Potential—and Potential Pitfalls—of Screening Newborns for Critical Congenital Heart Disease

James A. Taylor, MD; Carrie A. Phillipi, MD, PhD

Before discharge from a newborn nursery, most US infants undergo screening for a diverse list of medical conditions. In addition to the newborn metabolic screen, recommended screening in each newborn includes testing for hearing loss, a serum or transcutaneous measurement of bilirubin level for jaundice, and, for infants born at less than 37 weeks' gestation, a 90-minute “car seat challenge” to evaluate for possible hypoxia. The most recent addition to this menu is the recommendation that newborns undergo screening with pulse oximetry for critical congenital heart disease (CCHD) by assessing preductal and postductal oxygen saturation levels before discharge. That's a lot of screening to be completed during a birth hospitalization averaging less than 48 hours for term infants born vaginally. In addition, this screening is expected to be accomplished in an atmosphere supportive of maternal-infant bonding and the initiation of successful breastfeeding.

Given this crowded agenda, critical assessment of the utility of the various recommended screening tests is appropriate. In this issue, Peterson et al provide useful data on the potential benefit of pulse oximetry screening. The goal of these investigators was to more precisely quantify the proportion of US infants with late detection of CCHD (defined as detection >3 days after birth) who would theoretically benefit from pulse oximetry screening. For their analysis, the authors used data from the National Birth Defects Prevention Study. This database has several advantages compared with administrative databases, including multisite population-based ascertainment of infants with birth defects. Cases were actively identified and verified using multiple medical records as sources. Demographic information was compiled by telephone interview. Most important, the investigators excluded infants with syndromic features, rightfully assuming that these newborns would be more likely to undergo evaluation for cardiac disease than would normal-appearing and seemingly healthy neonates. Infants identified with late detection of nonsyndromic CCHD at autopsy or through diagnostic echocardiography were reasonably labeled as those who might benefit from screening, that is, 29.5% of all children with CCHD in their final analysis.

Because congenital heart disease is the most common birth defect and because a significant proportion of affected children have life-threatening cardiac lesions, the suggestion by Peterson et al that late diagnosis might be avoided in as many as 29.5% of those with CCHD with the use of pulse oximetry screening is potentially quite significant. However, before concluding that this finding provides definitive evidence of the benefit of screening, we should put the results in context by objectively evaluating how well the tenets of a good screening test are fulfilled.

A fundamental problem of the analysis by Peterson et al is the lack of a denominator; the reader is left wondering, 29.5% of what? Estimates of the incidence of CCHD are in the range of 1 to 3 cases per 1000 births, but these incidence rates include newborns with syndromic and nonsyndromic disease, which biases extrapolation of the results from the present study. The underlying assumption that early diagnosis via screening leads to better outcomes than late diagnosis (ie, after children develop CCHD symptoms) presents an additional problem. Peterson et al describe 6 infants included in their analysis who were found at autopsy to have CCHD potentially amenable to treatment that could have been detected by early oxygen saturation screening. We can reasonably assume that these children would have benefited from CCHD screening during their newborn nursery stay. However, whether early diagnosis would have been beneficial to most newborns with CCHD detected late remains unclear. As a
known by the authors, 44.9% of the infants identified with late-detected CCHD had a coarctation of the aorta. Although a portion of such children may have a catastrophic outcome if a ductal-dependent lesion goes undetected in the first few days of life, outcomes in many, if not most, children with coarctation of the aorta are not dependent on diagnosis during the newborn nursery stay.

Before the widespread implementation of pulse oximetry screening in the United States, many clinicians were concerned that screening would result in a high number of false-positive test results, leading to expensive, unnecessary, and anxiety-inducing workups of infants without CCHD. Anecdotal evidence suggests that these concerns are largely unfounded. This impression was confirmed in a report on the statewide screening program in New Jersey. In this report, only 27 asymptomatic newborns without CCHD had false-positive pulse oximetry results of more than 70,000 infants undergo screening, yielding a false-positive rate of approximately 0.04%. False-negative screening results may be more problematic. In a recent meta-analysis, the overall sensitivity of pulse oximetry screening was estimated at 76.5%. Barriers to and problems with large-scale implementation have recently been identified, notably, the need to consider simplification of the currently recommended algorithm. Single-foot oximetry, with sensitivity and specificity statistically similar to those of combined preductal and postductal testing, may prove to be the preferred method of screening for its sheer simplicity.

Pulse oximetry screening for CCHD is probably the right thing to do. The data from the study by Peterson et al provide a high-end estimate of the potential benefit of screening. However, whether the actual benefits outweigh the downsides (overall costs of screening, delayed diagnoses because of false-negative screen results, the costs of evaluation and the heightened anxiety ensuing for families of children with false-positive screen results, and identification of children with CCHD for whom early diagnosis offers no added benefit over later detection) remains an unanswered question. Unfortunately, because pulse oximetry screening has become the recommended and/or mandated standard of care, this dilemma will be difficult to resolve.

ARTICLE INFORMATION
Author Affiliations: Department of Pediatrics, University of Washington, Seattle (Taylor); Department of Pediatrics, Oregon Health and Science University, Portland (Phillipi).
Corresponding Author: James A. Taylor, MD, Department of Pediatrics, University of Washington, PO Box 354920, Seattle, WA 98195 (uncjat@uw.edu).
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REFERENCES