

Hypoplastic Right Heart Syndrome

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1 April 2025

Abstract

In the diagnosis and treatment of Hypoplastic Right Heart Syndrome in fetuses and children, there is still an adequate amount of research to be discovered. Hypoplastic Right Heart Syndrome (HRHS) is a rare congenital cyanotic disease process caused by underdevelopment of the right heart during fetal life. Patients may be born with a prenatal diagnosis of HRHS and suffer from a multitude of respiratory complications including cyanosis, shortness of breath, and poor growth. The cause of HRHS remains unknown, but research has correlated the diagnosis of HRHS with hereditary components of congenital heart disease and poor maternal environment. It is crucial for a patient to be treated within the first week of life. There is a three-step surgical process that the patient must undergo by the time they are four years of age. Throughout this literature review, the diagnosis and treatment process will be presented with additional data and statistics.

Background

Hypoplastic Right Heart Syndrome (HRHS) is a rare cyanotic congenital heart condition in which the structures throughout the right side of the heart are abnormally developed. Important structures such as the tricuspid and pulmonic valves are underdeveloped along with the right ventricle appearing small or absent. This is a serious congenital abnormality if left untreated because the right side of the heart is responsible for delivering oxygen-deprived blood to the lungs and pulmonary system for exchange. Research states that the first discussions of pulmonary valve atresia began in 1783. While there is no first case confirmed, HRHS is believed to be first described in the 1850s.¹ The prevalence of HRHS is lower than that of left ventricular hypoplasia.

Pathophysiology and Etiology

During fetal life, the cardiovascular system is the first to develop within the first eight weeks of pregnancy. Hypoplastic right heart syndrome is the result of an interruption in the network's ability to properly form the right ventricle and valvular structures responsible for delivering deoxygenated blood to the lungs. In patients with normal anatomy, blood is brought to the right atrium from the head, neck, and extremities. During the diastolic filling phase, the tricuspid valve opens and allows blood to pass from the right atrium to the right ventricle. When the heart is ready to contract, blood is pushed through the pulmonic valve where blood travels through the pulmonary arteries and to the lungs. The right heart's primary objective is to deliver deoxygenated blood to the lungs so that oxygen-rich blood may travel through the left heart and return to the body. In patients with an underdeveloped or absent pulmonic valve, it is difficult for an adequate amount of blood to travel to the lungs. Once there is a complication in pulmonary circulation, the risk increases for deoxygenated blood to filter through the systemic circulation.

This may result in a series of respiratory-related complications upon birth as the lungs and pulmonary system continue to develop. The etiology of hypoplastic right heart syndrome remains unclear. Some researchers believe that there is a genetic component related to the diagnosis of HRHS. It is also known that the mother's environment, medication use, and dietary patterns directly affect the fetus and their ability to properly develop in utero.²

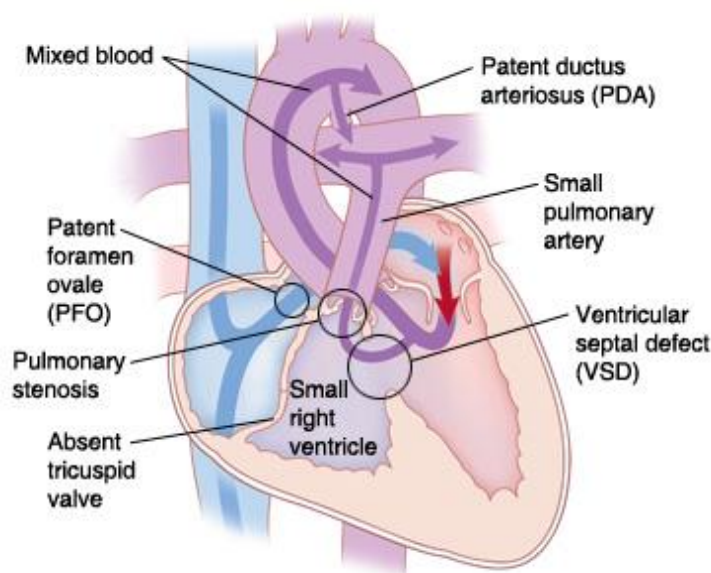


Figure 1: this image is a diagram of the heart, focusing on the dysplastic right ventricle.³

Signs and Symptoms

A common symptom associated with hypoplastic right heart syndrome is hypoxemia due to oxygen-deprived blood mixing with the systemic circulatory system.¹ In most cases of HRHS, the most common clinical sign is the development of cyanosis upon birth. The patient may also present with various respiratory symptoms such as shortness of breath and dyspnea. It is important for the care team to understand the severity of disease for each patient because HRHS lies on a spectrum. For example, a patient with mild HRHS may not start showing symptoms until early adolescence or adulthood. In severe cases, it is possible for the patient to present with

dilated jugular veins, congestive heart failure, liver failure, and respiratory distress shortly after they are born. Research also found a positive correlation between HRHS and clubbed hands, as well as exertional dyspnea.⁴

Echocardiographic findings

The first diagnostic tool to monitor and evaluate hypoplastic right heart syndrome is fetal and pediatric echocardiography. Other recommended diagnostic imaging modalities may include cardiac MRI, X-Ray, CT angiography, and cardiac catheterization.⁴ Echocardiography is a non-invasive and portable ultrasound examination of the heart and its structures. Compared to other imaging modalities, echocardiography is relatively safe and does not require the use of radiation. Hence, why it is the safest way to follow and monitor pregnancies and young adolescents. As hypoplastic right heart syndrome is the underdevelopment or absence of right heart structures, echocardiography has the ability to visualize and measure the severity of the abnormalities during fetal life and after the child is born. On a normal echo, the right ventricle and atrium are low pressure chambers that are separated by the tricuspid valve. In cases of HRHS, the right ventricular cavity will be reduced with hyperechoic regions along the walls that indicate trabeculation and hypertrophy. Since the dimensions are significantly reduced in the right ventricle, stroke volume decreases along with cardiac output. In an apical four chamber view, the right ventricular base will measure twice as close to the left ventricular apex than the mitral valve base.⁵ Within the interventricular septum, it is common to detect a ventricular septal defect with color and spectral Doppler. In HRHS, the tricuspid valve will appear atretic on 2D. The valve leaflets are difficult to evaluate throughout the cardiac cycle due to its inability to properly open and close. Because of this, systolic function measurements like TAPSE and tissue Doppler will be abnormal. In normal systolic function, TAPSE measurements that are less than

17mm indicate poor systolic function. Another parameter used in the evaluation of right ventricular function is tissue Doppler of the lateral free wall. Anything measuring less than 0.095 m/s reflects reduced or worsening function.⁵ Pulmonic valve atresia is a common finding in the diagnosis of HRHS and contributes to the reason why blood is unable to reach the lungs. Echocardiographic findings of pulmonary atresia include an absent pulmonic valve orifice, hypoplastic branch pulmonary arteries, and cessation of flow with color Doppler.⁶ Along with this, the right atrium will appear dilated which indicates a volume and pressure overload. The right heart is intended to be a low pressure system, which is why reduced function and pulmonary hypertension result when pressure elevates.⁵



Figure 2: the hypoplastic right ventricle is shown in an apical 4-chamber view on fetal echocardiography.⁷

Differential Diagnosis

Hypoplastic right heart syndrome is closely associated with other congenital disease processes of the right heart including pulmonic stenosis, ventricular septal defects, Ebstein's anomaly, and Tetralogy of Fallot. Congenital pulmonic stenosis indicates that there is an obstruction of the right ventricular outflow tract by narrowing of the pulmonary artery, displacement of the pulmonic valve, or complete absence of the outflow tract. Clinical symptoms of congenital pulmonic stenosis include shortness of breath, bluish skin, lower extremity edema, and venous congestion upon birth. In mild cases of congenital pulmonic stenosis, the patient may appear asymptomatic until later in adolescence or adulthood.⁸ Ventricular septal defects (VSD) are abnormal connections in the ventricular septum and are relatively common amongst children and young adults. The reason why ventricular septal defects are closely associated with HRHS is due to the abnormal formation of the right ventricle. If a patient is diagnosed with HRHS, a VSD would not be a bad finding to further evaluate. Pressure in the right side of the heart may be optimized if a patient has a congenital septal defect as the blood will have a chance to mix. If blood is unable to reach the right ventricle, ventricular septal defects offer a left to right shunt in which more flow can reach the lungs.⁶ Ebstein's anomaly is described as an abnormal formation and displacement of the tricuspid valve. This anomaly represents 40% of all tricuspid valve abnormalities in pediatrics. Ebstein's anomaly on echocardiography will show a dilation of the right atrium and tricuspid valve regurgitation. Symptoms of Ebstein's anomaly also include respiratory issues such as cyanosis and dyspnea.⁹ Tetralogy of Fallot is a complex congenital disease process consisting of four defects upon birth. These components include congenital pulmonic stenosis, ventricular septal defects, right ventricular hypertrophy, and a large overriding aorta. Tetralogy of Fallot is closely related to HRHS because it mainly affects the

right side of the heart and proposes a risk of oxygen-poor blood being sent to the rest of the body.¹⁰

Treatment and Prognosis

Treatment options for hypoplastic right heart syndrome consist of multiple surgical corrections to the tricuspid and pulmonary valves. Based on the severity of disease, not all patients may qualify for these interventions and it is the care team's job to correctly navigate the patient's treatment plan. The first stage of HRHS correction is the Norwood procedure and is done within the first week of life. This procedure introduces an atrial shunt by removing part of the interatrial septum to allow for pulmonary and systemic flow return to properly mix before ventricular contraction. The Norwood technique then introduces an artificial conduit to the pulmonary artery to establish a more controlled pulmonary circulation.¹¹ By the time the patient is four to six months old, the Glenn procedure is done to reduce right ventricular load by connecting the superior vena cava directly to the pulmonary artery. This intervention eliminates the need for the shunt placed from

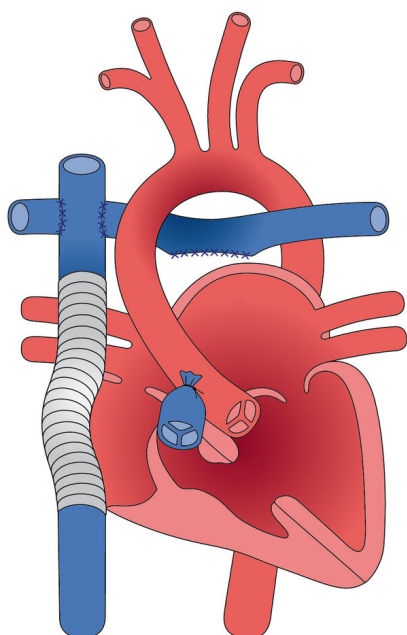


Figure 3: this is a diagram showing the completed Fontan procedure in patients with HRHS.¹³

the Norwood procedure. A common contraindication to receiving this procedure is the development of pulmonary hypertension, worsening cyanosis, and persistent left vena cava.¹² The final step in surgical treatment of HRHS is the Fontan procedure. The Fontan was introduced and developed by Dr. Francis Fontan and Eugene Baudet in 1971 to correct tricuspid valve atresia. The primary goal of this surgery is to completely bypass the right side of the heart by connecting the

inferior vena cava to the pulmonary artery. In patients with single ventricle pathology, the Fontan procedure provides long term solutions before the discussion of a heart transplant. Patients that qualify for this surgery must be at least four years of age, reduced pulmonary pressures of less than 15mmHg, and an ejection fraction of 55% or greater.¹⁴ Surgical complications resulting from HRHS repair may include continued heart failure, cyanosis, pulmonary vascular dysfunction, and low cardiac output. The Norwood procedure proposes the highest risk with postoperative complications including shunt thrombosis, infection, arrhythmia due to surgical interruption of the conduction system, and neurological impairment due to hypoxia.¹¹ The patient may also suffer from recurrent pleural effusion and chylothorax from venous pressure overload. Later in life, there is a risk of developing liver dysfunction and protein-losing enteropathy as a result of intervention. Patients with a Fontan procedure may still be considered for heart transplantation if the procedure is failing later in life.¹⁴ Modern technological advancements have improved the rate of mortality for patients with hypoplastic right heart syndrome. Many patients survive into adulthood with surgical intervention, but must be routinely monitored for the duration of their life to evaluate late complications. Without intervention, neonates have an increased mortality rate as they age and will not live to the stage of adolescence.¹⁵

Conclusion

Hypoplastic right heart syndrome is a congenital cyanotic disease process that affects all right sided structures and pulmonary circulation. Blood is unable to reach the lungs through atretic tricuspid and pulmonary valves, resulting in vascular dysfunction. Research suggests that the cause of HRHS is due to genetic components and disruptions in the mother's environment. If left untreated, the patient will experience heart failure, pulmonary hypertension, worsening cyanosis and dyspnea, and early death. HRHS can be diagnosed prenatally with the use of fetal

echocardiography and closely monitored once the patient is born. Symptoms of hypoplastic right heart syndrome include respiratory distress and cyanosis upon birth, shortness of breath, fatigue, clubbing of the digits, and poor growth. HRHS is closely associated with other congenital disease processes such as Tetralogy of Fallot, Ebstein's anomaly, ventricular septal defects, and congenital pulmonic stenosis. The most common surgical intervention to treat severe HRHS consists of a Norwood, Glenn, and Fontan procedure by the time the patient has reached four years of age or older. So far, research has suggested that the most successful way to treat HRHS is to bypass the heart completely by means of connecting the superior and inferior vena cava to the pulmonary artery. Patients will require lifetime follow-up and possible heart transplantation if the Fontan has failed. In summary, patients may live well into adulthood with minimal complications if surgically treated. There is still much research to be done as the rise in prevalence increases.

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