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Leonard B. Goldstein *

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Review Article

Hypoplastic Left Heart Syndrome: Management and Treatment

Katherine Welch, James Keane, Leonard B. Goldstein*

Assistant Vice President for Clinical Education Development, A.T. Still University.

***Corresponding Author**: Leonard B. Goldstein, Assistant Vice President for Clinical Education Development, A.T. Still University.

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Abstract

Stroke remains a leading cause of disability and death in the United States, disproportionately affecting Black, Latinx, and Asian or Pacific Islander populations. Atrial Fibrillation (AF), a prevalent arrhythmia, further elevates stroke risk. This article reviews the impact of culturally tailored prevention programs on health equity, particularly in underserved populations. Evidence from studies highlights the effectiveness of culturally specific education in improving stroke awareness, prevention behaviors, and emergency response in minority communities. Programs that integrate cultural beliefs and address systemic barriers show promise in reducing stroke-related disparities. However, challenges in sustaining long-term behavior change and addressing healthcare access persist. Future research should focus on refining these programs, fostering trust between patients and providers, and overcoming systemic obstacles to improve stroke outcomes for all populations.

Keywords: acutest elevation myocardial infarction; cardiogenic shock; percutaneous coronary intervention; double culprit vessel occlusion

Introduction

Pre-natal diagnosis of congenital heart disease (CHD) was difficult if not impossible until transvaginal ultrasound imaging began to be used and reported in the 1990s [1]. Thereafter, other patient groups with a high risk of CHD have been defined including fetuses with increased nuchal translucency, exposure to teratogenic drugs and/or previous pregnancies with CHD [2]. One form of CHD, hypoplastic left heart syndrome presents with a stark anatomical and highly morbid clinical presentation [3]. While accounting for only about 4% of CHD cases, hypoplastic left heart syndrome is responsible for between 15%-25% of CHD-associated deaths [3, 4]. A well-established correlation has been identified between pre-natal diagnosis of CHD and better outcomes. This may include fewer pre-operative risk factors, less mortality, and increased parental preparedness [5, 6]. With the aim of improving clinician familiarity with this rare but impactful syndrome, and call for additional research, this article will review the management and treatment of hypoplastic left heart syndrome and discuss the importance of pre-natal diagnosis in the management and treatment.

Overview of Hypoplastic Left Heart Syndrome

Hypoplastic Left Heart Syndrome is a congenital heart defect characterized by the underdevelopment of the left sided heart structures. This includes the left ventricle, mitral valve, aortic valve, ascending aorta, aortic arch, and left atrium. The left ventricle is typically significantly underdeveloped and therefore cannot support circulation. There is typically aortic valve atresia or stenosis which leads to an underdeveloped aorta. The mitral valve is typically underdeveloped as well which contributes to overall underdevelopment of all the left sided heart structures. The aorta is underdeveloped and sometimes coarctation is seen as well, further contributing to the severity of this condition. In addition to the left sided structures sometimes the interatrial septum is closed or the foramen ovale is stenotic which slows or stops blood flow from the left atrium to the right atrium.[7]

The pathophysiology of this condition is not fully known but there are known genetic and environmental factors that contribute. It can present as an isolated phenotype or as a part of a larger genetic disorder[3]. Because of the multifactorial etiology that is associated with Hypoplastic left heart syndrome it is difficult to pinpoint one specific cause but the MYH6 variants are present in >10% of patients[3]. This mutation is also associated with many other congenital heart disease conditions. Genes that have also been implicated in HLHS patients include NOTCH1, NKX2.5, ERBB4, HAND1, and GJA1.[3]. During fetal development HLHS typically doesn't interfere with the fetal blood supply which allows for normal somatic growth and tolerance of the defect; although as a result of the retrograde perfusion of the upper body children with HLHS can have a reduced head circumference and/or brain volume.[7]

Clinical presentation can vary slightly but most infants with HLHS will show signs of cyanosis and tachypnea after birth. In some cases, the cyanosis will be mild and not noticed right away but will show signs of cardiogenic shock around day 3-7 of life as a result of the ductus arteriosus closing. Infants who have interatrial restriction will present immediately after birth with respiratory failure with no delay.[7] As a result of the severity of this disease, physical exam and diagnosis will occur either immediately or very shortly after birth. Infants that immediately show signs of cyanosis will present with an increased right ventricular impulse and a systolic murmur, an oxygen saturation level below 94%, and if the ductus arteriosus is restrictive then peripheral pulses may be diminished or absent. [7] In an infant that shows signs of shock at 3-7 days of life it is important to consider HLHS because it

can have a similar clinical picture to neonatal sepsis. Echocardiography is the main diagnostic tool when evaluating neonates and children with HLHS to gain information about the degree and location of the defects. Echocardiography also helps to evaluate interatrial communication which can rule out potential obstruction and looks at pulmonary venous drainage. Cardiac catheterization is typically not used for diagnosis of HLHS but may be used to perform interventional procedures. MRI is also not routinely used for diagnosis or management in children with HLHS but may be used later on for follow up care. CT is not routinely used but may provide similar information instead of cardiac catheterization that can be used for surgical planning. [7]

Prenatal Diagnosis and Counseling

Echocardiography is the primary technique for prenatal diagnosis of congenital heart disease, including Hypoplastic left heart syndrome. This imaging allows detailed assessment of cardiac anatomy and function and is typically done around 18 to 22 weeks gestation. These imaging modalities can be performed earlier around 12 to 14 weeks in high-risk pregnancies and have a repeat one done later on.[8] The American Heart Association recommends fetal echocardiography for pregnancies with certain risk factors including maternal diabetes, autoimmune conditions, drug or toxin exposure, abnormalities noted in umbilical-placental development, and know or suspected genetic conditions. Serial imaging is also recommended in 2-to-8-week intervals until around 34 to 36 weeks especially for conditions like HLHS.

Early diagnosis is critical from a medical preparedness standpoint as well as a psychological standpoint for the family of the infant. Early diagnosis allows for prostaglandin to be available in the delivery room for the fetus, planning of cardiac surgeries, and able to prepare for additional interventions the baby may need in the delivery room. Counseling should be provided as soon as possible so the family understands the condition as well as future steps. The American Heart Association highlights the importance of addressing parental stress and anxiety, offering psychosocial support, and involving palliative care. There should be a multidisciplinary approach.[9]

Postnatal Management Strategies

The initial treatment of HLHS is to maintain the patency of the ductus arteriosus using prostaglandin, this should be done in the delivery room or as early as possible. Intubation should be avoided if possible. In infants that go into cardiogenic shock after the ductus arteriosus closes high doses of prostaglandin should be used to reopen it. Other supportive measures included correction of acidosis, catecholamine support, ventilation, and antibiotic therapy should be started. In cases where the interatrial septum is restrictive, a balloon septostomy may be performed, which is a case in which cardiac catheterization would be used.[7]

Surgical Treatment Options

The three-stage palliative surgery is the Norwood Operation and consists of the Norwood procedure (Stage I), the Glenn procedure (Stage II), and the Fontan procedure (Stage III).

The Norwood procedure is typically performed at days 2-5 of life and involves reconstructing the aorta and pulmonary artery to establish unobstructed systemic perfusion, balance pulmonary perfusion, and unobstructed venous return. The goal of this procedure is for the right ventricle to empty into the new aorta. The mortality rate for this operation can range from 5% to 40% depending on surgeon's experience, associated anomalies, and other factors. There is also a risk of interstage mortality which is around 5-10%.[7]

The second stage is performed around 4-6 months of life and involves ligating the shunt from the first procedure, repairing distortions of the pulmonary tree and performing a bidirectional Glenn anastomosis. This unloads the ventricle, but cyanosis can still persist. The second stage carries an additional mortality risk of about 5-10%.

Stage three of the Norwood operation is the Fontan procedure and is performed around age 2-3 years of life. It involves using an extracardiac conduit from the inferior vena cava to the underside of the right pulmonary artery in order to further reduce cyanosis and achieve nearly normal saturations. There is an estimated survival rate of 70=85% at 10 years post operation. [7]

Alternative and Adjunct Therapies

Cardiac transplantation is an alternative treatment strategy for infants with HLHS, but it is normally limited. There is a scarcity of organ donors especially outside of the Unted States. In addition, heart transplantations carry their own risks associated and there is no guarantee of success. These risks include rejection, infection, renal failure, graft coronary artery disease, and posttransplant lymphoproliferative disease. [7]

Some places utilize a hybrid approach which is called the "Giessen" approach which is used instead of the Norwood I procedure. The Giessen approach uses cardiac catheterization procedures to maintain systemic perfusion, banding of the pulmonary arteries to reduce pulmonary blood flow and development of pulmonary hypertension and ultimately unrestricted interatrial communication. [7]

Mechanical circulatory support (MCS) is used when waiting for a transplant, as a bridge for recovery, or in cases of advanced heart failure or cardiogenic shock. The devices that can be used in infants and small children for mechanical circulatory support include pulsatile devices and continuous flow devices. Device selection and timing needs to be determined based on the individuals anatomy, physiology, and individual needs.[10]

Long-Term Outcomes

Children born with hypoplastic left heart syndrome are at higher risk for long-term neurological and psychosocial impairments. The risk of these impairments is further increased in patients who have received mechanical circulatory support (MCS). The American Heart Association says that MCS is associated with a higher rate of stroke and hypoxic ischemic brain injury which can lead to long term neurological impairment.[11] Patients with HLHS also have increased rates of anxiety disorders, ADHD, and other psychiatric disorders. Most patients with HLHS have other factors that increase and cause challenges for them including a right systemic ventricle and tricuspid regurgitation which worsens long term outcomes. Many patients will develop heart failure and will become candidates for heart transplant later in life. Long term complications known to be associated with the Fontan procedure are atrial arrhythmias, protein-losing enteropathy, liver cirrhosis, hepatocellular carcinoma, plastic bronchitis, and left ventricular dysfunction.

Ethical and Psychosocial Considerations

Family counseling is a critical part of the process for infants with HLHS. Palliative care is an option for patients because of the severity of the condition and potential for serious long term poor outcomes. Palliative care includes stopping prostaglandin, extubating, IV fluid therapy, morphine for pain management, and psychological family support.

Conclusion

In summary, the management of Hypoplastic Left Heart Syndrome has advanced significantly, with current standards centering on staged surgical palliation—most commonly the Norwood, Glenn, and Fontan procedures—or, in select cases, hybrid approaches and transplantation. These procedures have changed HLHS from an almost certainly fatal diagnosis to one with increasing survival, though long-term outcomes remain an area of active investigation. Early and accurate diagnosis, particularly through fetal echocardiography, is essential for optimizing management and counseling,

timely intervention, and improved outcomes. Individualized treatment plans are essential for optimizing survival and quality of life. Despite these advances, many challenges are still associated with HLHS, including neurodevelopmental morbidity, psychosocial impacts, and long-term prognosis. Continued research into long-term outcomes, interventional techniques, and genetic and environmental determinants of HLHS is important for further increasing survival and good outcomes for patients. Interdisciplinary collaboration among cardiologists, surgeons, intensivists, and other health professionals is vital to advancing care and improving the lives of patients with HLHS.

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