

OREGON PUBLIC HEALTH DIVISION • OREGON HEALTH AUTHORITY

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 ~40,000 annual births, this extrapolates to about 300 to 350 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 known as Critical Congenital Heart Disease (CCHD). About one-quarter of neonates with CHD have CCHD³ (~75-85 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 7 CCHD conditions that require intervention and present most or all of the time with neonatal hypoxemia. These account for about 17-31% of all CHD³ 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 condi-

tions 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 Screening was therefore targeted for the second day of life (≥ 24 hours of age or shortly before discharge if < 24 hours of age) in the committee's recommended screening protocol (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 neonates with CCHD in regions without routine pulse oximetry screening were more likely to be discharged with undiagnosed ductus arteriosus-dependent circulation (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 com-

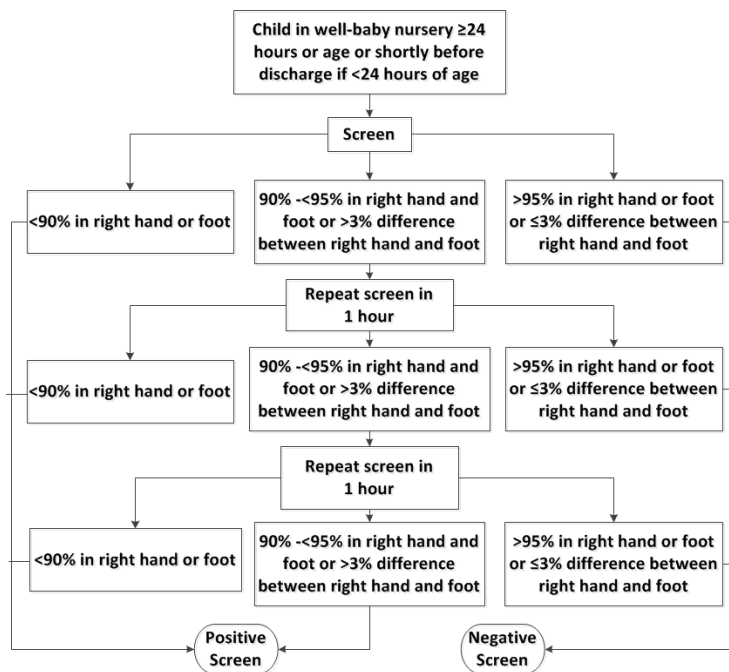
* Images available at: www.mayoclinic.com/health/congenital-heart-defects/CC00026 and www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/Common-Types-of-Heart-Defects_UCM_307017_Article.jsp



If you need this material in an alternate format, call us at 971-673-1111.

IF YOU WOULD PREFER to have your CD Summary delivered by e-mail, zap your request to cd.summary@state.or.us. Please include your full name and mailing address (not just your e-mail address), so that we can purge you from our print mailing list, thereby saving trees, taxpayer dollars, postal worker injuries, etc.

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



plications from undiagnosed CCHD. In fact, the Swedish study mentioned above estimated the cost of complications from one missed CCHD case 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.

We don't have Oregon-specific data on implementation, but anecdotal information from Oregon, Idaho, and SW Washington during 2012 indicates that about 60% of hospitals and birthing centers are currently using pulse oximetry screening, with about 15% of facilities planning to implement it soon.

- Despite existing uncertainty, there are a few things we can say for sure:
- 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 hypoxemia⁴

- In the absence of other findings to explain hypoxemia on pulse oximetry screening, CCHD needs to be excluded on the basis of diagnostic echocardiogram; a pediatric cardiologist should be consulted, when feasible before obtaining an echo⁴
- As implementation becomes more complete, primary care providers will need to ensure that all newborns are appropriately screened and receive necessary follow-up.⁴

REFERENCES

1. Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. HHS Secretary adopts recommendation to add critical congenital heart disease to the Recommended Uniform Screening Panel. September 21, 2011. Washington, DC: US Department of Health and Human Services; 2011. See: www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/correspondence/cyanoticheartsecr09212011.pdf. (Accessed 28 Dec 2012).
2. Centers for Disease Control and Prevention. Newborn screening for critical congenital heart disease: Potential roles of birth defects surveillance programs—United States, 2007-2009. *MMWR* 2012;61:849–53.
3. Knapp, AA, Metterville, DR, Kemper, AR, Prosser, L, Perrin, JM. Evidence review: Critical congenital cyanotic heart disease, Final Draft, September 3, 2010. Prepared for the Maternal and Child Health Bureau, Health Resources and Services Administration. See: www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/nominate-condition/reviews/cyanoticheart.pdf. (Accessed: 28 Dec 2012).
4. Kemper AR, Mahle WT, Martin GR, et al. Strategies for Implementing Screening for Critical Congenital Heart Disease. *Pediatrics* 2011;128:e1259–67.

The **CD Summary** (ISSN 0744-7035) is published fortnightly free of charge, by the Oregon Health Authority, Public Health Division, 800 NE Oregon St., Portland, OR 97232

Periodicals postage paid at Portland, Oregon.

Postmaster—send address changes to:

CD Summary, 800 NE Oregon St., Suite 730, Portland, OR 97232

^{Oregon}
Health
Authority

CD SUMMARY

PERIODICALS
POSTAGE

PAID

Portland, Oregon

If you need this material in an alternate format, call us at 971-673-1111.

If you would prefer to have your *CD Summary* delivered by e-mail, zap your request to cd.summary@state.or.us. Please include your full name and mailing address (not just your e-mail address), so that we can purge you from our print mailing list, thereby saving trees, taxpayer dollars, postal worker injuries, etc.
