Prenatal Sonographic Diagnosis of Hypoplastic Left Heart Syndrome

Abstract

Hypoplastic left heart syndrome (HLHS) represents a variety of cardiac malformations that may result from errors in the early stages of cardiac development. HLHS includes a wide spectrum of cardiac malformations including hypoplasia of the left ventricle, ascending aorta, hypoplasia, or atresia of the aortic and mitral valves. Over the recent years, the improved resolution of advanced equipment with awareness and increased performance of second-trimester ultrasound examinations for the assessment of fetal anomalies have helped in understanding the spectrum and have expanded our knowledge of HLHS. They are one of the causes which constitute for neonatal morbidity and mortality and hence the rapid need for prenatal evaluation with ultrasound to detect cardiac anomalies. Prenatal recognition of disease also allows families to prepare for a child with a life-threatening defect by consultation with the multidisciplinary team that will care for their newborn and discussing the short- and long-term prognosis.

Keywords: *Fetal echocardiography, four-chamber view, hypoplastic ventricle*

Raghu Teja Sadineni, B Santh Kumar, NB Chander, Durga Mahita Boppana[1]

Department of Radiology, Dr. Pinnamaneni Siddhartha Institute of Medical Sciences and Research Foundation, [1]Department of Pediatrics, Rainbow Hospitals, Vijayawada, Andhra Pradesh, India

Introduction

The cardiovascular anomalies constitute the most common major congenital anomalies. At birth, the incidence of cardiovascular anomalies is 6.5 times higher than that of chromosomal anomalies and is up to four times more common than neural tube defects. However, the prenatal sonological diagnosis is difficult and hence many cases are missed undiagnosed. Congenital cardiac anomalies account for up to 20% of neonatal deaths and 50% of infant deaths and are four to five times more commonly seen in stillbirths than in live-born infants.[1] The incidence of hypoplastic left heart syndrome (HLHS) is approximately 1 in 10,000 live births;[2] however, their prenatal diagnosis by sonography remains very less. HLHS accounted for a quarter of the neonatal deaths due to congenital heart disease until the 1980s.[3,4]

Case Report

We describe the case of a 26-year-old primigravida who came for routine antenatal ultrasonography for the first time. There was no history of consanguinity. A single live fetus of 33 weeks gestation was assessed. Cardiac screening showed mild bradycardia with heart rate persistently between 100 and 120 bpm. Further assessment of cardia demonstrated small sized, echogenic left ventricle, reduced size of mitral valve orifice, capacious right ventricle and atria, and small defect in membranous part of interventricular septum. However, there was no significant reduction in size of aorta further confirming the continuum of spectrum of abnormalities forming the HLHS [Figure 1].

Discussion

Neonatal HLHS is an end point which can result from a spectrum of left heart cardiac abnormalities. Simpson in 2000 has described "Classic" HLHS which includes heart anomalies such as both aortic valve atresia and either atresia or stenosis of the mitral valve.[5] There is a reduction in the size of the left side of heart as a result of which it is unable to support systemic circulation. In the effected fetus, from early gestation itself, the left side of heart is poorly developed. Second-trimester ultrasound findings are similar to those at term and include the left heart showing markedly diminished capacity and contractility.

If not treated, HLHS is a uniformly fatal form, which is responsible for 22% of deaths from congenital heart disease in

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Address for correspondence:
Dr. Raghu Teja Sadineni, Flat 503, Aruna Residency, Netaji Street, Patamata Lanka, Vijayawada - 520 015, Andhra Pradesh, India. E-mail: sadineniraghu@gmail.com

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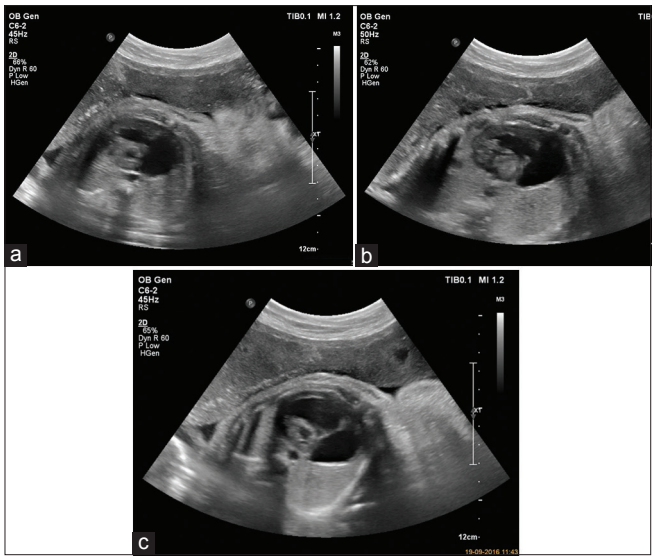

Figure 1: (a-c) Ultrasonography images of a fetus of 32-week gestational age shows small sized and hyperechoic left ventricle on four-chamber view of heart with reduced mitral valve orifice size

the 1st year of life.[6] HLHS can be detected on prenatal sonography between 18 and 22 weeks of gestation with a 4-chamber view of the fetal heart. HLHS carries a poor prognosis with a survival rate reported of 40%–55% after prenatal diagnosis.[7,8] Prenatal sonological diagnosis has been shown to improve outcome when compared with neonates not diagnosed before birth.

Second-trimester ultrasonography reveals not only a small, dysfunctional left ventricle but also critical aortic stenosis and/or severe coarctation of the aorta. Real-time images will delineate the obstructed outflow and dysfunctional contractility of the left ventricle wall.[9] When they are followed through gestation, these fetuses can develop increased cardiac wall echogenicity reflecting endocardial fibroelastosis, and the left ventricle diminishes in size relative to the normally growing right ventricle.[10] *In utero* aortic valve dilation has been proposed for HLHS due to critical aortic stenosis.

In a study by Stoll *et al.*, sonographic detection for isolated left heart syndrome had a sensitivity of 61.9%.[11] In general, the cardiac defects affecting the size of the ventricles have the highest detection rate. In various other studies, sensitivities are 36.6% and 37% for prenatal sonographic diagnosis.[11]

The most important sonographic findings include the following:[11] a small, thick-walled and hyperechoic left ventricle, with weak contractility. An absent or minute left ventricle and an anterior mitral leaflet, of 5 mm or less, is diagnostic of hypoplastic left heart ventricle, enlarged right ventricle with increased excursion of tricuspid valve, absence of antegrade flow through the aortic valve, and poor motion of aortic valve; ascending aorta hypoplasia of variable degree, which is small or not visible, and small amount of flow through it. An aortic root of 5 mm or less is consistent with aortic atresia, mitral valve hypoplasia with or without measurable flow, and poor mitral valve motion.

Extracardiac anomalies may be seen associated with hypoplastic left heart, which include two-vessel cord, craniofacial, gastrointestinal, genitourinary, and central nervous system abnormalities.

The HLHS usually presents during the 1st week of life with signs of low systemic perfusion due to constriction of ductus arteriosus because of the falling pulmonary vascular resistance. These infants tolerate their defect for a few days until the ductus remains widely open. Severe metabolic acidemia develops when the ductus constricts as the arterial pressure decreases. If not treated, almost all the affected infants die within 6 weeks.

No active obstetrical interventions are needed during pregnancy other than determining the karyotype and looking for associated anomalies. Prenatal diagnosis is important for pregnancy counseling and for planning the delivery, due to the severity of this condition and the specialized surgical treatment that is required. Prenatal diagnosis helps in preventing ductal shock (by avoiding closure of ductus arteriosus) after birth usually by the use of prostaglandin E14.

There are various palliative surgical procedures that have been proposed which include modified Norwood procedure, bidirectional cavo-pulmonary shunt, modified Fontan procedure, aortic valvuloplasty, and heart transplantation which have increased the survival rate of these children. Therapeutic approach is initially staged surgical palliation, which if unsuccessful may require cardiac transplantation.

Conclusion

HLHS which is a complex combination of cardiac malformations can be detected with prenatal evaluation with ultrasound providing an option of pregnancy termination to the parents, or intrauterine interventions may be performed if the parents wish to continue the pregnancy. It also guides the clinicians to prepare for the postnatal interventions as and when required.

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Conflicts of interest

There are no conflicts of interest.

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