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A UNIQUE PRESENTATION OF HYPOPLASTIC LEFT HEART SYNDROME – A SERIES OF 2 CASE REPORTS

KEY WORDS: congenital heart disease, hypoplastic left heart syndrome, Mitral Valve Insufficiency.

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ABSTRACT

Background: Hypoplastic left heart syndrome (HLHS) is a critical congenital heart defect marked by underdevelopment of left-sided heart structures. With an incidence of about 1 in 10,000 live births, prenatal detection is challenging but crucial. This report presents two distinct cases of HLHS diagnosed via prenatal ultrasound during the second trimester, highlighting the unique variations in presentation and the challenges associated with early detection. **Case Reports:** This article details two cases of hypoplastic left heart syndrome (HLHS) identified through prenatal ultrasound in the second trimester. In Case 1, a 31-year-old woman at 21 weeks and 5 days gestation had an ultrasound showing a hypoplastic left atrium and ventricle, thick echogenic left ventricle walls, no mitral valve flow, and a very thin ascending aorta. Despite recommendations for a dedicated fetal echocardiogram, intrauterine fetal death occurred. In Case 2, a 26-year-old at 22 weeks gestation was found to have a small echogenic left ventricle, a reduced mitral valve orifice, and a capacious right ventricle and atria, along with a small interventricular septum defect. There was no significant reduction in aortic size, but HLHS was suggested. Follow-up also resulted in intrauterine fetal death. **Conclusion:** Ultrasound plays a crucial role in diagnosing and assessing hypoplastic left ventricle (HLHS) by detecting abnormalities at the 4-chamber view (4CV) level. Prenatal echocardiography, particularly between 18 and 22 weeks of gestation, is vital for planning postnatal care and interventions. Despite advances in diagnosis and treatment, HLHS remains a significant cause of neonatal cardiac mortality. Early detection through ultrasound is essential for improving treatment outcomes and survival rates.

INTRODUCTION

Cardiovascular anomalies are the most prevalent major congenital defects, with an incidence at birth that is 6.5 times higher than chromosomal anomalies and up to four times greater than neural tube defects. Despite this, prenatal detection remains challenging, leading to many cases being missed. [1]

Congenital heart defects are responsible for up to 20% of neonatal deaths and 50% of infant deaths, and are seen four to five times more frequently in stillbirths compared to live births.[2] Hypoplastic left heart syndrome (HLHS) occurs in approximately 1 in 10,000 live births, yet its prenatal detection through sonography is still limited. Until the 1980s, HLHS was responsible for a quarter of neonatal deaths from congenital heart disease. [1]

Hypoplastic left heart syndrome (HLHS) is a lethal congenital heart defect involving the underdevelopment of the left-sided structures of the heart, such as the mitral valve, left ventricle, aortic valve, ascending aorta, and aortic arch. First described as a syndrome in 1958 by Nadas and Noonan, HLHS was initially referred to as combined aortic and mitral atresia.[3]

The incidence of HLHS is approximately 1 in 5,000 neonates, accounting for about 3% of all congenital heart disease cases in infants.[4] Historically, without treatment options, the mortality rate for HLHS was 100% within the first week of life.[5]

HLHS includes various lesions with a dominant right ventricle (RV) and systemic outflow obstruction including various shunt lesions e.g. ventricular septal defect (VSD) and atrial septal defect (ASD). [6] Underdevelopment of the left-sided structures of the heart may involve aortic valve atresia, atresia or stenosis of the mitral valve and hypoplasia of the ascending aorta and aortic arch. [7]

The etiology of HLHS is multifactorial. HLHS is a form of severe congenital heart disease characterized by a failure of the left heart structure to grow normally [8,9]. This syndrome plays a critical role because LV hypoplasia can accompany some types of heart defects or other congenital abnormalities that lead to morbidity and mortality in the first week of life [10,11]. Current evidence suggests that left ventricular hypoplasia is caused by primary defects in ventricular development [12].

There are 2 types of defect, leading to 2 distinct groups based on obstruction of blood flow: the defect of the first group is when there is obstruction of blood flow entering the LV such as mitral valve atresia (MVA) or mitral valve stenosis (MVS). The defect of the second group is a restrictive foramen ovale. This condition of the second group is when there is an obstruction in the outflow from the LV, such as atresia or aortic valve stenosis. Aortic valve stenosis is the most common cause of LV retardation. The pathophysiology of HLHS is evolutionary during fetal life: the LV will decrease in size as the pregnancy progresses, and the defect may not be detected by echo-cardiography until the third trimester. The right ventricle (RV) and right atrium (RA) will often appear dilated from increased volume loading during the prenatal period. Visualization by 4-chamber view (4CV) ultrasound is ideal for comparing the sizes of both ventricles [13].

Neonates with HLHS depend on a patent ductus arteriosus and an interatrial communication for survival until surgical intervention, and a continuous infusion of prostaglandin E1 (PGE1) is essential for maintaining ductal patency. The main treatment is surgery. Neonates with HLHS are in critical status, require care in the neonatal intensive care unit (NICU), and need to be stabilized prior to surgical intervention. Initial management includes: (1) maintaining an open ductus arteriosus; (2) avoiding massive blood flow to the lungs; and (3) ensuring blood flow from the left atrium (LA) to the RA [13]. Despite advancements in treatment, HLHS is still responsible for 23% of all cardiac deaths within the first week of life.[3]

Prenatal ultrasound with fetal echocardiography has made it possible to recognize and monitor fetuses with HLHS. This has demonstrated the progressive nature of HLHS and

emphasized the significance of aberrant flow patterns in the mechanisms of development of HLHS. Here in this case report we present 2 cases of variants of HLHS diagnosed by prenatal fetal ultrasound in second trimester at the 4CV level.

CASE REPORTS

This article describes 2 cases with multiple variations of HLHS that were referred by obstetrics and gynecology specialists based on preliminary information on congenital abnormalities in the 2nd trimester that were visible on ultrasound. A frequently observed HLHS sign at the 4CV level is that the heart in these cases is typically characterized by a small or narrow LV, with numerous variations. It is crucial to identify the morphology of the left & right heart including the moderator band and tricuspid valve to confirm the occurrence of hypoplasia. When HLHS occurs, the apex of the heart will be filled almost entirely by the RV.

Case Report 1

We describe the case of a 31-year-old primigravida who came for routine antenatal ultrasonography for in her second trimester for the first time. There was no history of consanguinity. A single live fetus of 21 weeks 5 days gestation was assessed. Cardiac screening showed normal cardiac position and mild bradycardia with heart rate persistently between 100 and 120 bpm. Further assessment of cardia in four chamber view demonstrated hypoplastic left atrium and ventricle. Left ventricle shows thick echogenic walls with reduced volume. No flow was seen through mitral valve on colour doppler. Three vessel view shows very thin calibre ascending aorta. A possible diagnosis of Hypoplastic left side heart syndrome was made and dedicated fetal echo was recommended. Follow up after 3 weeks was suggested. However, this neonate did not survive and intrauterine fetal death was reported.

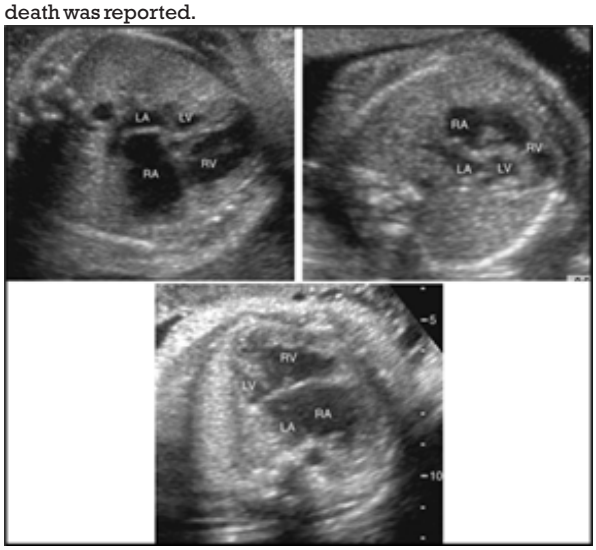

Figure 1. (a-c) Ultrasonography images of a fetus of 21-week 5 days gestational age shows small sized and hyperechoic left ventricle on four-chamber view of heart with no flow through mitral valve orifice.

Case Report 2

We report the case of a 26-year-old primigravida who presented for her first routine antenatal ultrasonography. There was no history of consanguinity. The ultrasound revealed a single live fetus at 22 weeks of gestation. Cardiac screening identified mild bradycardia, with the fetal heart rate consistently ranging between 100 and 120 bpm. Further cardiac evaluation revealed a small, echogenic left ventricle, a reduced mitral valve orifice, and a capacious right ventricle and atria. Additionally, a small defect was noted in the membranous part of the interventricular septum. Notably, there was no significant reduction in the size of the aorta, which suggested that the fetus exhibited a spectrum of abnormalities consistent with hypoplastic left heart syndrome (HLHS). On follow up, IUFD was reported.

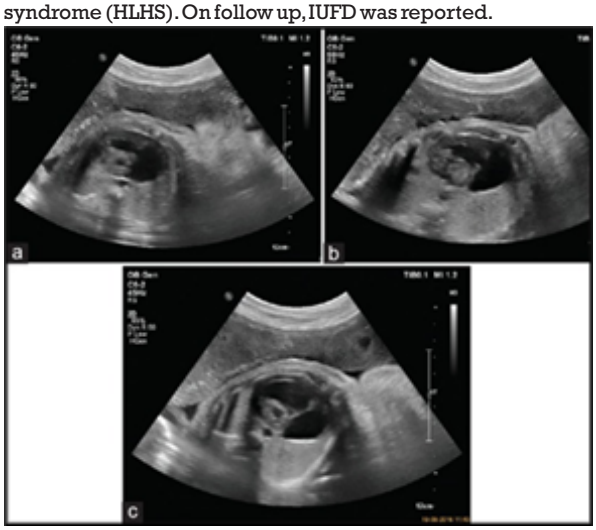

Figure 2. (a-c) Ultrasonography images of a fetus at 22 weeks of gestation reveal a small, hyperechoic left ventricle in the four-chamber view of the heart, along with a reduced mitral valve orifice size.

DISCUSSION

Hypoplastic left heart syndrome (HLHS) represents a spectrum of left-sided cardiac abnormalities, leading to severe underdevelopment of heart structures. Simpson (2000) described the "classic" form of HLHS, which includes anomalies such as aortic valve atresia and either atresia or stenosis of the mitral valve.[14] This condition results in a significantly reduced size and impaired function of the left side of the heart, compromising its ability to support systemic circulation. From early gestation, the left side of the heart is underdeveloped in affected fetuses. Second-trimester ultrasound findings, which mirror those at term, show a marked reduction in the capacity and contractility of the left ventricle.

HLHS is a uniformly fatal condition if left untreated, accounting for 22% of congenital heart disease-related deaths in the first year of life.[15] Prenatal sonography can detect HLHS between 18 and 22 weeks of gestation using a four-chamber view of the fetal heart. Survival rates post-prenatal diagnosis are reported between 40% and 55%. [16,17] Prenatal detection significantly improves outcomes compared to those diagnosed after birth.

Ultrasound findings in the second trimester may reveal a small, dysfunctional left ventricle, critical aortic stenosis, and/or severe coarctation of the aorta. Real-time imaging can demonstrate obstructed outflow and impaired contractility of the left ventricle wall.[18] Additionally, these fetuses might develop increased cardiac wall echogenicity indicative of endocardial fibroelastosis, with the left ventricle decreasing in size relative to the normally growing right ventricle.[19] In utero dilation of the aortic valve has been considered as a potential intervention for critical aortic stenosis.

A study by Stoll et al. found a sensitivity of 61.9% for the sonographic detection of isolated left heart syndrome.[20] Sensitivities for prenatal diagnosis in other studies range from 36.6% to 37%.[20] Key sonographic features indicative of HLHS include a small, thick-walled, hyperechoic left ventricle with poor contractility; a very small or absent left ventricle and mitral valve leaflet (5 mm or less); an enlarged right ventricle with increased tricuspid valve excursion; absence of antegrade flow through the aortic valve; and variable hypoplasia of the ascending aorta. Associated extracardiac anomalies may include a two-vessel umbilical cord, and abnormalities in the craniofacial, gastrointestinal,

genitourinary, and central nervous systems.

HLHS typically presents in the first week of life with signs of low systemic perfusion due to constriction of the ductus arteriosus, as pulmonary vascular resistance decreases. Infants with HLHS may initially tolerate the defect if the ductus remains open but will develop severe metabolic acidosis when it constricts. Without treatment, nearly all affected infants die within six weeks.

During pregnancy, the primary interventions involve karyotyping and screening for associated anomalies. Prenatal diagnosis is crucial for counseling and planning delivery, given the severity of HLHS and the need for specialized surgical treatment. Prenatal detection helps prevent ductal shock, often managed with prostaglandin E1 therapy.

Palliative surgical options, such as the modified Norwood procedure, bidirectional cavo-pulmonary shunt, modified Fontan procedure, aortic valvuloplasty, and heart transplantation, have improved survival rates. The initial treatment typically involves staged surgical palliation, with cardiac transplantation considered if palliative surgery is unsuccessful.

CONCLUSION

Hypoplastic left heart syndrome (HLHS), a complex condition involving a combination of cardiac malformations, can be identified through prenatal ultrasound evaluation. This detection offers parents the option to consider pregnancy termination if desired. Alternatively, if parents choose to continue the pregnancy, intrauterine interventions may be considered. Additionally, prenatal diagnosis aids clinicians in preparing for necessary postnatal interventions, ensuring timely and appropriate care for the newborn.

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