CASE REPORT

Hypoplastic left heart syndrome with parchment left ventricle: A rare perinatal autopsy case report

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Abstract

Hypoplastic left heart syndrome (HLHS) refers to the abnormal development of the left-sided cardiac structures, resulting in obstruction to blood flow from the left ventricular outflow tract. In addition, the syndrome includes underdevelopment of the left ventricle, aorta, and aortic arch, as well as mitral atresia or stenosis. The syndrome can be diagnosed by fetal echocardiography between 18 and 22 weeks of gestation. This is a fetal autopsy case report of HLHS, which was diagnosed on ultrasonography and medically terminated at 20 weeks of gestation. The autopsy and histopathological findings are discussed in detail. Some important aspects of epidemiology, prenatal diagnosis, and current literature on HLHS are highlighted.

Keywords

Autopsy, hypoplastic left heart syndrome, parchment left ventricle

Introduction

The most common major congenital anomalies are cardiovascular which occur in approximately 8 of 1000 births and account for approximately 20% of neonatal deaths and 50% of infant deaths, and are seen four to five times more frequently in stillbirths than in live-born babies.[¹]

Hypoplastic left heart syndrome (HLHS) is a rare uniformly fatal disease if it is not treated. No geographical or ethnic predilection in the worldwide distribution. Death results within few weeks after birth because of variable underdevelopment of the left ventricle that is unable to sustain the systemic circulation. In HLHS, the structural defect is variable but generally involves all or most components of the left side of the heart, i.e. the left atrium, mitral valve, and aorta are underdeveloped or absent.[²]

Even in resource-limited countries, the use of echocardiogram has made it possible to make an accurate diagnosis of specific heart lesions, even though the multidisciplinary approach is still a mirage in these settings.[³]

Case Report

We report a case of fetal autopsy with left hypoplastic heart syndrome, which was antenatally confirmed on ultrasonography (USG) and hence medically terminated at 20 weeks of gestation in a 23-year-old mother who was gravida 3, para 0, and abortion 3. Parental consent for autopsy was obtained. The autopsy revealed the body to be that of a well-developed 430 g male infant of approximately 20 weeks’ gestation. External appearance was normal without any gross deformities. The baby was cyanotic. Abnormal fluid collection in pleural and peritoneal cavities was seen. All body organs were present and in normal anatomical position; there was no malrotation of the organs. The most notable findings involved the heart, which was abnormally shaped, weighed 10 g with measurement of 2 cm × 1 cm × 1 cm, the left ventricle was pale and markedly thinned out [Figure 1]. Right ventricle was thickened with both aorta and pulmonary artery arising from it. Aorta was equal in size with pulmonary artery suggesting aortic atresia. Both ventricular and atrial septal defect and patent ductus arteriosus were present. Left atrium was hypoplastic with mitral atresia.

Histological examination showed absence of myocardium in the free left ventricular wall with epicardium separated by only wisps of cardiac muscle from the endocardium [Figure 2].

Discussion

Lev in 1952, termed HLHS initially as hypoplasia of the aortic tract complex. HLHS was first used in 1958, by Noonan and Nadas to collectively describe their series of specimens with
multiple malformations involving left-sided structures of the heart.

HLHS refers to the abnormal development of the left-sided cardiac structures, resulting in obstruction to blood flow from the left ventricular outflow tract. The syndrome also includes underdevelopment of the left ventricle, aorta, aortic arch, mitral atresia or stenosis. As in most congenital heart diseases, the embryologic cause is not fully known. A multifactorial influence is attributed to be the cause of up to 90% of cardiac anomalies, with a 2% to 6% recurrence rate in further offspring.\[4\]

A normal fetus has a parallel circulation that adequately supports single-ventricle physiology before birth. Three communications (the ductus venosus, foramen ovale, and ductus arteriosus) shunt oxygenated placental blood largely past the hepatic and pulmonary beds to supply the splanchnic circulation. HLHS is well supported in this situation, and as a result, it is rarely a cause of fetal demise. And hence HLHS is probably a secondary result of early obstructive lesions of either mitral or aortic valvular development. However, the primary cause of the obstructive flow lesion that leads secondarily to HLHS is unknown. This argues for a complex early multifactorial event or, more likely, a transient early insult.\[5\]

Patients are categorized into three primary subsets on the basis of atrioventricular and semilunar valvular morphology:
1. Aortic atresia with mitral atresia (40%),
2. Aortic stenosis with mitral stenosis (30%), and
3. Aortic atresia with mitral stenosis (30%)\[5\]

The said fetus had both aortic atresia and mitral atresia and was supported by ductus venosus, foramen ovale, and ductus arteriosus. The free left ventricular wall was parchment like, an unusual feature, often described in the right ventricle in cases of Uhl’s syndrome.\[6\] Histological examination confirmed the absence of muscle, a thin layer of elastic and fibrous tissue being all that separated the epicardium, and the endocardium.

Prenatal echocardiography can be used to detect unbalanced ventricles as early as 20 weeks of gestational age. Postnatal echocardiography establishes the diagnosis and guides medical and surgical decision-making.\[5\]

An increase in the number of cases in which HLHS is detected prenatally is seen because of the widespread use of ultrasound screening for fetal malformations. Despite this advantage which provides the opportunity to plan perinatal management, the outlook for these fetuses is still poor, with an overall survival rate below 40% and highlights the importance of presenting these figures when counseling parents with affected foetuses. Several factors explain the discrepancies between fetal and surgical series. These limitations worsen with the negative feedback generated by the high rate of termination of pregnancy (TOP) following prenatal diagnosis of HLHS.\[7\]

With the incidence of HLHS about 0.1-0.25/1000 live births, in the Indian scenario, we should expect about 2000 babies with HLHS born every year. In reality, the total number of babies with HLHS seen in major centers in India will not be more than 100 children/year. Most of the fetuses with a diagnosis of HLHS get terminated by parents’ options and counseling offered to them by perinatologists.\[8\]

The main reason for referral was suspected heart defect on a routine ultrasound scan (82%). The mean gestational age at diagnosis was 21 weeks. Most cases were detected at ≤22 weeks (72%), the upper limit for TOP in Spain. 79% (58/73) of the cases in which HLHS was detected at ≤22 weeks were terminated.\[7\]

HLHS constitutes 5% of all cases of congenital heart disease and is responsible for 25% of cardiac deaths in the 1st week of life. Among 10,000 live births, approximately 1.8 will be born with HLHS, with a slight male predominance. The recurrence risk is 2.2% for one affected sibling and 6% for two affected siblings, suggesting a genetic predisposition.\[5\]

Autosomal recessive, autosomal dominant, and polygenic inheritances have been suggested. Associated extracardiac
abnormalities of craniofacial, gastrointestinal, genitourinary, and central nervous system are seen. Prenatal diagnosis with good counseling is necessary because of high perinatal mortality. A karyotype analysis and detailed ultrasonographic scan should be done to rule out other anomalies.[9]

In our case, prenatal USG established the diagnosis and guided the decision of termination. Autopsy examination demonstrated the HLHS without extracardiac anomalies. In view of the previous pregnancy loss, and associated recurrence risk counseling and karyotyping was advised.

References
