

*Case Report*

Hypoplastic Left Heart Syndrome - A Rare Congenital Anomaly as a Cause of Fetal Death

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ABSTRACT

Hypoplastic left heart syndrome (HLHS) is one of the major congenital anomaly reported to occur in 0.016-0.036% of all live births. Generally, these children if born alive, present with profound cyanosis, non-specific heart murmur and signs of cardiogenic shock. In our case, HLHS was diagnosed antenatally on ultrasonography and further confirmed on fetal echocardiography. However, intra-uterine death occurred at 20 weeks of pregnancy. Autopsy findings confirmed HLHS.

Key words: Hypoplastic heart, Congenital heart disease, Fetal autopsies

INTRODUCTION

Hypoplastic left heart syndrome (HLHS) refers to the abnormal development of left sided cardiac structures resulting in obstruction of blood flow from left ventricular outflow tract. Most of the time, it results in fetal death. ^[1] Prevalence of HLHS is 0.016-0.036% of all live births. ^[2] The etiology of HLHS is not precisely known however autosomal recessive, autosomal dominant and polygenic inheritances have been suggested. ^[3] We present a case of fetal autopsy due to HLHS in an intra-uterine fetal death at 20 weeks of gestation.

CASE REPORT

A 29 year old pregnant female with G2P1A0 presented with history of 20 weeks of amenorrhea with pain in abdomen. There was no significant contributory past, systemic or family history of any disorder.

There was no history of exposure to drug or radiation. Routine investigations reveal no significant abnormalities. On clinical examination, internal os was closed and cervix was healthy. On auscultation, fetal heart sounds were not audible. Prenatal diagnosis was confirmed by obstetric ultrasonography (USG) and fetal echocardiography which revealed hypoplasia of both left atrium and ventricle with complete absence of mitral and aortic valve. Hypertrophy of right ventricle and pulmonary trunk was noted. Aortic atresia was noted. Mild polyhydramnios was seen. USG confirmed intra-uterine death. After proper counselling, parents opted for oxytocin induced vaginal delivery. The fetal autopsy was performed after proper consent.

The fetal autopsy findings were-On external examination, premature female fetus weighing 1 kg with evidence of severe

cyanosis, pallor and oedema. All anthropometric measurements were - head circumference = 26 cm, chest circumference = 23 cm, crown heel length = 35 cm, crown rump length = 20 cm, foot length = 15 cm. No gross skeletal abnormality detected. Anal opening was patent. In situ examination showed congested lungs. The gastrointestinal tract, liver, adrenals and kidneys show congestion and focal areas of haemorrhages. The cardiac position was situs solitus with all major vessels in their normal position. Heart was dissected along the flow of blood. On opening the heart, it revealed only presence of right atrium and ventricle. Left sided chambers were completely hypoplastic with absence of mitral and aortic valve. Aortic atresia was noted. Pulmonary trunk was prominent and right ventricle was hypertrophied (Fig 1). The pericardium was normal.

Based on these findings, the diagnosis of hypoplastic left heart syndrome was made. The other cranio-facial, gastrointestinal, genito-urinary, central nervous system, etc anomalies were not found. The umbilical cord and the placenta were normal.

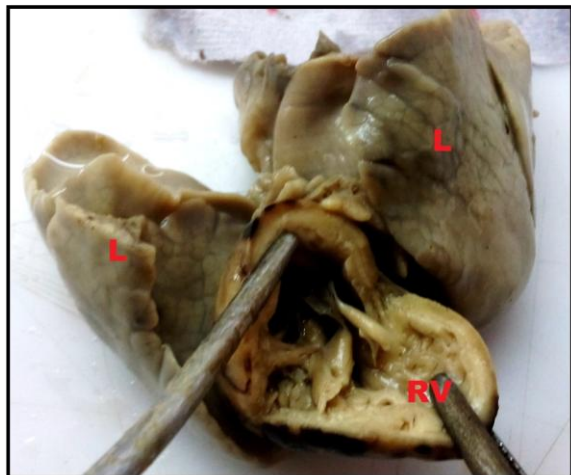


Fig 1. Gross photograph showing hypoplastic left atrium and ventricle with dilatation and hypertrophy of right ventricle with bilateral lungs. (L=lung; RV= right ventricle)

DISCUSSION

HLHS was initially termed hypoplasia of aortic tract complex by Lev in 1952. [4] HLHS refers to abnormal development of left sided cardiac structures resulting in obstruction to blood flow from left ventricular outflow tract. Severity may vary from normal birth to intra-uterine death. [5] The syndrome has been reported to occur in approximately 0.016-0.036% of all live births. [2] It accounts for 1-3.8% of all congenital cardiac malformations. The recurrence risk in siblings is 0.5%, with other forms of congenitally malformed hearts seen in 13.5%. [6]

Risk factors for development of HLHS are maternal, gestational and familial condition, exposure to teratogenic drugs, maternal infection such as rubella, herpes virus, coxsackie virus and cytomegalovirus, are further additive. Chromosomal aberrations resulting from gene defect account for about 6%. [7] HLHS is responsible for 20% of cardiac deaths in 1st week of life. Almost all the affected infants die within 6 weeks if left untreated.

These patients typically show profound cyanosis attributed to cardiogenic shock, if survived they are likely to die in absence of any intervention such as catheter based or surgical septostomy performed soon after birth, to relieve obstruction or to create a means of communication in the atrial septum. [8]

The other significant obstructive cardiac disorder mimicking this condition should be carefully rule out like critical aortic stenosis, coarctation of aorta, interrupted aortic arch, as their clinical presentation is cardiogenic shock like state. [8]

Genetic counselling and testing should be offered to both parents as multiple genetic syndromes are identified like Turner's, Noonan's, etc. Fetal echocardiography is the main diagnostic modality for antenatal diagnosis particularly

between 18-22 weeks of gestation. [8] The prenatal USG diagnosis is reported to be about 37%. [9] Comprehensive counselling by health care team is critical component of care for family of child with HLHS. Currently, HLHS can be detected in fetuses in 1st trimester of pregnancy and may choose different treatment options like medical termination of pregnancy, supportive care, Norwood (3 staged) surgical procedure and/or cardiac transplantation.

CONCLUSION

As a result of advanced technology, refined surgical techniques and catheter based interventions, antenatal diagnosis of HLHS is of utmost importance and these should be managed with utmost intranatal care to avoid intrauterine death. We present this case for rarity, more emphasis on protocol and most important counselling of parents and their genetic testing to reduce further such birth defects in next siblings.

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