmonary veins, right ventricle and distal aortic arch. The confluence of the right pulmonary veins in the frontal view resembled the left atrium in shape and position, but no atrial appendage was visualized. Joined by the left pulmonary veins, the entire pulmonary venous return flowed in retrograde fashion along a large ascending levoatrial-cardinal vein to join the dilated left innominate vein. The latter continued medially to join the dilated right superior cava, resulting in unrestrictive, supracardiac total anomalous pulmonary venous return. (Although the dilated region may have represented the left atrium with mitral atresia, it could also have represented the primitive common pulmonary vein with atresia of its connection to the left atrium.)

The right ventriculogram demonstrated a large right ventricle supplying moderate-sized pulmonary arteries. Selective injection into the main pulmonary artery demonstrated

Figure 4. Selective contrast injection into the main pulmonary artery, demonstrating its confluence across the ductus arteriosus with the descending aorta. A flow dividing shelf (arrow) is evident on the posterior aortic wall opposite the distal ostium of the ductus arteriosus.

Figure 5. Aortogram in frontal (A) and lateral (B) projections. A displays filling of the transverse aortic arch vessels and aberrant origin of the right subclavian artery. Retrograde flow in both views opacifies the classical hypoplastic ascending aorta (arrow) and more proximal bulbous, atretic aortic root. The coronary arteries arise from the atretic proximal aorta (not demonstrated). Opacification of the distal arch is interrupted by nonopaque blood entering the aorta across the large patent ductus arteriosus during ventricular systole.
its continuity with the descending aorta through the ductus arteriosus. A flow dividing shelf was noted opposite the entry of the patent ductus into the aorta (Fig. 4). No left ventricle was demonstrated.

The selective retrograde aortogram in the distal transverse arch showed the origin of the carotid and left subclavian arteries and retrograde filling of the hypoplastic ascending aorta (Fig. 5). The right subclavian artery was noted arising aberrantly as the last brachiocephalic branch. The proximal aorta had the classical appearance of congenital aterias with the slightly bulbous origin of the coronary arteries.

In summary, the angiocardio graphic data are diagnostic of hypoplastic left heart with mitral and aortic atresia associated with total anomalous pulmonary venous connection of the supracardiac type. Systemic arterial blood flow is maintained across a patent ductus arteriosus with diastolic reversal of flow consistent with the calculated low pulmonary vascular resistance.

Discussion

Factors in mortality and survival. The combination of defects that make up hypoplastic left heart syndrome account for more than 25% of all cardiac deaths in the first week of life (2). Most infants born with these defects die within the neonatal period (an average lifespan of 4 to 23 days has been reported [7]). A number of factors alone or in combination lead to the early death of infants with hypoplastic left heart syndrome. In most cases, death occurs as a result of narrowing of the ductus arteriosus, which causes decreased systemic perfusion. If the ductus remains patent, death in the neonatal period results from congestive heart failure with pulmonary congestion. This may be due to massive pulmonary blood flow and myocardial preload, obstruction of pulmonary venous return (restrictive foramen ovale), insufficient coronary blood flow due to reduced ascending aorta caliber or a combination of these factors.

For our patient to have survived without operation, several unique anatomic and physiologic adjustments were required. First, the ductus remained widely patent to maintain systemic blood flow through the right ventricle. Second, sufficient coronary blood flow was provided by retrograde flow through the hypoplastic ascending aorta into the coronary vessels. Third, although the atrial septum is intact, unobstructed pulmonary venous return occurred through an anomalous system of pulmonary venous connections. Finally, and perhaps most surprising, there was no severe pulmonary vascular disease despite long-standing systemic pressure in the pulmonary artery. The systemic arterial saturation and angiocardio graphic data, indicating excellent pulmonary blood flow retrograde across the ductus arteriosus during diastole, are consistent with low calculated pulmonary vascular resistance (3.4 U/m²).

In a previously reported case (7) of a patient with aortic atresia who died at the age of 3 1/2 years with pulmonary vascular disease, an unrestrictive atrial septal defect provided decompression of the left atrium and pulmonary veins. Other reports (8,9) have also attributed long survival in aortic atresia to a large-sized septal defect, but no cases have been described that are similar to our own.

Surgical correction. Recently, Norwood et al. (10) reported early success with a two-stage palliative approach for hypoplastic left heart syndrome. The initial procedure consists of reconstruction of the hypoplastic ascending aorta and aortic arch by anastomosis of a transected proximal pulmonary artery to the ascending aorta and aortic arch, creating unobstructed anterograde flow from the right ventricle through the aorta and improved access to the coronary arteries. Pulmonary blood flow is then provided by a conventional Blalock-Taussig shunt. The second stage consists of construction of an interatrial baffle providing continuity between the left atrium and right ventricle through the atrial septal defect and tricuspid valve, isolating the right atrium. Pulmonary artery circulation is established by anastomosis of the right atrium to the pulmonary artery according to the Fontan principle with closure of the Blalock-Taussig shunt. Two infants with aortic atresia are reported to be alive and well after the second stage procedure, whereas 21 infants have survived the first stage and are awaiting the Fontan operation (Lang P, personal communication, 1985). A final judgment regarding the efficacy of this operation must await long-term follow-up study of a larger group of survivors. However, the prolonged and relatively uneventful clinical course of our patient suggests that, with optimal hemodynamic adjustments, patients with hypoplastic left heart syndrome may indeed survive for a long time and lead a life of good quality. Aggressive attempts at surgical palliation in the neonatal period and after appear to be justified.

We express our appreciation for the referral of this unique case and recognition of the care received by the patient under the direction of James Sutherland, MD, Letterman Army Medical Center, Presidio of San Francisco, California

References

