Case Reports

Hypoplastic left heart syndrome and valvular pulmonary stenosis

A rare association that limits the management options

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ABSTRACT

A term baby had cyanosis 2 hours after delivery; echocardiography showed hypoplastic left heart syndrome (HLHS) and valvular pulmonary stenosis (PS). The opinions of the cardiac centers in Saudi Arabia were taken; however, the baby was not accepted probably because the associated PS made him a poor candidate for a Norwood procedure and because cardiac transplant is not available for infants in Saudi Arabia. He died after 15 days. This mother should have fetal echocardiography in future pregnancies, and if HLHS is suspected she might be advised to seek medical advice antenatally at highly specialized cardiac centers abroad, as delivery there may increase the chance of surgical intervention.

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We describe a rare association of hypoplastic left heart syndrome (HLHS) with significant valvular pulmonary stenosis (PS). The latter made the option of Norwood procedure unsuitable; the surgical management is cardiac transplant which is not available for infants in Saudi Arabia. The aim of this paper is to draw attention to this extremely rare association and to discuss the management of future pregnancies for this couple in view of the recurrence risk and the available options in Saudi Arabia.

Case Report. This baby boy was born at term to a healthy couple, a 22-year old gravida 3 para 2 mother and a 29-year old father. The mother had a regular antenatal care during which oligohydramnios and intrauterine growth retardation (IUGR) were detected. No congenital malformation was diagnosed antenatally. There is no history of maternal diabetes, exposure to any radiation or infection; the mother was on folic acid and iron tablets only. There is no history of congenital heart disease (CHD) in the family. The baby was delivered normally at another hospital. His Apgar scores were 5, 7 and 9 at 1, 5 and 10 minutes respectively. Weight was 2.2 Kg; length was 43 cm (both were below 5th percentile). Head circumference was 33 cm (at 5th percentile). Two hours after delivery the baby was noted to have mild cyanosis, and his oxygen saturation was 85%. Prostaglandin E1 infusion was started and the boy was referred to Aseer Central Hospital (ACH) at 18 hour of age as a case of HLHS; he was put on mechanical ventilation shortly after his arrival to our neonatal intensive care unit. Physical examination in ACH revealed O_2 saturation of 90%, a heart rate of 160 beats per minute, blood pressure in the right arm and in the lower limb of 50/35 mm Hg and 58/40 mm Hg respectively, weak peripheral pulses in the upper and lower parts of the body, hyperactive right ventricle (RV), normal first heart sound, single soft second heart sound, systolic ejection click, systolic ejection murmur grade 3/6 at upper left sternal border, absence of diastolic murmur, and normal respiratory and abdominal examination. There was no gallop, edema, dysmorphic features or obvious anomalies. Blood gases revealed pH 7.35, PCO_2 33 mm Hg, bicarbonate 17 mEq/L, BE -8, and PO_2 49 mm Hg. Serum creatinine was 1.9 mg/dl. Chest x-ray showed normal heart size and normal lung

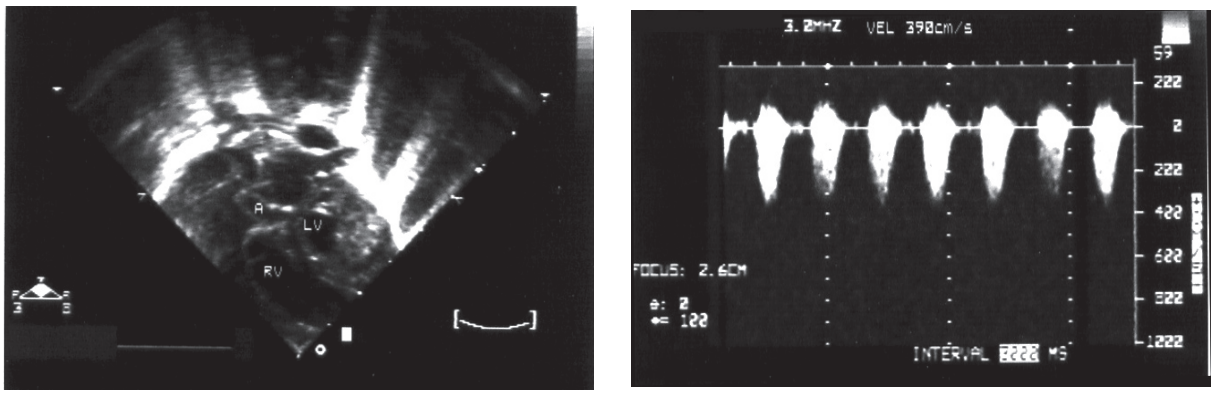

Figure 1 - a) Echocardiographic apical view shows hypoplastic muscle-bound left ventricle (LV), hypoplastic ascending aorta (A), and aortic atresia. **b)** Doppler signal across the pulmonary valve indicating a peak velocity of 3.9 m/sec (peak gradient 60 mmHg).

fields. ECG revealed normal sinus rhythm, QRS axis of +190°, right atrial (RA) enlargement, RV hypertrophy and no ischemic changes. Echocardiography (**Figure 1**) showed: situs solitus, Levocardia, normal pulmonary venous drainage, persistent left superior vena cava draining into coronary sinus, mitral valve atresia, hypoplastic left ventricle (LV), aortic valve atresia, hypoplastic aortic root (3 mm), hypoplastic ascending aorta (4 mm), mild narrowing of the descending aorta below the left subclavian artery (but no gradient), small secundum atrial septal defect (3 mm) with left to right shunt, intact interventricular septum, dilated RA and RV, moderately thickened pulmonary valve with moderately severe pulmonary stenosis (peak gradient 60 mm Hg), left aortic arch and large patent ductus arteriosus. At 3 days of age while the boy was on PGE1 infusion (0.1 µg/kg/min) and intravenous Lasix (1.5 mg/Kg/day), he developed congestive heart failure (CHF) with generalized edema, hepatomegaly (4 cm below right costal margin, span of 7 cm), escalation of creatinine to 2.2 mg/dl and electrocardiographic evidence of ischemia. The PGE1 infusion was increased to 0.15 µg/kg/min resulting in a slight improvement. Ampicillin and Cefotaxime, which had been started on admission, were discontinued after 7 days as all cultures were sterile. On day 10, the baby developed hypotension and looked septic, and therefore dopamine and adjusted doses of vancomycin and meropenem were started. Unfortunately, the condition deteriorated and the baby died at 15 days of age. The repeated blood and urine cultures were sterile. Of note, this patient was not accepted by any of the leading cardiac centers in the country because of the complexity of the cardiac lesions.

Discussion. Hypoplastic left heart syndrome is fortunately uncommon.[1] The combination of HLHS and valvular PS is extremely rare; a few patients with this combination have been described.[2] The frequency of valvular PS among 230 autopsy cases of HLHS was 0.4%.[3] The surgical options for HLHS include Norwood reconstruction followed by a modified Fontan procedure, and cardiac transplantation either primarily or following Norwood reconstruction. The third option is a compassionate care, namely, no surgical intervention. The informed parents may ask for termination of pregnancy (in some countries) when the diagnosis is confirmed antenatally, or opt to do nothing when the diagnosis is made after birth; this last option seems reasonable under certain circumstances as the long-term prognosis following Fontan's procedure for HLHS is not very clear, and the overall mid-term survival is only 39% 10 years postoperatively.[4] As for the transplantation, it is known that 19-25% of these patients die while waiting for donors[5,6] and only 72% of the survivors remain alive 5 years postoperatively.[6] In Kingdom of Saudi Arabia, a "pediatric cardiac transplantation program" does not exist, and therefore affected patients may be candidates for the Norwood procedure as an initial palliation. Basically, this procedure uses the patient pulmonary valve as "neoaortic" valve, and the pulmonary trunk to widen the ascending aorta; the blood flow to the distal pulmonary arteries, which are surgically disconnected from the pulmonary trunk, is maintained through Blalock-Taussig shunt. This procedure, which is scarcely carried out in Saudi Arabia, was not suitable for our patient as it would have left him with a systemic RV and a stenosed "neoaortic" valve, which may cause progressive deterioration of this systemic ventricle. Ballon dilatation could be carried out but may leave

residual stenosis or regurgitation of the dysplastic valve; these are relative contraindication to Norwood/ Fontan reconstruction. The optimal management is transplantation; balloon dilatation of the pulmonary valve and atrial septostomy may be needed to improve the hemodynamics[2] while waiting for transplantation.[2] The fact that pediatric cardiac transplantation was not available left us with "do nothing" approach. The parents need counseling for future pregnancy as the risk of recurrence is relatively high. The recurrence risk of CHD in the siblings has previously been estimated to be 2%; however, in some families the recurrence is high, approaching 20%, and the predominant types of CHDs among them are left-sided obstructive lesions.[7] Hypoplastic left heart syndrome may rarely be inherited as an autosomal recessive condition.[8] We would advise the mother to have a fetal echocardiography in the second trimester by an expert as the antenatal diagnosis of HLHS would allow the willing families to seek medical advice antenatally at highly specialized centers abroad; this entails executing the delivery near the specialized center as this will at least optimize the preoperative condition of the baby and increase the chances of early Norwood surgery and transplantation. A less practical option is fetal intervention by balloon dilatation of a severely stenosed or atretic aortic valve as this may prevent its progression to HLHS increasing the future chance of biventricular repair;[9] this may be attempted if fetal echocardiography shows signs predictive of progression to HLHS namely, reversed flow in the transverse aortic arch and foramen ovale, and a monophasic mitral inflow.[10] Fetal intervention for HLHS is still experimental; in one study there was a 20% fetal death and at least 15% chances of preventing HLHS.[9] Further studies are needed to improve the outcome.

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