

Prenatal Imaging of Congenital Fetal Anomalies: A Case Series.

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ABSTRACT

Hypoplastic left heart syndrome (HLHS) comprises 2 to 3 percent of all congenital heart disease. HLHS is uniformly lethal if left untreated; however, there is currently an estimated 70 percent 5-year survival rate for infants who undergo surgical correction. The hypoplastic left heart syndrome encompasses a spectrum of cardiac malformations that are characterized by significant underdevelopment of the components of the left heart and the aorta, including the left ventricular cavity and mass. Its main anatomical characteristics include mitral stenosis or atresia, aortic stenosis or atresia, and hypoplasia of left ventricle. The apex of the heart is usually formed by the right ventricle. The right ventricle is the single functional ventricle maintaining fetal circulation in these patients. After births, due to decreased pulmonary vascular resistance and closure of the ductus arteriosus, infants with HLHS develop a constellation of symptoms that include cyanosis, dyspnea, heart failure, hypoxemia and acidosis. Surgery is the only effective treatment. Unfortunately, even when treated, this disease is associated with significant morbidity including an increased risk of thrombotic complications, decrease in exercise tolerance, and neuro-developmental impairment. Continuous intravenous prostaglandin E1 is used to maintain a PDA to allow adequate systemic perfusion. Nitrogen may be used in some centers to decrease inspired oxygen which increases pulmonary arterial resistance. This therapy encourages systemic rather than pulmonary blood flow and enhances systemic perfusion. Double outlet right ventricle (DORV) is a type of ventriculoarterial connection in which both the aorta and pulmonary artery arise entirely or predominantly from the right ventricle (RV). The only outlet from the left ventricle (LV) is a ventricular septal defect (VSD). DORV is usually associated with concordant atrioventricular connections with the right atrium draining into the RV and the left atrium draining into the LV. DORV is almost always associated with a VSD and occasionally with an atrial septal defect. Transposition of great arteries is characterized by both the outflows being parallel to each other at the origin. On color Doppler, they show similar color flows, indicating that the direction of flow in both the great vessels is same. Most atrial septal defects involve either the septum primum or septum secundum. Primum atrial septal defect is the simplest form of the atrioventricular septal defect. Secundum atrial septal defect which is the commonest are usually isolated septal defects, but may be related to other cardiac anomalies like mitral, tricuspid, pulmonary or aortic atresia and are occasionally found as part of syndromes like Holt-Oram syndrome.

Keywords: HLHS, congenital, cardiac, prostaglandin, prenatal diagnosis, DORV, TGA, ASD.

INTRODUCTION

Hypoplastic Left Heart Syndrome (HLHS) is rare congenital heart defect characterized by faulty or absent development of the left ventricle. Normally, oxygenated blood returns from the lungs to the left side of the heart to be pumped systemically; however, in HLHS the right ventricle must support both pulmonary and systemic circulations.^[1]

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There are likely several inciting mechanisms resulting in the underdevelopment of the left ventricle (LV). In fetal life, the LV is predominantly

filled by flow through the foramen vale and any perturbation of flow into or out of the LV may result in growth impairment. It has been observed that the fetus with HLHS has a smaller foramen ovale than the fetus with a normal heart. In addition, there is a known association between HLHS and an anatomic abnormality of the atrial septum, namely posterior deviation of the septum primum. In this anomaly, the superior edge of the septum primum is deviated posterior and leftward, attaching anomalously to the left atrial wall, restricting atrial level shunting.^[2] An intact atrial septum in association with HLHS has also been observed in utero; often, there is a small communication early in gestation that closes over time. This diagnosis carries a very poor prognosis. Hypoplastic left heart syndrome (HLHS) can be easily recognized on prenatal ultrasound and is

proportionately one of the most common serious cardiac defects diagnosed prenatally. The standard “4-chamber cardiac view” used for screening of congenital heart disease demonstrates either a small left side or an echogenic left ventricle from endocardial fibroelastosis.^[3] Prenatal diagnosis of HLHS, however, has not been demonstrated to improve surgical outcomes. Prenatal diagnosis of HLHS affords the opportunity for counselling and perinatal planning. For patients in whom surgical palliation is elected, prenatal diagnosis provides an opportunity to avoid the preoperative hemodynamic and metabolic insult so frequently seen in those diagnosed postnatally. Prenatal diagnosis of HLHS affords time for physicians both to counsel parents and to plan perinatal management. Without the benefit of prenatal diagnosis, most infants with HLHS are born outside of tertiary care centers, which delays diagnosis and appropriate resuscitation. The delay in diagnosis, in turn, may lead to systemic hypoperfusion, shock, and multiorgan damage, which can diminish chances for surgical success and lead to long-term sequelae.^[4]

Double outlet right ventricle is characterized by the aorta and the pulmonary trunk both arising from the right ventricle. This anomaly accounts for 2% of live births with a congenital heart defect.^[5] Because of its diverse morphology, DORV does not have a typical ultrasound appearance. The 4 chamber view may be abnormal in fetuses with a large VSD, a single ventricle or coexisting multiple valve atresia.^[6] Often there is a conspicuous malposition of the great vessels with a discrepancy in the calibers of the aorta and the pulmonary trunk. The origin of the two great vessels from the right ventricle is defined more clearly with color Doppler.^[7] Transposition of the great arteries is an abnormality in which aorta arises entirely or in large part from the right ventricle and the pulmonary artery arises from the left ventricle. Clue to the diagnosis is the demonstration that the two great vessels do not cross but arise parallel from the base of the heart.^[8]

Atrial septal defect is very difficult to diagnose prenatally. 4 types of defects have been identified which include a sinus venosus defect, an ostium secundum defect, a septum primum defect and a common atrium.^[9] When a large septum secundum defect is suspected, it can be confirmed with color Doppler, which demonstrates a left to right shunt in addition to the physiologic right to left shunt. With a septum primum defect, the examiner can use color flow to confirm his suspicion. Color Doppler shows any communication that exists between the right atrium and left ventricle or between the left atrium and the right ventricle.

CASE REPORT

Case 1: A Baby boy delivered normally at term weighing 2430 gm, length 52 cm, presented on 3rd

day of life with respiratory distress. On Examination there was central cyanosis and pallor with cold extremities, weak peripheral pulses, poor perfusion in shock. Auscultation revealed loud S2. Due to progressive deterioration, mechanical ventilation was provided with inotropic support. However, baby could not be revived. On Ultrasonography 29 weeks single, live intrauterine pregnancy with dilated fetal aorta. Fetal echocardiography showed heart more in left hemi thorax, two chambered heart—morphological RA and RV. LA severely hypoplastic to atretic and LV atretic. Large RA and large RV, single pulmonary vein draining into almost atretic LA. Large pulmonary trunk arising from the RV, dividing into RPA and LPA; ductus arch large and continuing into descending thoracic aorta. Ascending aortic-atretic. Postnatal echocardiography done on 1 day of life which revealed Mitral atresia, aortic atresia, Rudimentary and Hypoplastic LV. Small, restrictive PFO measuring 3.2 mm, left to right shunt with severe PH, Hypoplastic arch, Pulmonary artery largely continuing as descending aorta suggesting Hypoplastic left heart syndrome.



Figure 1 and 2: Antenatal ultrasound images demonstrating the size discrepancy between the right and left ventricles—suggesting a diagnosis of hypoplastic left heart syndrome (HLHS).

Case 2: A male infant aged 8 days and weighing 3 Kg was presented with tachypnea and cyanosis. His heart auscultation revealed a continuous murmur (+++/4+) on the upper left sternal border and a

systolic murmur of regurgitation (++/ 4+) on the lower left sternal border. His second heart sound had a normal intensity and showed no splitting. His pulses were symmetrical and had a normal amplitude. His electrocardiogram electro-cardiogram showed right ventricular hypertrophy. His chest X-ray revealed an enlarged cardiac silhouette due to enlarged cavities, flat pulmonary trunk segment and increased pulmonary vasculature. Fetal echocardiography showed the following findings: normal pulmonary venous drainage and atrioventricular concordance; doubly committed VSD remote from the great arteries; atrioventricular valves with normal echoes and mobility. A diagnosis of DORV with TGA was made.

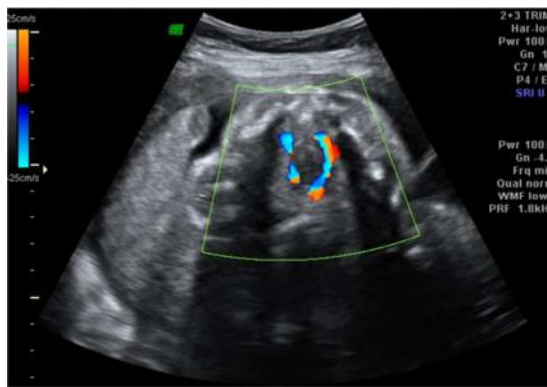


Figure 3 and 4: Antenatal ultrasound images demonstrating two great vessels that do not cross but arise parallel from the base of the heart – suggesting transposition of great arteries (TGA) and double outlet right ventricle (DORV) with common origin of the aorta and the pulmonary artery from the right ventricle.

Case 3: A male neonate aged 28 days and weighing 2800 gm who is currently asymptomatic. Fetal ultrasonography revealed a defect in the central portion of the septum secundum in the vicinity of the foramen ovale of the fetal heart. On M-mode fetal echocardiography, an abnormal oscillatory pattern of the foramen ovale flap motion was detected which gave us the clue to the existence of septum secundum type of atrial septal defect as at birth a

patent foramen ovale also could be mistaken for an ostium secundum atrial septal defect.



Figure 5 and 6: Antenatal ultrasound images of the heart showing an atrial septal defect (ASD) of septum secundum type.

DISCUSSION

In HLHS, in utero shunting across the atrial septum is reversed from the normal pattern. The minimal blood flow that enters the left atrium from the pulmonary veins must predominantly cross the atrial septum into the right atrium. The mixture of pulmonary and systemic venous blood then passes from the RV into the pulmonary artery. A small amount of blood enters the branch pulmonary arteries, whereas the majority goes through the ductus arteriosus. In the most extreme form of HLHS with aortic atresia, the myocardial and cerebral circulations are supplied solely by the ductus in a retrograde fashion. The lower body blood flow is also provided by the ductus arteriosus.^[10] This “adaptation” allows for hemodynamic stability during fetal life. However, flow inefficiencies are poorly tolerated in the fetus with HLHS.

Prenatal diagnosis of severe congenital cardiac malformations has been made with four-chamber ultrasonography of the fetal heart performed to evaluate ventricular size and symmetry and the direction of flow within the ascending aorta. Ultrasonography is certainly used to assess the fetus when risk factors for congenital heart disease are suggested by family history or when an

abnormality is suspected. In screening for fetal anomalies, ultrasonography is generally used during the 16th–18th gestational weeks.^[3] Although the critical events of organogenesis are completed in weeks 3–8 of the embryonic period, the growth that occurs in the next several weeks allows improved resolution of fetal anatomy. Echocardiography provides the information required for surgical planning or for recommending no intervention in severely ill infants. Angiographic studies are no longer routinely performed.

Many infants with HLHS are now diagnosed prenatally and are physiologically stable at presentation; some infants are diagnosed due to a murmur or cyanosis that is discovered in the newborn nursery prior to discharge; and still other infants are diagnosed only after becoming acutely and critically ill following ductal closure. A variety of chest radiographic findings are seen in patients with HLHS, including an enlarged cardiac silhouette (prominent right atrium), pulmonary venous hypertension, an atrial septal defect, and valvular stenosis or atresia. The recent evolution of palliative surgical procedures (modified Norwood procedure, bidirectional cavopulmonary shunt, modified Fontan procedure, aortic valvulo plasty, heart transplantation) has increased the survival rate in children with HLHS.^[11] Echocardiography allows accurate assessment of the size and location of the ductus arteriosus, the hemodynamics of the aortic root, the patency and size of the foramen ovale or atrial septal defect, and the presence of a ventricular septal defect to facilitate planning.

Despite the many potential benefits, the increasing frequency of prenatal diagnosis of HLHS has not previously been shown to result in significant improvements in outcome.^[12] Several previous studies have not found a significant difference in survival between infants with HLHS diagnosed prenatally and those diagnosed after birth. In addition, fetal diagnosis has been associated with improved surgical outcome for some biventricular heart defects. Associated cardiac malformations include pre and postductal coarctation of the aorta, patent ductus arteriosus, patent foramen ovale, dilated pulmonary artery, ventricular septal defect, dilatation of the right atrium, enlarged right ventricle, and various forms of double-outlet right ventricle.^[13] Fibrotic thickening of the endocardium (endocardial fibroelastosis) can occur in either side of the heart and contribute to poor cardiac function. Additional complicating factors include right ventricular failure and tricuspid valve or common atrioventricular valve regurgitation.

History of fetal bradycardia heart block during the first trimester of pregnancy has been associated with double outlet right ventricle. Fetal heart block can be diagnosed on ultrasound depending on the subtype of DORV with or without transposition of the great arteries and clinical history. In patients

with DORV and transposition of the great arteries, the clinical presentation depends on the location of the ventricular septal defect (VSD) and the presence of pulmonary valve stenosis (PS), the degree of PS, or both.^[14] In patients with DORV and transposition of the great arteries, the VSD may be doubly committed or remote from the great arteries. If the VSD is doubly committed, the conus septum is deficient and the VSD usually lies above the crista supraventricularis, closely related to both semilunar valves. Clinical presentation is cyanosis with signs of pulmonary over circulation. TGA as well as other congenital heart diseases are best diagnosed using the segmental approach of the fetal heart.^[15]

Atrial septal defects are not a cause of impairment of cardiac function in utero, as a large right to left shunt at the level of the atria is a physiological condition in the fetus. Most affected infants are asymptomatic even in the neonatal period. Atrial septal defects are the second most common congenital heart defect after ventricular septal defects and most commonly become symptomatic in adulthood. High output congestive cardiac failure eventually develops, usually becoming symptomatic by the age of 30.^[17]

CONCLUSION

Fetal congenital heart anomalies are the leading cause of infant death. Prenatal diagnosis of cardiac disease provides prognostic information prior to birth whether to terminate pregnancy or to undergo in utero intervention which helps improve the neonatal outcome. Hypoplastic left heart syndrome (HLHS) is a complex combination of cardiac malformations that probably results from multiple developmental errors in the early stages of cardiogenesis and that, if left untreated, invariably proves fatal. HLHS comprises a wide spectrum of cardiac malformations, including hypoplasia or atresia of the aortic and mitral valves and hypoplasia of the left ventricle and ascending aorta. In HLHS, prenatal US is most effective for diagnostic evaluation of ventricular size beginning in the 18th week of gestation and is most reliable beginning in the 22nd week of gestation. In double outlet right ventricle (DORV) with a co-existent transposition of great arteries (TGA), the ventricular septal defect may be doubly committed or remote from the great arteries. Transposition of great arteries is characterized by both the outflows being parallel to each other at the origin on color Doppler. Septum secundum type of ASD is a difficult diagnosis on antenatal ultrasonography and becomes symptomatic by adulthood with high output congestive failure as the presentation.

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