Hypoplastic left heart syndrome

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Abstract

Hypoplastic left heart syndrome (HLHS) represents a severe spectrum of congenital cardiac malformations, which lead to systemic blood supply being dependent on patency of the arterial duct after birth. Antenatal diagnosis, advances in postnatal care and modifications of surgical palliation strategies have led to improved outcomes with the majority of patients born in the current era being expected to live into adulthood. This review discusses the surgical treatment strategies for HLHS and the wider implications these may have on the wellbeing of patients and families. The 'Fontan' circulation constitutes a common endpoint for HLHS patients who often experience physiological circulatory compromise rather than pump failure of the univentricular heart. Heart transplantation has been performed in HLHS, but donor shortage in countries such as the United Kingdom make this a viable option only for a few patients. Protein losing enteropathy and plastic bronchitis may be signs of physiological failure in 'Fontan' patients and require specialist input. Family-centred care should enable paediatricians to build relationship outside specialised cardiac centres and optimise healthcare experiences and outcomes through multidisciplinary care systems for these vulnerable patients.

Keywords aortic atresia; congenital heart disease; Norwood procedure; univentricular circulation

Prevalence and definition

Hypoplastic left heart syndrome (HLHS) encompasses a spectrum of complex congenital cardiac lesions. This term was first used by Noonan and Nadas and the reported incidence of HLHS ranges in different reports from 0.16 to 0.36 per 1,000 live births with a male predominance. Physiologically, HLHS describes the case where systemic circulation is dependent on the morphologically right ventricle due to aortic atresia or severe hypoplasia of the aortic valve and arch. In day-to-day clinical practice, there often is a less stringent differentiation, which has led to the term HLHS being applied to a wider spectrum of cardiac malformations that lead to left ventricular hypoplasia such as in the setting of severe mitral valve hypoplasia or mitral atresia.

Fetal diagnosis and management

Fetal diagnosis of HLHS can usually be suspected during the first, and confirmed during the second trimester. Regional variations in antenatal diagnosis rates may reflect local concentration of specialised fetal medicine services and differences in fetal echocardiography training standards. Fetal diagnosis allows for appropriate counselling and planning of postnatal care with reduced likelihood of neonatal circulatory collapse, which should ultimately lead to improved outcomes.

As pulmonary vascular resistance remains higher than systemic vascular resistance during fetal life, the right ventricle maintains systemic perfusion through the arterial duct *in utero*. Pulmonary venous blood flow will be directed from left to right across the atrial septum via the foramen ovale into the right atrium. This pattern of flow effectively bypasses the left side of the heart. Restriction to this flow can lead to pulmonary venous congestion and pathological changes to the pulmonary vasculature resulting in irreversible pulmonary hypertension after birth.

Embryologically, normal development of the aortic arch in fetal life is dependent upon the flow of blood the aorta. Thus, severe aortic stenosis may progress to aortic atresia and HLHS during fetal life. This has led to fetal catheter intervention programmes in a small number of cardiac centres around the world. Fetal balloon dilatation of the stenotic aortic valve has been performed with the aim of enabling better growth of left heart structures. Similarly, stenting of a restrictive atrial septal defect has been performed to prevent pulmonary hypertension in selected cases. Short-term results of fetal interventions have been promising, but patient selection and timing of intervention remains challenging. Maintaining local expertise in this evolving field of fetal cardiac interventions is difficult due to the small numbers of cases.

If left untreated, HLHS will lead to death in infancy. Pulmonary oedema and symptoms of congestive heart failure due to an imbalance of pulmonary and systemic circulations will compromise organ functions and lead to circulatory collapse commonly within the first month of life. Gradual closure of the arterial duct will result in poor systemic perfusion and subsequent hypoxia. Rarely, pulmonary vascular resistance remains high and the arterial duct patent even without treatment. In these cases survival can be expected to exceed the first month of life, but evolving pulmonary hypertension in these cases will ultimately prevent successful heart transplantation or surgical palliation strategies, which both rely on normal pulmonary vascular resistance.

Some genetic syndromes are associated with HLHS and chromosomal anomalies are seen in 4–10% of cases. Significant extra-cardiac anomalies occur in 1% of HLHS and may influence overall clinical management and prognosis. Interestingly, neonatal magnetic resonance imaging of the brain in HLHS before surgical intervention has shown an 'open operculum' in a number of infants. These structural brain findings may play a role in fine motor coordination necessary for sucking and swallowing which may in turn explain why some babies with HLHS experience difficulties establishing oral feeds.

Postnatal presentation and management

If fetal growth is adequate to term and neonatal cardiac management can be facilitated directly after birth, babies with HLHS can be delivered by normal vaginal birth and stabilised outside a specialised cardiac centre. It is necessary to start the baby on a prostaglandin E1 (PGE1) or E2 infusion early to ensure continued patency of the arterial duct. Recommendations regarding enteral feeds vary amongst centres but the increased incidence of necrotising enterocolitis (NEC) in neonates with cyanotic heart

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disease and duct dependent systemic circulations may justify a 'nil by mouth' strategy to minimise the risk of NEC in babies born with HLHS.

Clinical examination directly after birth may reveal normal cardiovascular findings. A ductal murmur will become audible as the pressure gradient between pulmonary and aortic side increases. 12 lead electrocardiogram (ECG) will typically show right heart dominance, which may resemble a normal neonatal ECG pattern. Over the first few days of life, falling pulmonary vascular resistance will lead to the evolution of pulmonary over-circulation with often normal oxygen saturations and signs of tachypnoea. Chest radiograph at this stage will show evidence of evolving pulmonary plethora. Subtle signs of low systemic cardiac output such as cool peripheries, reduced urine output and weak pulses may become evident. This situation necessitates specialist input and may require intensive care management to stabilise the circulation prior to surgical intervention.

On echocardiography, the length of the left ventricle is measured from the annular plane of the atrioventricular valves to the apex of the heart on four chamber view (Figure 1). The size of the mitral and aortic valve annulus and aortic arch will be hypoplastic. Endocardial fibroelastosis (EFE) of the small left ventricle may be present. Arterial duct flow should be maintained with a prostaglandin infusion (either PGE1 or PGE2).

Supplementary video related to this article can be found at http://dx.doi.org/10.1016/j.paed.2014.07.006.

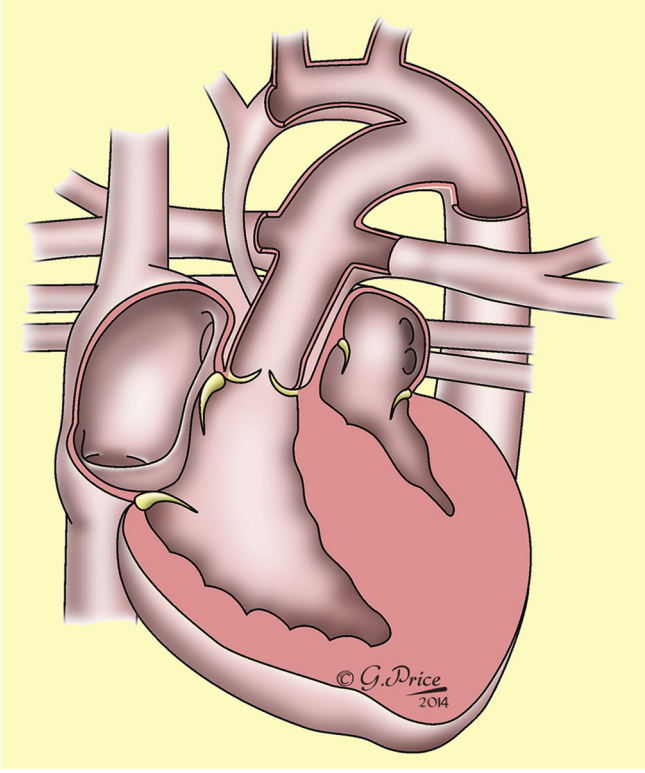

Figure 1 Hypoplastic left heart. The left-sided heart structures are hypoplastic and the arterial duct is maintained patent to sustain life in the immediate postnatal period. Four chamber transthoracic echocardiography view of hypoplastic cleft heart syndrome.) (see video clip).

Heart transplantation

The long-term outlook for children after heart transplantation is improving, but organ shortage remains a problem for infants awaiting heart transplantation in the United Kingdom (UK). Many babies born with HLHS would realistically not survive to transplantation in the current UK setting making alternative management strategies necessary. For older HLHS patients, heart transplantation may be considered when signs of circulatory failure emerge and successful transplantation is technically feasible in children providing the pulmonary vascular resistance remains acceptable. The surgical approach may, however, be technically more challenging due to the altered anatomy of caval, pulmonary and aortic blood flows following often multiple cardiac procedures.

Surgical palliation strategies

All patients undergoing surgical palliation strategies for HLHS will require long-term anticoagulation. Usually this will be in the form of aspirin. After completion of the 'Fontan' procedure patients may be switched to long-term warfarin to minimise the risk of thrombus formation in the pulmonary circulation, which will be driven by respiratory forces alone. Angiotensin converting enzyme inhibitors have not shown a survival benefit at one year of age in patients after the first neonatal surgical palliation, but may well play a role in selected patients with impaired ventricular function.

Norwood procedure Figure 2

The Norwood procedure for HLHS creates a common right ventricular to aortic outflow tract by dividing the pulmonary trunk from the rest of the pulmonary circulation and anastomosing the proximal section to the ascending aorta. The aortic arch is augmented along its hypoplastic course — usually with homograft material — and the arterial duct ligated. A modified Blalock—Taussig (BT) shunt, typically made of a 3.5 mm PTFE tube (Gore-Tex®), is fashioned from the innominate artery or proximal right subclavian artery to the central right pulmonary artery. The atrial septum is resected to ensure unrestrictive inter-atrial blood flow where necessary. These surgical modifications allow systemic and pulmonary venous blood to flow via the right atrium into the right ventricle and out into the neo-aortic arch. Pulmonary blood flow is exclusively reliant on patency of the modified BT shunt. Balancing systemic and pulmonary circulations is crucial to achieve stability following this complex neonatal surgery and oxygen saturations are expected to be in the range of 70—80% in air.

Sano procedure

Instead of a modified BT shunt, as seen after the Norwood procedure, an approximately 5 mm PTFE tube is positioned between the anterior aspect of the right ventricle and the central portion of the pulmonary arteries during the 'Sano' modification. This requires a ventriculotomy and allows pulsatile flow from the right ventricle to the pulmonary circulation. This has been advocated as improving growth of the pulmonary arterial bed and deemed more physiological than the continuous modified BT shunt flow, which may cause low diastolic pressures in the ascending aorta