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CLINICAL UPDATE

Critical Congenital Heart Diseases as Life-threatening Conditions in the Emergency Room

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ABSTRACT

Critical congenital heart disease (CHD) represents a special type of cardiovascular emergency due to the complexity of the associated pathology. In many cases, urgent surgery or catheter-based intervention is required as the condition might be life-threatening. In patients with ductal-dependent lesions, closure of the patent ductus arteriosus (PDA) within the first few days postpartum, can cause sudden clinical deterioration with potentially life-threatening consequences. The diagnostic challenges, clinical presentation and particularities related to the closure of PDA in life-threatening critical CHD are presented.

Keywords: congenital heart disease, emergency room, neonatologic emergencies

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CRITICAL CONGENITAL HEART DISEASES AS LIFE-THREATENING CONDITIONS IN THE EMERGENCY ROOM

Congenital heart diseases (CHD) are the most common congenital disorders in the newborn, affecting about 6–13 per 1000 live births.[1,2] There is an increased risk of CHD with familial, maternal and pregnancy-related factors and these anomalies are more common in some genetic syndromes.

Critical congenital heart disease occurs in approximately two of every 1,000 live births and 25% of newborn with CHD,[3,4] and represents a special type of cardiovascular emergencies due to the complexity of the associated pathology. In many cases, urgent surgery or catheter-based intervention is required in the first year of life.[5–7] The morbidity and mortality increases with any delay in diagnosis and timely referral to a center with experience in treating these pathologies is vital.[8,9]

Understanding fetal circulation and physiologic changes at birth allows us to appreciate why critical heart disease is tolerated in utero but is fatal postnatally without immediate intervention.

This group of lesions includes ductal-dependent and cyanotic lesions, as well as a severe form of CHD that is not depending on patent ductus arteriosus (PDA). The ductal-dependent congenital heart lesions are dependent upon a PDA to supply pulmonary or systemic blood flow or to allow adequate mixing between the two parallel circulations.

DIAGNOSTIC CHALLENGES IN CRITICAL CHD

The majority of infants with critical CHD are symptomatic and consequently are identified soon after birth. However, a few are diagnosed with the condition only after discharge from the neonatology units. In patients with ductal-dependent lesions, closure of the PDA within the first few days postpartum can cause sudden clinical deterioration with potentially life-threatening consequences.[10]

When the disease remains undiagnosed during hospitalization at the time of birth, the risk of mortality is higher than 30%.[8,9,11]

Prenatal diagnosis has an impact on early diagnosis and treatment of critical CHD. Fetal echocardiography has become an important tool in the prenatal diagnosis of critical CHD, avoiding the hemodynamic compromise often accompanying a late postnatal diagnosis.

Many infants with critical CHD show, during the hospitalization at the time of birth, life-threatening clinical findings that require immediate intervention.[1] However, some of them may appear normal on routine examination and develop symptoms after discharge, the timing depending on the underlying lesion and it's dependence upon a PDA.

The most common conditions associated with delayed diagnoses are coarctation of the aorta, interrupted aortic arch, aortic stenosis, or hypoplastic left heart syndrome (HLHS).

CRITICAL CHD AS LIFE-THREATENING CONDITIONS

Neonates with critical CHD can present with life-threatening manifestations as shock, cyanosis, tachypnea, pulmonary edema. All these situations require urgent referral to a pediatric cardiologist.

Infants with ductal-dependent lesions are at increased risk of death and significant morbidity until prostaglandin therapy is initiated to maintain patency of the ductus arterious.

In the case of HLHS, critical aortic stenosis, critical coarctation of the aorta or interrupted aortic arch, infants may present in cardiogenic shock as the ductus arterious closes and sistemic perfusion decreases. In these patients, it is imperative to initiate treatment with prostaglandin E1 to reopen or maintain the patency of the ductus arteriosus.

Infants with a total anomalous pulmonary venous return and significant obstruction at the atrial communication, have impaired systemic perfusion. This is one of the rare situations when lesions might benefit from ductal patency until urgent surgical intervention is instigated. When the obstruction occures within the pulmonary venous pathway, commonly below the diaphragm, the most appropriate management option is the expedient and speedy resort to surgery.

Cardiac lesion, such as HLHS, transposition of the great arteries with restrictive or intact atrial septum, that are unstable in the delivery room, represent abnormalities of oxygen delivery that are not stabilized by prostaglandin E1 alone and require immediate intervention to sustain life.

PDA IN CRITICAL CHD — WHY IS IT SO IMPORTANT?

An important sign of critical CHD is cyanosis secondary to the presence of 3 to 5g/dl deoxygenate hemoglobin. In mild desaturation, pulse oximetry is helpful in detecting patients with cyanotic CHD.

In ductal-dependent lesions, closure of the PDA soon after birth can precipitate profound cyanosis and should therefore be avoided. Such ductal-dependent lesions include:

- Critical obstructive right heart lesions (critical pulmonary valve stenosis, pulmonary atresia with intact ventricular septum), in which pulmonary blood flow is provided via PDA retrograde flow from the aorta; ductus closure would lead to a decrease of the flow to the lung, with secondary progresive severe cyanosis.
- Critical obstructive left heart lesions (HLHS, coarctation of the aorta, interrupted aortic arch, critical aortic stenosis), in which postductal saturation will be lower secondary to the right-to-left flow by PDA, ensuring the lower body circulation. With the closure of the ductus, systemic circulation is compromised, resulting in cyanosis and cardiogenic shock. With a restrictive atrial communication, the cyanosis will be more profound even with ductal patency,[12] while in the case of an adequate atrial communication and patent ductus the desaturation will be minimal.
- Parallel pulmonary and systemic circulation (transposition of the great arteries), when the degree of cyanosis is dependent upon atrial and ductal communication; ductal closure and inadequate atrial commmunication result in profound cyanosis in these cases.
- Ebstein's anomaly with functional pulmonary atresia, in which the neonate may be cyanotic.

In all these cases with severe cyanosis or shock, reopening and maintaining patency of ductus arteriosus by initiation of prostaglandin E1 is imperative.

Non-ductal-dependent lesions with cyanosis that can progress to a critical condition requiring urgent intervention may include:

- total anomalous pulmonary venous connection;
- truncus arteriosus;
- Fallot's tetralogy and tricuspid atresia (depending on the degree of right ventricular outflow tract obstruc-

tion and the presence and size of the ventricular septal defect in tricuspid atresia).

CLINICAL PRESENTATION IN CRITICAL CHD

Differential cyanosis occurs in critical CHD when the lower half of the body is supplied by PDA with deoxygenated blood, with a difference of more than 3% in the oxygen saturation measured at the right hand (preductal) and foot (postductal). Together with critical coarctation of the aorta, interrupted arch, critical aortic stenosis, differential cyanosis also occures in infants with persistent pulmonary hypertension of newborn with structurally normal heart. Pulse oximetry is a simple, safe, feasible test that is acceptable for screening, allowing the identification of critical CHDs that would otherwise go undetected.[13]

Respiratory signs and symptoms in critical CHD include: tachypnea, feeding difficulties, increased work of breathing secondary to increase in pulmonary blood flow with fall of pulmonary vascular resistance shortly after delivery. Cardiac tachypnea in the neonate reflects an increase in pulmonary venous pressure or volume secondary to a large left-to-right shunt, pulmonary venous obstruction or increased left ventricular end-diastolic pressure.[14] All these respiratory signs should be distinguished from those secondary to pulmonary disease.

These conditions may occur in cases of truncus arteriosus or total anomalous pulmonary venous connection, with more severe symptoms in cases of obstruction within the extracardiac pulmonary venous channel or restrictive atrial communication, or PDA in premature infants.

The presence of any symptoms in the neonate with suspected CHD requires urgent refferal to a pediatric cardiologist for diagnosis.

The diagnostic evaluation includes: physical examination, pre- and post-ductal pulse oximetry, chest radiography, electrocardiogram, hyperoxia test useful in differentiation of cardiac from noncardiac causes of cyanosis (a failed hyperoxia test is consistent with an intracardiac mixing lesion and CHD with normal hyperoxia test include coarcation of the aorta, aortic stenosis, isolated interrupted aortic arch). Echocardiography provides a definitive diagnosis of CHD.

In the event of late presentation with an asymptomatic critical CHD during hospitalization at the time of birth, symptoms develop following discharge from the hospital, usually at two weeks of age. The most commonly missed diagnoses immediately after birth are: HLHS, coarctation of the aorta, interrupted aortic arch, aortic stenosis, transposition of the great arteries, pulmonic stenosis.

In these infants, parents notice feeding difficulties, respiratory distress, persistent cough or wheezing. Other clinical manifestations include: central cyanosis or persistent pallor, irritability, increased sweating with feeding and poor weight gain. In these cases evaluation includes a general evaluation and detailed cardiac examination, with heart rate and upper and lower extremity blood pressure, assessment of peripheral pulses and checking for hepatomegaly including, when possible, pulse oximetry. In cases of cyanosis, respiratory symptoms, poor weight gain, with difficulty in feeding, a genetic disorder, an abnormal ECG or chest radiograph, or a physical exam suggestive for CHD, the patient should be reffered to a pediatric cardiologist.

CONCLUSIONS

The outcome of a neonate with critical congenital heart disease depends on timely assessment and accurate diagnosis of the underlying defect and prompt evaluation of potential secondary end-organ damage. However, survival for infants with critical CHD has improved with prenatal diagnoses.

Timely diagnosis and proper perioperative care are crucial in the management of CHD. To ensure the provision of appropriate care, it is essential that there is communication among obstetrician, neonatologist, pediatric cardiologist, surgical and nursing disciplines.

CONFLICT OF INTEREST

None declared.

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