

## UK consultation on pulse oximetry screening for critical congenital heart defects in newborns



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Universal screening allows potentially life-threatening diseases to be detected while presymptomatic. UK neonatal mortality is rising and in 2015 was ranked 19th out of 28 European countries.<sup>1</sup> Congenital anomalies and infections are the main causes of UK term neonatal mortality, and most deaths from congenital anomalies are from cardiac defects.<sup>2</sup> Critical congenital heart defects (CCHD) occur in two per 1000 livebirths and, if undetected, can result in collapse and death following closure of the ductus arteriosus.<sup>2</sup> Most such defects are amenable to surgical or transcatheter intervention, but survivors of acute collapse have worse outcomes.<sup>2</sup>

In the UK, antenatal screening detects only 43% of CCHD, with wide regional variation.<sup>3</sup> Routine newborn clinical examination fails to identify up to 45% of CCHD before acute collapse<sup>4</sup> and up to a third of cases present after hospital discharge.<sup>2</sup>

Newborn pulse oximetry screening (POS) detects babies with CCHD before clinical deterioration, is cost-effective,<sup>2</sup> and meets criteria for a screening test.<sup>2,5,6</sup> In 2017, 40% of UK hospitals used some form of POS<sup>7</sup> and more have begun screening since then.

In February, 2019, the UK National Screening Committee (NSC) decided not to recommend routine POS in the UK, citing insufficient evidence of overall improvement in newborn outcomes, concerns about parental anxiety following a positive test, and that harms (delayed discharge and unnecessary investigations and treatment) outweighed benefits. Importantly, they have invited a public consultation on this decision until Aug 9, 2019.<sup>8</sup>

POS improves detection of CCHD compared with examination alone.<sup>4,9</sup> Meta-analysis of 437 000 screened babies showed consistent test accuracy with a sensitivity of 76.3% and a specificity of 99.9% for detection of CCHD.<sup>5</sup> Studies suggest that overall detection of CCHD rises to over 92% with the addition of POS to existing screening tests.<sup>6,9</sup>

The low prevalence of CCHD means large implementation studies are needed to show statistically significant improvements in newborn outcomes. POS is mandatory for all babies in the USA,<sup>10</sup> and in a birth cohort of over 26 million infants, overall mortality from

CCHD was reduced by 33% after introduction of POS in individual states.<sup>11</sup>

POS does generate false-positive results, but these occur ten times less frequently than with clinical examination alone.<sup>9</sup> The rate of false positives with POS varies according to the time of screening.<sup>5</sup> Screening later than 24 h after birth leads to fewer false positives, but up to half of CCHD cases can present before screening.<sup>4</sup> Early discharge from hospital is commonplace in the UK and other countries, so screening in the first 24 h is pragmatic and reduces the risk of acute collapse prior to screening,<sup>4</sup> which is the outcome screening aims to prevent.

In UK studies,<sup>2,6,12</sup> including the 2015 NSC pilot study,<sup>13</sup> the positive test rate was consistently between 0.7% and 0.8%. Importantly, up to 80% of babies who are admitted to a neonatal unit after a positive test have a non-cardiac condition, such as pneumonia or sepsis, that required treatment<sup>8,12</sup> and some of these conditions are potentially life-threatening if treatment is delayed. Concerns about an increase in the demand for echocardiography following a positive test have not been realised, with less than a third of babies with a positive test undergoing this investigation.<sup>8,12,13</sup>

Data from the NSC UK pilot<sup>13</sup> suggest that 70 in every 10 000 babies screened with POS will test positive and 35 will be admitted to a neonatal unit for further investigations. Of these, 28 will have a condition that



requires treatment and only seven will be healthy (true false positive).<sup>8</sup>

Despite these reassuring data, the NSC is concerned about potential overdiagnosis and overtreatment of infants with false-positive screening tests, and therefore convened a workgroup of neonatologists and other health professionals to consider the balance between benefit and risk of POS for these babies. The group concluded that most infants admitted to a neonatal unit after a positive test would benefit and there would be moderate harms relating to delayed discharge and unnecessary investigations and treatment in a minority of babies.<sup>8</sup> The question of whether parental anxiety is unnecessarily increased when a baby has a positive test on screening is important. Psychometric analysis has shown no significant increase in anxiety among mothers of babies with false-positive results compared with mothers of babies with true-negative results.<sup>2,14</sup>

Moreover, it will never be possible to assess the detrimental effect of discharging non-cardiac, hypoxaemic babies who might benefit from early treatment. However, parents should be aware of the potential risk for newborn babies who might be discharged home with suboptimal oxygen levels.

We believe there is clear evidence that early diagnosis of CCHD with POS is beneficial and cost-effective and that potential harms associated with false-positive tests are not serious or common.<sup>2,5,8,12,13</sup> Universal screening is recommended in North America and some European countries<sup>15</sup> and is already used in over 40% of UK hospitals.<sup>7</sup> We think that routine POS should be recommended in the UK. We urge parents, patients, and health professionals to voice their views on this important consultation.

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SO, BS, and AKE were all part of an expert group, convened by the UK NSC in 2018 to review aspects of implementation of possible POS for CCHD. AKE was a clinical adviser to the NSC regarding POS and the clinical lead on the NSC pulse oximetry pilot.<sup>13</sup> None of us have a direct role on the NSC, but we have reported to the committee. We were not involved in the decision not to recommend screening, but our submissions were used as part of the evidence considered in the NSC recommendation that is discussed in this Comment. AKE has received travel and accommodation expenses to speak at scientific meetings from Masimo and Medtronic. JW was an originator of the idea that gave rise to POS and is President of the Resuscitation Council and Vice Chair of the Neonatal Task Force, International Liaison Committee on Resuscitation.

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