e-ISSN: 0976-822X, p-ISSN:2961-6042

Available online on http://www.ijcpr.com/

International Journal of Current Pharmaceutical Review and Research 2025; 17(12); 815-819

Case series

Fetal Echocardiographic Diagnosis of Hypoplastic Left Heart Syndrome with Associated Fndings and Pregnancy Outcome: A Case Series

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Received: 03-10-2025 / Revised: 02-11-2025 / Accepted: 03-12-2025

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Conflict of interest: Nil

Abstract:

Objectives: To analyse the main prenatal anatomical spectrum of hypoplastic left heart syndrome, its characterstics and associated other system anomalies.

Method: we retrogradely analysed our study done for screening of CHD by fetal echocardiography in 6634 consecutive obstretic patients out of which 29 cases were identified with CHD out of which 3 cases were hypoplastic left heart syndrome and its spectral disease. The study done during the period of November 2018 to December 2019.

Introduction

The term hypoplastic left heart syndrome (HLHS) describes a spectrum of congenital heart defects (CHD) characterized by severe obstruction at one or more levels of left ventricle inflow or outflow.[1] In classical HLHS, the left ventricle is unable to support the systemic circulation after birth and it is uniformly fatal if left untreated [2].

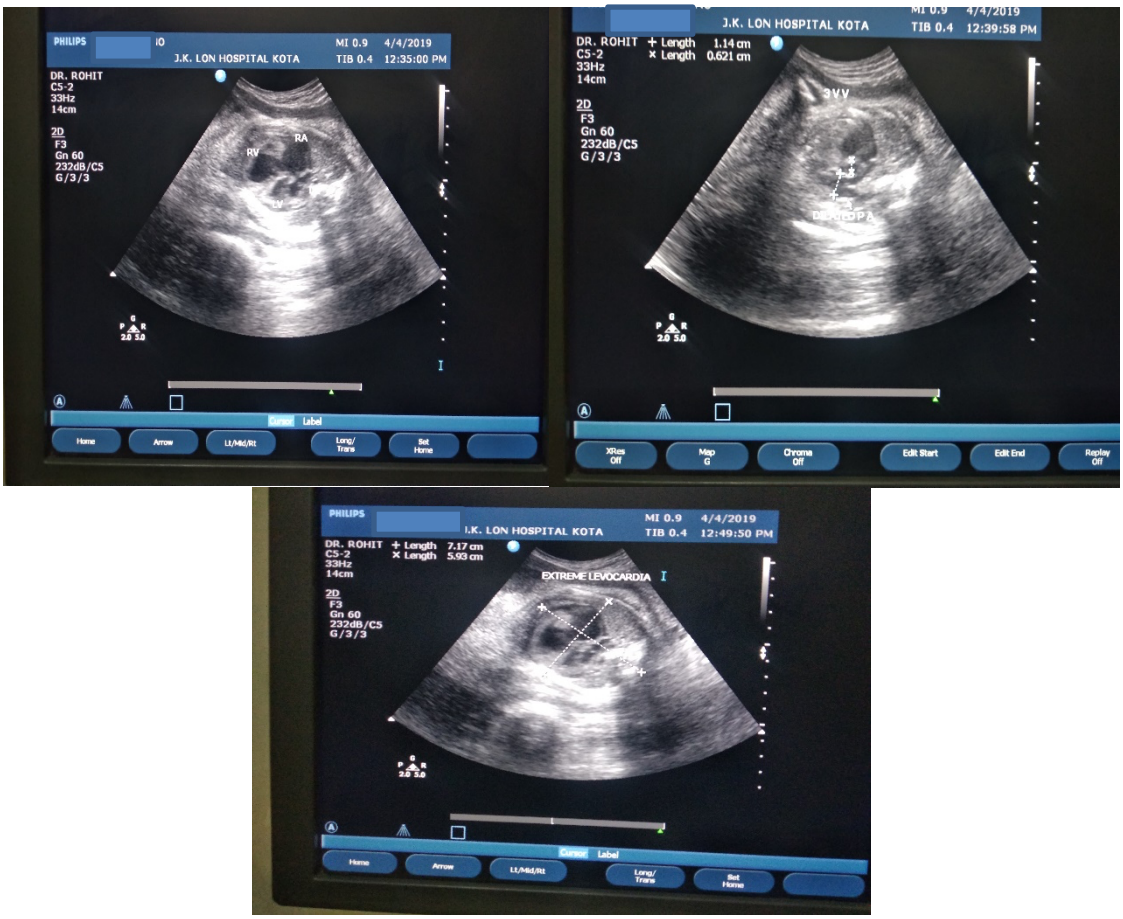
HLHS is potentially detectable on prenatal sonography with a four-chamber view of the fetal heart. Moreover, the great distortion of the cardiac anatomy associated with HLHS theoretically facilitates its detection in the routine second-trimester ultrasound scan [3, 4]. Although the prenatal diagnosis of HLHS has been shown to improve the outlook for affected patients [5] and HLHS is generally considered to be a CHD at the worst end of the severity spectrum [6]. Nevertheless, in order to provide parents without look forcurate counselling following prenatal diagnosis of HLHS, the prognosis should be based on updated fetal experience, ensuring thereby that they are in a position to make a fully informed decision about the future management of their baby. The aim of this study was to analyse the main prenatal characteristics of HLHS, its association with extracardiac anomalies and the outcome for these patients.

Case 1

We describe a case of 32 years old third gravida female, who came for routine antenatal ultrasound examination.

The patient conceived of first-degree consanguineous marriage.

A single live fetus of 34 weeks of gestation was assessed. The cardiac assessment done by fetal echocardiogram revealed mild bradycardia with heart rate between 100-120 bpm, left atrium and ventricle were small for gestational age as per the normograms. Also, there was small atretic mitral orifice and aortic annulus. Right atrium and ventricle were capacious and were showing normal contractility. Small septum secundum ASD noted, however interventricular septum was intact. On colour doppler index, no flow was detected across the mitral valve. The ductal arch was dilated and showing antegrade cardiofugal flow. The diagnosis of hypoplastic left heart syndrome was made. (figure 1A, 1B &1C)

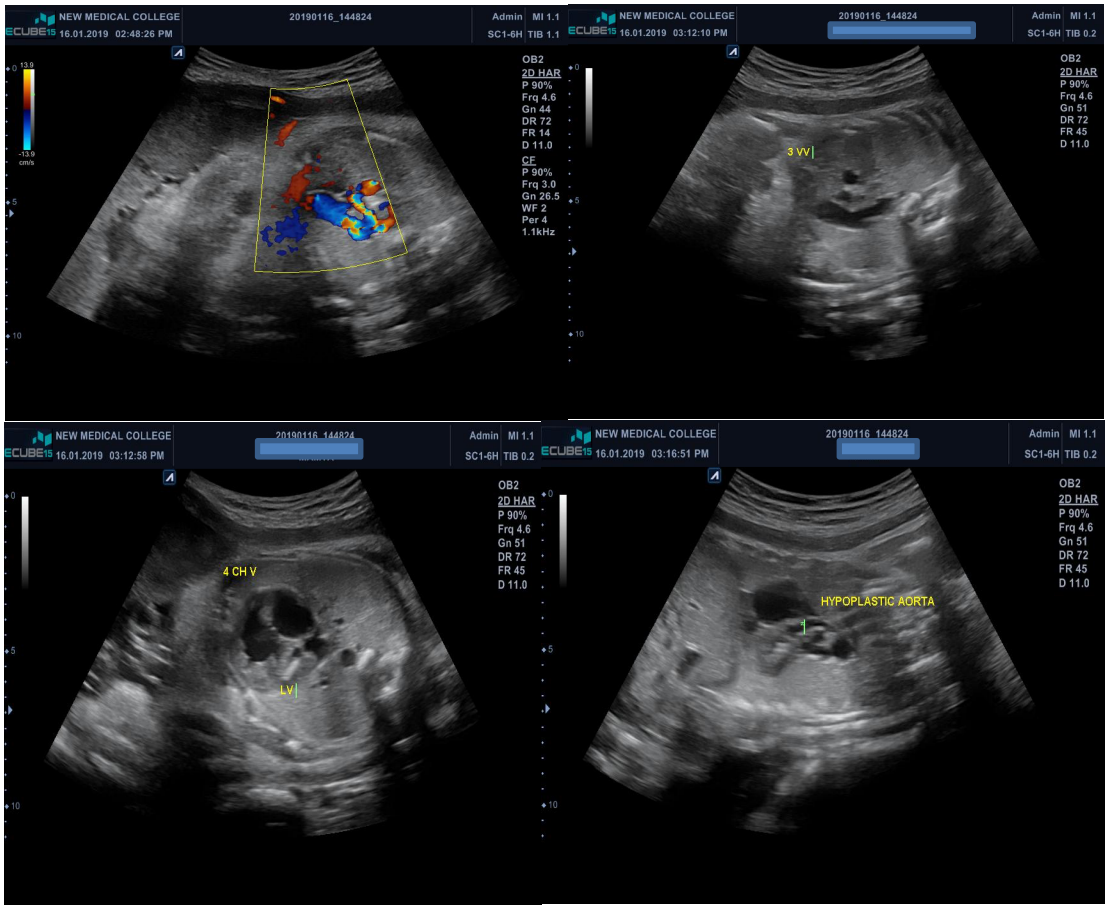


Case-2

A case of 29 years old fourth gravida female, who came for routine antenatal ultrasound examination for the first time.

She conceived of non-consanguineous marriage.

Single live fetus of 29 weeks of gestation was assessed. The cardiac evaluation showed extreme levocardia with cardiac axis of 90-degree, small hypoplastic left atrium and ventricle, small aortic orifice. The lining of hypoplastic left ventricle was thickened and echogenic suggestive of endocardial fibroelastosis. The three-vessel view showed small hypoplastic aorta. Interatrial septum and interventricular septum were intact. Right atrium and ventricle were normal for gestational age. On colour doppler index, minimal flow was detected across the mitral valve. The diagnosis of hypoplastic left heart syndrome was made. The extracardiac anomalies were bilateral cleft lip and palate, right PUJ obstruction, ambiguous genitals and unilateral club foot. (figure 2A to 2D)

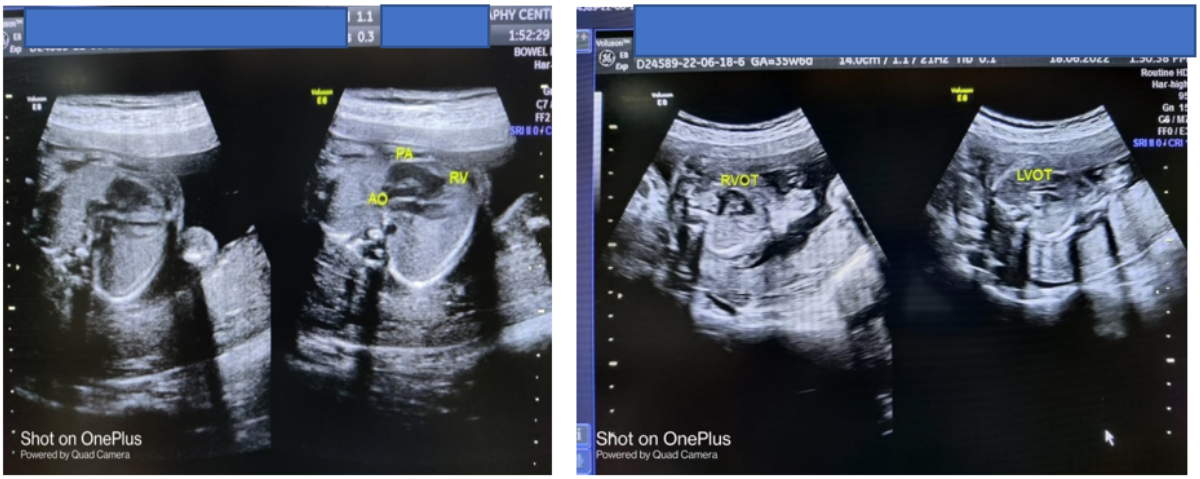


Case-3

A case of 27 years old primigravida female came for routine antenatal scan.

The patient conceived of assisted reproductive technology.

A single live feu of 35 weeks of gestation was assessed. The cardiac evaluation showed extreme levocardia with cardiac axis of approximately 85 degree, relatively smaller left ventricle, left atrium and significantly small aortic annulus. However, normal sized mitral orifice. The three-vessel view showed small hypoplastic aorta. Interatrial septum was intact with bulging of foraminal flap in right atrium. Small defect noted muscular part of interventricular septum. Right atrium and ventricle were normal for gestational age. On colour doppler index, minimal flow was detected across the mitral valve. The diagnosis of hypoplastic aortic arch with left hypoplasic heart was made. Post natal 2-D echo confirmation confirmed the diagnosis. (Figure 3A and 3B)



Discussion

Hypoplastic left heart syndrome is one of the most severe cyanotic congenital cardiac anomalies, characterised by severe hypoplasia of left ventricle, ascending aorta and atretic or hypoplasic mitral valve. The reported birth incidence of HLHS is 0.1 to 0.25 per 1000 live births [7]. HLHS accounts for 3.8% of all congenital cardiac abnormalities, and up to seven-tenths of cases occur in boys [8].

Although HLHS is one of the most commonly diagnosed congenital heart abnormalities in utero, it is still missed in a significant proportion of fetuses. Recurrence of left-sided congenital heart disease has been reported in the range of 2% to 13% [9].

In HLHS, Left atrium can be small or normal sized. The intact interatrial septum is uncommon finding. The ductus arteriosus is dilated to support the systemic circulation and thus the increased pulmonary circulation leads to severe congestive cardiac failure.

HLHS is associated with a 4% to 5% incidence of chromosomal abnormalities [10] such as Turner syndrome, trisomies [13, 18], and others. Extracardiac malformations have been reported in 10% to 25% of infants with HLHS [11,12] with associated genetic syndromes such as Turner syndrome, Noonan syndrome, Smith-Lemli-Opitz syndrome, and Holt-Oram syndrome [11, 13]. Growth restriction may be seen in fetuses with HLHS probably due to a 20% reduction in combined cardiac output [14].

If not treated, HLHS is a uniformly fatal form, which is responsible for 22% of deaths from congenital heart disease in the 1st year of life. [15]. 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. [16,17]. 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.[18] 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 [19]. 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%. [20]. 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. [20]

Conclusion

Hypoplastic left heart syndrome is one of the complex congenital heart diseases which shows anatomical variation in degree and severity of its components. Fetal echocardiography plays important role in diagnosis as well as prognosis of the disease. It can be detected in early second trimester on four chamber view, however extended fetal echocardiography may reveals addition information i.e. function and contractility of left ventricular myocardium, aortic annulus and associated secondary changes in hemodynamic function.

References:

1. Tchervenkov CI, Jacobs ML, Tahta SA. Congenital heart surgery nomenclature and database project: hypoplastic left heart syndrome. Ann Thorac Surg. 2000; 69: S170–S179.
2. Simpson JM. Hypoplastic left heart syndrome. Ultrasound Obstet Gynecol. 2000; 15: 271–278.
3. Sharland GK, Allan LD. Screening for congenital heart disease prenatally. Results of a 2 1/2 year study in the South East Thames Region. Br J Obstet Gynaecol. 1992; 99: 220–225.
4. Nikkil¨a A, Bj ¨ orkhem G, K¨all´en B. Prenatal diagnosis of congenital heart defects – a population based study. Acta Paediatrica. 2007; 96: 49–52.
5. Tworetzky W, McElhinney DB, Reddy VM, Brook MM, Hanley FL, Silverman NH. Improved surgical outcome after fetal diagnosis of hypoplastic left heart syndrome. Circulation. 2001; 103: 1269–1273.
6. Allan LD, Huggon IC. Counselling following a diagnosis of congenital heart disease. Prenat Diagn. 2004; 24: 1136–1142.
7. Ferencz C, Rubin JD, McCarter RJ, et al. Congenital heart disease: prevalence at livebirth: the Baltimore Washington infant study. Am J Epidemiol. 1985;121:31–36.
8. Morris CD, Outcalt J, Menashe VD. Hypoplastic left heart syndrome: natural history in a geographically defined population. Pediatrics. 1990;85:977–983.
9. Boughman JA, Berg KA, Astemborski JA, et al. Familial risks of congenital heart defects assessed in a population based epidemiologic study. Am J Med Genet. 1987;26:839–849.
10. Raymond FL, Simpson JM, Sharland GK, et al. Fetal echocardiography as a predictor of

chromosomal abnormality. Lancet. 1997; 360: 930.
11. Natowicz M, Chatten J, Clancy R, et al. Genetic disorders and major extra-cardiac anomalies associated with the hypoplastic left heart syndrome. Pediatrics. 1988;82:698–706.
12. Callow LB. Current strategies in the nursing care of infants with hypoplastic left heart syndrome undergoing first-stage palliation with the Norwood operation. Heart Lung. 1992; 21(5):463–470.
13. Connor JA, Thiagarajan R. Hypoplastic left heart syndrome. Orphanet J Rare Dis. 2007; 2:23.
14. Rosenthal GL. Patterns of prenatal growth among infants with cardiovascular malformations: possible fetal hemodynamic effects. Am J. Epidemiol. 1996;143:505–513.
15. Gillum RF. Epidemiology of congenital heart disease in the United States. Am Heart J. 1994; 127(4 Pt 1):919-27.
16. Allan LD, Apfel HD, Printz BF. Outcome after prenatal diagnosis of the hypoplastic left heart syndrome. Heart. 1998;79:371-3.
17. Brackley KJ, Kilby MD, Wright JG, Brawn WJ, Sethia B, Stumper O, et al. Outcome after prenatal diagnosis of hypoplastic left-heart syndrome: A case series. Lancet. 2000;3 56: 1143-7.
18. Allen RH, Benson CB, Haug LW. Pregnancy outcome of fetuses with a diagnosis of hypoplastic left ventricle on prenatal sonography. J Ultrasound Med. 2005; 24: 1199-203.
19. Hornberger LK, Need L, Benacerraf BR. Development of significant left and right ventricular hypoplasia in the second and third trimester fetus. J Ultrasound Med. 1996; 15: 655-9.
20. Stoll C, Dott B, Alembik Y, De Geeter B. Evaluation and evolution during time of prenatal diagnosis of congenital heart diseases by routine fetal ultrasonographic examination. Ann Genet. 2002;45:21-7.