Screening for Critical Congenital Heart Disease in Newborns
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While we have come a long way in our ability to treat critical congenital heart conditions, which affect about 3 of every 1000 live births and account for 30% of all infant fatalities in the United States annually, prenatal sonograms can often identify structural heart disease; however, the sensitivity of congenital heart disease (CHD) detection is highly variable, depending on operator expertise, gestational age, fetal position, and the type of cardiac defect. As a result, prenatal sonography will miss some patients with critical CHD and is known to miss many newborns with simple CHD. Newborns who might benefit from early treatment can often be identified in their first days of life through pulse oximetry screening—a painless, readily available noninvasive examination that is easy to incorporate into newborn assessments. This noninvasive test measures the percentage of hemoglobin in the blood that is saturated with oxygen and pulse rate. The addition of pulse oximetry to fetal ultrasound and physical examination as a screening tool can reduce the chance of overlooking critical congenital heart disease (CCHD) in newborns.

### Physiology of Pulse Oximetry
Oxygen breathed in through the lungs attaches to the red blood cell protein hemoglobin. The newly oxygenated blood then circulates to the tissues. Arterial hemoglobin saturated with oxygen is bright red, and venous hemoglobin with less oxygen is darker. Pulse oximetry uses the principles of spectrophotometry and leverages the light absorption characteristics of hemoglobin and the pulsatile nature of arterial blood flow to determine the saturation of oxygen in the blood. An oximetry probe placed on the foot can transcutaneously measure the blood that is carrying oxygen. The oximetry probe includes a light source, detector, and microprocessor which calculate the differences in the oxygen-rich versus oxygen-poor blood hemoglobin. Red and infrared light–emitting diodes are positioned so that they are opposite their respective detectors and transmit through the toe to the light detector side of the probe (Figure). The oxygen-rich hemoglobin absorbs more of the infrared light, and the hemoglobin with less oxygen absorbs more of the red light. The digital microprocessor in the probe calculates the differences and converts the information to readout of oxygen saturation and heart rate on the pulse oximeter.

### The Ductus Arteriosus in CCHD
Before birth, the aorta and pulmonary arteries are connected by a blood vessel called the ductus arteriosus. This blood vessel is a vital part of fetal blood circulation and normally closes shortly after birth. Many CCHD lesions are ductus dependent. The affected neonate may not be symptomatic during the birth hospitalization because the ductus arteriosus has not yet closed before discharge. Some children with CCHD are given medicine to keep the ductus arteriosus open to maintain blood flow and oxygen levels until surgery can be performed. For example, this may be done if a child is born with a heart defect that decreases blood flow to the lungs or the rest of the body.

### Critical Congenital Heart Disease
The risk of morbidity and mortality in CCHD increases when there is a delay in diagnosis and referral to a tertiary
center with expertise in treating these patients. Babies with CCHDs usually require surgery or catheter intervention in the first year of life and represent more than one third of all congenital heart defects. These structural heart defects are often associated with abnormally low levels of oxygen in the blood, known as hypoxemia, and can potentially be detected using pulse oximetry screening. Screening is usually done when a baby is 24 to 48 hours of age. Newborns with CCHD have a significant risk for death or disability if their condition is not diagnosed soon after birth.

The 7 main CCHD screening targets are:

1. Hypoplastic left heart syndrome
2. Pulmonary atresia
3. Tetralogy of Fallot
4. Total anomalous pulmonary venous return
5. Transposition of the great arteries
6. Tricuspid atresia
7. Truncus arteriosus

Without appropriate screening, some newborns with CCHDs might be missed because the signs of CCHD might not be evident before an infant is discharged from the hospital.

Screening Recommendations

In 2011, Health and Human Services Secretary Kathleen Sebelius adopted the recommendations set forth by the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) to add newborn screening for CCHD to the Recommended Uniform Screening Panel (RUSP) for States. Increasingly, pulse oximetry screening is being adopted by individual hospitals, and more states now require this test, offering another tool for early diagnosis of CCHDs. The American Academy of Pediatrics, American College of Cardiology, American Heart Association, and March of Dimes recommended that screening should be done no earlier than 24 hours after birth and prior to the newborn being discharged from the hospital or birthing center. Infant screenings are usually performed in the well-baby nursery and intermediate care nurseries or other units in which discharge from the hospital is common during an infant’s first week of life. Screening is normally conducted using pulse oximeters that report oxygen saturation and have been cleared by the FDA for use in newborns. Screening should be based on the recommended algorithm developed by the SACHDNC and should be completed by a member of a physician-led team educated in the screening algorithm and trained in pulse oximetry monitoring of newborns. Pulse oximetry is a familiar skill to nurses, and screening newborns is easily incorporated into their routine work flow.

Conclusion

A pulse oximetry screen is considered positive if the measured oxygen saturation <90% and no repeat testing is necessary. The threshold is saturation <95% or a difference of ≥4% in saturation between the right hand and either foot saturation. Any infant with a positive pulse oximetry screen should have a diagnostic echocardiogram. Once identified, newborns with a CCHD can receive special care and treatment that can prevent death or disability early in life. Pulse oximetry screening does not detect all CCHDs, so it is possible to still have a CCHD or other congenital heart defect with a negative screening result. A complete medical and pregnancy history should be taken along with a physical examination, which sometimes can detect CHD before hypoxemia develops. Parents should know the signs and symptoms of CHD, so they don’t miss conditions not picked up by pulse oximetry screening. Early detection of CCHDs provides the opportunity to plan ahead for appropriate delivery and immediate initiation of the appropriate management of the heart defect once the baby is born. There continues to be wide variation and disparity in neonatal detection rates depending on type of lesion and birth hospital. To improve the care and outcomes for neonates with CCHD there is a need for national comprehensive validated registries and improved reporting.

Disclosures

None.

References


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