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CASE REPORT

False-Negative Pulse Oximetry Screening for Critical Congenital Heart Disease: The Case for Parent Education

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Abstract The American Academy of Pediatrics recently published evidence-based guidelines for a standardized approach to pulse oximetry as a screening tool for critical congenital heart disease (CCHD). The addition of CCHD screening to the standard newborn examination may lead to earlier detection of CCHD and subsequently decreased morbidity and mortality. We report a case of CCHD with excessive pulmonary blood flow that went undetected during routine newborn screening. Healthcare practitioners and families need to be aware of the limitations of CCHD screening.

Keywords Critical congenital heart disease · Pulse oximetry · Neonatal screening

Introduction

Pulse oximetry allows for a noninvasive, quick, and painless method to screen newborns for CCHD. Screening for CCHD is now part of the recommended uniform screening panel and supported by United States Health and Human Services (HHS) Secretary Kathleen Sebelius and the Advisory Committee on Heritable Disorders in Newborns and Children, the American Heart Association (AHA), the American College of Cardiology (ACC), the American Academy of Pediatrics (AAP) [10], and the March of Dimes. Early detection may result in decreased morbidity and mortality rates in babies that had delayed detection.

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Despite its ability to detect the majority of CCHD cases, pulse oximetry is not without its limitations. Certain cyanotic cardiac lesions can go undetected despite routine CCHD screening in the first few days of life, especially with single lower-extremity measurements. We report such a case.

Patient Presentation

The patient, weighing 3,180 g, was born by way of spontaneous vaginal delivery at 39 weeks' gestation to a group B *Streptococcus*-positive mother. The remaining serology was unremarkable. The patient underwent the standard institutional CCHD screening protocol, which at the time consisted of a single postductal pulse oximetry measurement (96 %). The patient was discharged home after an uneventful newborn nursery course.

At 8 days of life, the infant presented to an outside hospital, by way of ambulance from her pediatrician's office, secondary to a rectal temperature of 33.6 °C. In the emergency room, she was in moderate respiratory distress with poor peripheral perfusion and mottling. Initial arterial blood gas showed severe metabolic acidosis with a base deficit of 25. The patient was intubated; antibiotics were given; and prostaglandins were initiated before transfer to our tertiary institution.

Transthoracic echocardiogram on arrival in the cardiac intensive care unit showed ductal-dependent CCHD consisting of {S,L,L}, double-inlet left ventricle, hypoplastic left-sided right ventricle, juxtaductal coarctation of the aorta, and a small patent ductus arteriosus (PDA). Pulse oximetry in the upper and lower extremities was 90 and 88 %, respectively. At 16 days of life, the patient underwent stage 1 Norwood procedure with right-sided modified Blalock shunt. She was discharged home at 5 weeks of life.

Discussion

CHD is the most common birth defect and occurs in approximately 9/1000 live births [2]. CCHD is defined as a cardiac defect requiring surgical or catheter intervention during the first year of life [13]. CCHD is present in approximately 3/1000 live births and is associated with greater morbidity and mortality [7].

During the last several decades, advancements have been made to improve the diagnosis and treatment of CHD. This had lead to improved outcomes and decreased mortality associated with CHD [1, 11]. A key to these improved outcomes may be the earlier diagnosis of CHD, both antenatally and in the immediate newborn period. Future improvements in screening may lead to earlier detection. Delayed diagnosis, however, can result in significant morbidity and mortality. Pulse oximetry has been proposed as a low-cost, painless, noninvasive test to detect newborns with CCHD.

The AAP and the AHA first issued a scientific statement regarding the role of pulse oximetry in detecting newborns with CCHD in 2009 [9]. They concluded that more "studies in larger populations and across a broader range of newborn delivery systems" were needed before recommending universal screening of all newborns. Two prospective studies of nearly 40,000 newborns, each with screening pulse oximetry results, resulted in convincing data to institute routine newborn screening [4, 12]. In September 2011, the secretary of the HHS recommended that CCHD screening using pulse oximetry be added to the uniform newborn-screening panel.

This case brings attention to the fact that certain cyanotic congenital cardiac lesions can go undetected by routine CCHD screening. Most studies assessing the accuracy of pulse oximetry as a screening tool have focused on the false-positive rates of screening and its burden on families and the medical system. Little focus has been placed on the false-negative rates with routine screening. Meta-analysis of the major studies examining pulse oximetry screening for CCHD reviewed in the 2009 consensus statement from the AHA and AAP shows a false-negative rate of 0.01 % [9].

More recently, a prospective study of 20,055 asymptomatic infants who underwent routine CCHD screening found a similar false-negative rate of 0.02 % from the full cohort with CCHD [5]. Of the six babies who had false-negative pulse oximetry screening, only two were discharged home before an accurate diagnosis was obtained. The other four babies were indentified before discharge because of an abnormal routine examination or high suspicion from antenatal screening. Both babies discharged home ultimately presented with symptoms related to their heart defects as in our case.

Our patient was born at a referring hospital where the standard institutional protocol for CCHD screening at the time was a single postductal pulse oximetry measurement. The infant's CHD was classified as a mixing lesion with ductal-dependent systemic circulation. In the setting of a hypoplastic left-sided right ventricle, there is significantly more pulmonary blood flow (Qp) compared with the aorta (Qs), which increases overall saturation. Some of the blood flow through the main pulmonary artery will shunt right to left across the PDA to the lower extremity where the pulse oximetry measurement was taken. At the time of PDA closure, the patient developed the usually symptoms of low cardiac–output syndrome, including poor peripheral perfusion and the metabolic acidosis that occurs with coarctation of the aorta. Our case illustrates that some conditions with increased Qp and low Qs may fail detection with pulse oximetry screening.

The screening protocol developed by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, in conjunction with the AAP, recommends CCHD screening in the right hand and one foot, thus providing both a preductal *and* postductal saturation [8]. Oxygen saturations of $\geq$95 % in either extremity with a $\leq$3 % absolute difference between upper- and lower-extremity measurements would be considered passing. Obtaining both an upper- and lower-extremity measurement is supported by a study showing an increase in the sensitivity of CCHD screening without a decrease in specificity when this strategy is used [3]. Ductal-dependent systemic blood flow (coarctation) has been one of the conditions most likely to be missed. Adding the peripheral perfusion index (PPI) to screening has been shown to increase detection of coarctation and other ductal-dependent systemic circulation lesions [6].

Conclusion

Pulse oximetry is a simple, noninvasive, and painless test that can be used to detect CCHD during the newborn period. This case highlights some important limitations of pulse oximetry screening for CCHD. Use of upper- and lower-extremity measurements, in combination with PPI, may increase sensitivity of screening. As we move toward universal newborn CCHD screening, the caregiver should be reminded to consider CCHD as a diagnosis in patients who present in the neonatal period with shock and clinical decompensation. Furthermore, families need to know the limitations of CCHD screening and should be educated on the warning signs to look for after their baby discharge home.

Conflict of interest The authors have indicated they have no conflicts of interest relevant to this article.

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