Pulse oximetry screening: do we have enough evidence now?

The early detection of life-threatening, critical congenital heart defects in newborn babies still presents an important clinical challenge. Most defects are amenable to intervention but timely diagnosis (ie, before presentation with cardiovascular collapse or death) is crucial. In high-income countries, examination and, increasingly, antenatal ultrasound have formed the basis of screening, but test accuracy of these procedures is variable and many babies with critical congenital heart defects are discharged before diagnosis.32 Screening with pulse oximetry to detect hypoxaemia associated with most critical congenital heart defects has already been introduced in the USA1 and some Scandinavian countries, and is being considered by European countries including the UK.4

In The Lancet, Qu-ming Zhao and colleagues5 publish the results of a large study including 122 738 babies (120 707 asymptomatic and 2031 symptomatic), from 13 provinces in China. Because China does not have a national screening policy for congenital cardiac defects, individual clinicians were trained to undertake both a clinical assessment and pulse oximetry measurement in all eligible babies as part of the study. All babies who tested positive for either test had a diagnostic echocardiogram, and congenital heart defects were detected in 1071 babies (including 157 critical and 330 major defects). The investigators noted that addition of pulse oximetry to clinical assessment significantly increased the sensitivity for detection of critical congenital heart defects, from 77.4% (95% CI 70.0–83.4) to 93.2% (87.9–96.2).

This is not the first study to report the test accuracy of pulse oximetry screening,6 but is important for two reasons. It is by far the largest study of pulse oximetry screening with more than twice the number of babies screened than in the previous largest study,7 and it is the first to show the feasibility of universal screening and subsequent follow-up (including echocardiography) in a developing country. Although follow-up of false negatives was perhaps not as robust as in some studies8,9 (the researchers did not include interrogation of mortality databases or anomaly registers), test accuracy was consistent with that previously reported.6 These findings would seem to put to rest any remaining concerns about accuracy, and therefore, clinical applicability of pulse oximetry screening.

Critics might challenge this assertion given that the sensitivity for detection of critical congenital heart defects with pulse oximetry was only marginally higher than that for clinical assessment (84% [76.7–88.7] vs 77% [70.0–83.4]), and that pulse oximetry identified only a few more babies (nine) than did assessment. However, the addition of pulse oximetry significantly increased the overall sensitivity for detection of critical congenital heart disease to more than 90%. This added value is a consistent finding in previous studies of pulse oximetry.4 Riede10 described the idea of a diagnostic gap in the detection of critical congenital heart disorders—ie, babies missed by present screening methods. The size of this gap depends on the availability, expertise, and opportunity of ultrasound and clinical assessment, but the addition of pulse oximetry acts as a safety net and consistently reduces the gap to less than 10% irrespective of the proportion of babies identified by other methods.4

One of the major concerns about the introduction of pulse oximetry screening is the number of false positives.4 Findings of this study show that for critical congenital heart defects this number is much lower than those generated as a result of clinical assessment (0.3% [394 of 120 707] false-positive rate for pulse oximetry vs 2.7% [3272 of 120 707] for assessment [murmur]). Importantly, almost 47% of the false positives (180 babies) identified after pulse oximetry had a clinical disorder requiring further intervention or monitoring. These secondary targets are a key additional advantage of pulse oximetry screening because many of the disorders identified, including pneumonia and early-onset sepsis, might be as lethal as critical congenital heart defect if not diagnosed in a timely manner.4

The optimum timing of screening needs to be considered. Earlier screening (within the first 24 h) has a higher false-positive rate,4 as confirmed by results of this study. So when is the best time to screen? The median age at screening in this study was 43 h (range 6–72 h) with an overall false-positive rate of 0.3%. Only 16.6% (20055 of 120 707) of the asymptomatic population was screened within 24 h, but 55% (67 of 122) of the critical congenital heart defects were detected in this timeframe. In previous studies in which investigators screened only after 24 h,8,10 more than half of the babies...
with a critical congenital heart defect presented before screening, some in a collapsed state. In the USA much lower false-positive rates (0·04%) have been reported with screening after 24 h, but with significantly fewer critical congenital heart defects identified (three critical congenital heart defects after more than 72 000 screens). When clinicians consider the optimum time to screen, a lower false-positive rate needs to be balanced against the likelihood of a timely diagnosis.

Should every baby who tests positive undergo a routine echocardiogram? The researchers should be congratulated on their commitment to ensuring that 3898 babies with a positive result from either pulse oximetry or assessment received a diagnostic echocardiogram. However, only 298 (7·5%) of these babies had a major congenital heart defect and only 147 (3·7%) had a critical congenital heart defect. Clinical assessment was responsible for most of the false positives, and if this study’s protocol was introduced nationally, a huge number of unnecessary echocardiograms would be reduced, but could still be a challenge on a national scale, especially in low-income countries. Careful clinical assessment based on all available evidence might be the answer. Our own experience, based on this strategy, is that less than a third of babies positive for pulse oximetry ultimately need an echocardiogram.

The investigators also discuss an important limitation of pulse oximetry screening: the difficulty in detection of critical left heart obstructive defects, especially coarctation of the aorta and interrupted aortic arch (three of seven [43%] and two of five [40%], respectively, detected by pulse oximetry). The researchers acknowledge that, in their cohort, the proportion with these lesions was low (only 15% [22 of 146]) compared with previous studies (28–69%). and this might have contributed to the comparatively high reported sensitivity. Consistent identification of babies with such lesions remains problematic, and staff and parents need to be aware that no present screening test will identify all critical congenital heart defects; however, the combination of the three available tests will identify most defects.

Findings of this important study, along with others, have shown the added benefit of routine pulse oximetry screening. The suggestion that no baby with unexplained persistent hypoxemia should be discharged from hospital is not unreasonable, so where do clinicians go from here? Further trials are unnecessary. Now is the time for professional bodies to review the evidence and consider a pulse oximetry screening protocol that best suits their requirements.

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I declare that I have no competing interests.


