Screening of newborn babies: from blood spot to bedside

In *The Lancet*, Shakila Thangaratinam and colleagues’ report findings from a systematic review and meta-analysis of pulse oximetry screening for critical congenital heart defects in asymptomatic newborn babies. The investigators note that the overall sensitivity of pulse oximetry (76.5%, 95% CI 67.7–83.5) compared with other methods of detection (eg, prenatal ultrasound and routine physical exam), and the low false-positive rate (0·14%, 0·06–0·33), provide convincing evidence for introduction of this technique as a screening method in clinical practice. The investigators conclude that pulse oximetry for such defects meets criteria for universal screening.

These findings also provide some guidance for basic implementation—for example, the false-positive rate was especially low when screening was done 24 h or more after birth (0·05%, 95% CI 0·02–0·12), and screening in the foot alone (postductal) could be as effective as screening in legs and arms (preductal [sensitivity 80·2%, 69·5–87·8 vs 70·0%, 54·9–81·7; p=0·22]). However, important unanswered questions remain about the screening test, including the relative accuracy of different commercial pulse oximeters and how to adjust screening cutoff points for babies born in high-altitude hospitals.

Screening for critical congenital heart defects has been added to the recommended uniform newborn screening panel in the USA and individual states are now adjusting to this expansion. The USA is the only country so far to adopt pulse oximetry screening. To support adoption, the American Academy of Pediatrics, American College of Cardiology, and American Heart Association have developed an implementation plan with a consensus screening algorithm, which Andrew Ewer, one of the authors of this systematic review and meta-analysis, helped develop.

Pulse oximeters are widely available, testing is non-invasive and easy, and the cost per screen is low. However, screening is not simply the application of the first test. Guaranteeing follow-up after a positive screen will be the biggest barrier to adoption of screening. Many hospitals do not have access to paediatric echocardiography, which is needed for newborn babies with a positive screen not attributable to another cause. We have noted that hospitals in the USA already have a system for management of babies with clinical signs of congenital heart defects, which for some involves transfer to another centre for further diagnostic assessment and treatment. These systems can be adapted for the management of newborn babies with an abnormal pulse oximetry screen.

Although the benefit of screening asymptomatic full-term babies is compelling, the benefit of universal screening of all babies is unclear. Findings from an assessment of screening in seemingly healthy newborn babies in the UK found that screening for critical congenital heart defects costs about £6·24 per test and about £24 900 per additional timely diagnosis. By contrast, the value is likely to be low of screening a baby with a low birthweight who is continuously monitored by pulse oximetry in a neonatal intensive-care unit.

The decision to include screening of critical congenital heart defects in screening of newborn babies in the USA, instead of a simple expansion of recommended clinical care, was partly based on the success of newborn screening programmes in ensuring that all newborn babies are tested and have access to coordinated follow-up for diagnosis and treatment. Full engagement of public health programmes will most rapidly and effectively lead to the implementation of this technique. Furthermore, partnership with public health services will help to minimise geographical disparities in access to the screening test and therefore improve early detection of defects. In August, 2011, New Jersey became the first US state to implement state-wide screening of newborns.
babies for critical congenital heart defects. Although many concerns were evident about false-positive screens and the inability to obtain diagnostic follow-up, anecdotal reports suggest that screening has not been overly problematic, and we are aware of two unexpected cases of defects that were identified by screening. A rigorous assessment of the experience in New Jersey is underway.

The looming policy question is how to identify when a bedside screening test should be recommended for routine clinical care or as a component of newborn screening. The importance of this issue will grow as technology allows more screening tests, including those done as part of dried blood-spot newborn screening, to be done cheaply and accurately within individual nurseries. Public health agencies face tremendous financial stress. To increase the capacity of public health systems for point-of-care newborn screening will need novel solutions, including new information systems to allow high-quality support, reporting, and surveillance. Correct calculation of the benefits and costs of screening for critical congenital heart defects will provide an important platform for future point-of-care screening tests as they become available. However, we expect the debate about whether pulse oximetry screening should be part of newborn screening or clinical care to continue until we have better ways to assess explicitly the economic and health outcomes of each approach.

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We declare that we have no conflicts of interest.