RESEARCH PAPER

Prenatal Diagnosis of Hypoplastic Left Heart Syndrome: A Case Report

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Running Head: Prenatal Diagnosis of Hypoplastic Left Heart Syndrome

Abstract
Background
Hypoplastic Left Heart Syndrome (HLHS) is a rare and severe form of congenital heart disease characterized by underdevelopment of left-sided cardiac structures, resulting in inadequate systemic circulation. Prenatal diagnosis through fetal echocardiography plays a crucial role in parental counselling, pregnancy management, and optimization of neonatal outcomes.
Case Presentation
A 26-year-old primigravida at 23 weeks of gestation was referred to our tertiary care centre following detection of abnormal cardiac anatomy on routine anomaly scan. Detailed fetal echocardiography revealed marked hypoplasia of the left ventricle, severe mitral and aortic valve hypoplasia, and retrograde flow in the ascending aorta, consistent with HLHS. No extracardiac anomalies were identified. The couple received multidisciplinary counselling regarding prognosis, postnatal surgical options, and anticipated neonatal complications. Serial antenatal surveillance was performed, and delivery was planned at a tertiary cardiac centre. A live male neonate weighing 2.8 kg was delivered at 38 weeks by elective lower segment caesarean section. Postnatal echocardiography confirmed the prenatal diagnosis. Prostaglandin E1 infusion was initiated immediately after birth, and the neonate was referred for staged surgical palliation.
Conclusion
Prenatal diagnosis of HLHS enables timely counselling, delivery planning, and coordinated neonatal cardiac management. Early fetal echocardiography remains essential for improving perinatal preparedness and optimizing outcomes in complex congenital cardiac anomalies.
Keywords: Hypoplastic left heart syndrome, fetal echocardiography, congenital heart disease, prenatal diagnosis, fetal cardiac anomaly
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Introduction

Congenital heart disease (CHD) is the most common congenital malformation worldwide, affecting approximately 8–12 per 1,000 live births. Among these, Hypoplastic Left Heart Syndrome (HLHS) represents one of the most severe cyanotic congenital cardiac anomalies, accounting for nearly 2–3% of all congenital heart defects. HLHS is characterized by underdevelopment of the left ventricle and varying degrees of hypoplasia or atresia of the mitral valve, aortic valve, and ascending aorta.

Without surgical intervention, HLHS is uniformly fatal in the neonatal period because systemic circulation becomes dependent on a patent ductus arteriosus. Advances in fetal echocardiography have enabled antenatal detection of complex congenital heart diseases, allowing appropriate counselling, planned delivery at tertiary care centres, and early neonatal intervention.

Prenatal diagnosis of HLHS is commonly suspected during routine anomaly scans by identifying disproportion between the cardiac ventricles on the four-chamber view. Confirmation is achieved through detailed fetal echocardiography demonstrating left-sided obstructive lesions and altered flow dynamics.

This report describes the prenatal diagnosis, antenatal management, and immediate neonatal outcome of a fetus diagnosed with HLHS at a tertiary care centre.

Case Presentation

A 26-year-old primigravida with a spontaneous singleton pregnancy was referred to the fetal medicine unit at 23 weeks of gestation after an abnormal four-chamber cardiac view was identified during routine second-trimester anomaly screening. The patient had no significant medical disorders, history of teratogenic exposure, consanguinity, or family history of congenital heart disease.

Routine antenatal investigations were within normal limits. Ultrasonography revealed a live singleton fetus corresponding to gestational age with normal amniotic fluid volume and appropriate fetal growth. Detailed fetal cardiac assessment demonstrated marked asymmetry between the ventricles, with severe hypoplasia of the left ventricle. The right ventricle appeared enlarged and formed the cardiac apex.

Subsequent fetal echocardiography revealed:

- Severely hypoplastic left ventricle

Hypoplastic ascending aorta

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Severe mitral valve hypoplasia with near atresia

- Aortic valve stenosis with minimal forward flow

Retrograde flow within the ascending aorta on colour Doppler

Patent foramen ovale with right-to-left shunting

Preserved right ventricular systolic function

No extracardiac structural anomalies were identified. Fetal biometry corresponded with gestational age, and Doppler studies of the umbilical artery and middle cerebral artery were normal.

Following diagnosis, the patient and her family underwent multidisciplinary counselling involving obstetricians, fetal medicine specialists, neonatologists, and pediatric cardiologists. The poor prognosis, need for staged postnatal cardiac surgery, risk of neonatal morbidity and mortality, and long-term neurodevelopmental implications were discussed in detail. Options including continuation of pregnancy and postnatal palliation were explained.

The pregnancy was continued with serial antenatal surveillance every 3–4 weeks to monitor fetal growth, amniotic fluid, and cardiac status. No evidence of hydrops fetalis or fetal compromise developed during follow-up.

At 38 weeks of gestation, an elective lower segment caesarean section was performed due to obstetric indications. A live male neonate weighing 2.8 kg was delivered with APGAR scores of 7 and 8 at 1 and 5 minutes respectively. The neonate developed cyanosis and respiratory distress shortly after birth and was shifted to the neonatal intensive care unit.

Postnatal echocardiography confirmed the diagnosis of HLHS with severe left ventricular hypoplasia and critical left ventricular outflow tract obstruction. Prostaglandin E1 infusion was initiated to maintain ductal patency. After stabilization, the neonate was referred to a specialized pediatric cardiac centre for staged surgical palliation.

Discussion

Hypoplastic Left Heart Syndrome is a complex congenital cardiac anomaly involving underdevelopment of left-sided cardiac structures, resulting in inadequate systemic circulation. The condition includes a spectrum of abnormalities ranging from mitral stenosis or atresia to severe hypoplasia of the left ventricle and ascending aorta.

Prenatal diagnosis of HLHS has significantly improved over the past two decades due to advances in fetal echocardiography and increased implementation of routine anomaly screening. The classical antenatal sonographic finding is ventricular disproportion with a diminutive left ventricle visualized on the four-chamber view. Colour Doppler evaluation demonstrating retrograde flow in the ascending aorta is considered highly suggestive of HLHS.

Early antenatal diagnosis is clinically important because affected neonates require immediate postnatal stabilization with prostaglandin infusion to maintain ductal patency and ensure systemic perfusion. Prenatal diagnosis also facilitates planned delivery at tertiary centres equipped with pediatric cardiac surgery services, thereby reducing delays in intervention.

Despite advances in neonatal intensive care and staged surgical palliation procedures such as the Norwood, Glenn, and Fontan operations, HLHS continues to be associated with substantial morbidity and mortality. Long-term complications may include neurodevelopmental impairment, arrhythmias, ventricular dysfunction, and reduced exercise tolerance.

In the present case, antenatal diagnosis enabled timely multidisciplinary counselling and planned neonatal management. Absence of extracardiac anomalies and hydrops fetalis were favorable prognostic indicators. Nevertheless, the family was counselled regarding the guarded prognosis and need for multiple staged cardiac surgeries.

This case highlights the importance of detailed fetal cardiac screening during routine anomaly scans. Early referral for fetal echocardiography in suspected cases remains essential for accurate diagnosis and appropriate perinatal planning.

Conclusion

Prenatal diagnosis of Hypoplastic Left Heart Syndrome through fetal echocardiography plays a vital role in optimizing perinatal care. Early identification enables comprehensive parental counselling, coordinated multidisciplinary management, and planned delivery at specialized cardiac centres. Routine fetal cardiac evaluation during anomaly scans remains essential for improving early detection of complex congenital heart diseases.

DECLARATIONS

Ethics Approval

This case report was conducted in accordance with the ethical principles outlined in the Declaration of Helsinki (2013 revision). Formal Institutional Ethics Committee approval was not obtained as this was a retrospective single case report based on anonymized clinical data with no direct patient intervention. Patient confidentiality and privacy were strictly maintained throughout manuscript preparation. Written informed consent for publication of clinical details and imaging findings was obtained from the patient/guardian.

Author Contributions

SSB Fatima: Conceptualization, literature review, manuscript drafting, data interpretation, and final approval of the manuscript.

KP Patil: Supervision, critical intellectual revision, and final approval of the manuscript.

MC Metgud: Critical review of manuscript and final approval.

M Savanur: Critical review of manuscript and final approval.

All authors meet the four ICMJE criteria for authorship.

Conflicts of Interest

The authors declare no conflicts of interest.

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Data Availability
All relevant clinical data supporting the findings of this case report are included within the article. Additional de-identified information may be made available by the corresponding author upon reasonable request.

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