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SCREENING FOR CRITICAL CONGENITAL HEART DISEASE

On September 21, 2011, the U.S. Secretary for Health and Human Services (HHS) endorsed the addition of screening for Critical Congenital Heart Disease (CCHD) using pulse oximetry to the Recommended Uniform Screening Panel (RUSP) for all infants in the U.S.[1] How and why was this recommendation made? What does it mean for clinical practice and public health? And where do things stand in Oregon now? Read on, and all will be revealed (or at least what we know and what we don't know).

BACKGROUND

Universal newborn screening involves screening every newborn for certain serious genetic, endocrine, and metabolic conditions (e.g. PKU, sickle cell disease), as well as functional disorders that are not apparent at birth. The goal of newborn screening is to reduce infant morbidity and mortality through early identification and treatment. The Advisory Committee on Heritable Disorders in Newborns and Children (hereafter referred to as "the committee") reviews evidence and provides national guidelines on newborn screening that are reviewed and endorsed by the HHS Secretary. States use the RUSP as guidance when establishing their state-specific screening panels. In 2010, the committee recommended adding CCHD screening with pulse oximetry to the RUSP.[2]

WHY CCHD?

Congenital heart disease (CHD) describes a variety of structural defects that are present at birth. These defects change the normal flow of blood through the heart, and may result in hypoxemia (low blood oxygen saturation) during the neonatal period.[3]

CHD can range in severity from asymptomatic to life-threatening. CHD affects about 7 to 9 of every 1,000 live births in the United States and Europe and is the most common cause of death in the first year of life.[3] (Although we don't have Oregon-specific data, with ~45,000 annual births, this extrapolates to about 300 to 400 CHD cases per year in Oregon). When CHD causes severe and life-threatening symptoms requiring intervention, such as cardiac catheterization or surgery, within the first year of life, it is defined as Critical Congenital Heart Disease (CCHD). About one-quarter of neonates with CHD have CCHD[3] (~75-100 cases annually in Oregon). Screening is aimed at identifying and treating newborns with CCHD as early as possible to improve their outcomes.

WHY PULSE OXIMETRY?

Pulse oximetry has several things going for it when it comes to CCHD screening: it's a non-invasive test to estimate hemoglobin oxygen saturation in blood; it's a bedside test; and a positive screen is followed-up by an echocardiogram, just as a physical exam finding would be.[3] It therefore has potential to efficiently detect seven CCHD conditions that require intervention and present most or all of the time with neonatal hypoxemia. These account for about 17–31% of all CHD[3] and were the focus of the committee's review of pulse oximetry screening. They include*:

- Hypoplastic left heart syndrome (HLHS)
- Pulmonary atresia, intact septum
- Tetralogy of Fallot (TOF)
- Total anomalous pulmonary venous return (TAPVR)
- Transposition of the great arteries (TGA)
- Tricuspid atresia
- Truncus arteriosus

EVIDENCE REVIEW

The committee identified 11 studies that addressed the specificity and sensitivity of pulse oximetry screening for CCHD. In all but two, screening was >99% specific (test negative in those without disease) for the seven conditions listed above. Lower specificity (more false positives) appeared to be associated with screening at less than 6 hours after birth and may reflect lower oxygen saturations during the transition to postnatal circulation.[3] The committee's recommended protocol therefore targeted screening on the second day of life (24–48 hours of age or shortly before discharge if <24 hours of age) (see Figure 1, *verso)*.

Sensitivity (test positive in those with disease) was more variable, ranging from 42 to 100%. This was thought to be related to differences in the screened populations (e.g. if the study excluded newborns sent to the NICU or newborns who were symptomatic at birth, or if the institution had a large group of prenatal diagnoses) and the testing strategy employed.[3]

RESULTS

The committee determined that pulse oximetry identifies neonates with CCHD that prenatal ultrasound and postnatal clinical assessment miss. One large screening study of close to 40,000 newborns in Sweden found that, in regions without routine pulse oximetry screening, neonates with ductus arteriosus-dependent circulation were more likely to be discharged undiagnosed (28% vs. 8%) and that neonates diagnosed post-discharge had higher mortality than those diagnosed pre-discharge (18% vs. 0.9%).[3] The committee ultimately recommended that screening combine physical exam and pulse oximetry, as this had the highest sensitivity.

COSTS

Cost estimates for pulse oximetry screening range from less than $5 to $10 per infant, depending on the protocol.[4] This compares favorably with cost estimates for newborn hearing screening, which costs $30 or more per infant.[4] One British study found the cost per timely diagnosis of life-threatening CHD was £4,894 for pulse oximetry.[3] Granted, it's hard to know how that translates to dollars, depending on unit costs for care, exchange rates, or the current state of implosion of the European Union. But it ballparks to around $10,000 in today's dollars, and is likely less costly than complications from undiagnosed CCHD. In

* Images available at: www.mayoclinic.com/health/congenital-heart-defects/CC00026 and www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/Common-Types-of-Heart-Defects_UCM_307017_Article.jsp

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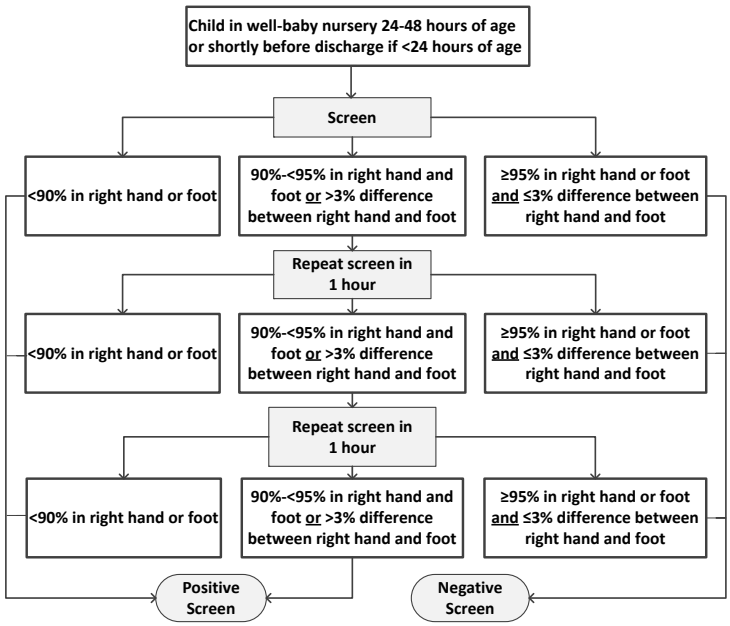
Figure 1. Recommended screening protocol for critical congenital heart disease using pulse oximetry.[4]



fact, according to estimates from the Swedish study mentioned above, the cost of one case of circulatory collapse from undiagosed CCHD may exceed the cost of screening 2,000 newborns.[4]

WHAT NOW?

Not to wax too Rumsfeldian on you, but this is where we have some known knowns and some known unknowns. In the known known category was the need for a standardized screening protocol (Figure 1).[4] Known unknowns include: how this protocol may need to be refined as screening data become available or adjusted for special conditions, such as high altitude settings; how to deal with out-of-hospital births; the true benefit of screening when it's done for the general population; the availability of echocardiography or telemedicine services to follow-up positive screening results, particularly in rural areas; and the exact role of public health in quality assurance and surveillance.

Research at Oregon Health and Science University (OHSU) shows that approximately 55% of hospitals with labor and delivery units in Oregon, Idaho, and SW Washington have implemented universal newborn pulse oximetry screening, with about 15% planning implementation.[5] Ongoing research at OHSU will re-evalutate the extent of implementation following a novel educational approach encouraging pulse oximetry screening.

Despite existing uncertainty, there are a few things we can say for sure, based on the committee's evaluation and recommendations:

- Pulse oximetry appears to be both effective and cost-effective for CCHD screening
- Positive pulse oximetry screening should be followed-up with a comprehensive evaluation for causes of hypoxemia[4]
- In the absence of other findings to explain hypoxemia on pulse oximetry screening, CCHD needs to be excluded on the basis of diagnostic echocardiogram or telemedicine evaluation; a pediatric cardiologist should be consulted, when feasible, before obtaining an echo[4]
- As implementation becomes more complete, primary care providers will need to ensure that all newborns are appropriately screened and receive necessary follow-up.[4]

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