A national 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 whilst pre-symptomatic. UK neonatal mortality is rising and in 2015 ranked 19th out of 28 European countries. Congenital anomalies and infections are the main cause of term neonatal mortality, and the majority of deaths from congenital anomalies are from cardiac defects. Critical congenital heart defects (CCHD) occur in 2 per 1000 livebirths and if undetected may result in collapse and death following closure of the ductus arteriosus. Most are amenable to intervention but survivors of acute collapse have worse outcomes.

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

Newborn pulse oximetry screening (POS) detects babies with CCHD prior to clinical deterioration, is cost-effective and meets the criteria for a screening test. In 2017 40% of UK hospitals used some form of POS 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 a lack of evidence of overall improvement in outcome, concerns about parental anxiety, and that harms outweighed benefits. Importantly, they have invited a public consultation on this decision.

POS unequivocally results in improved detection of CCHD compared with examination alone. Meta-analysis of 437 000 screened babies shows consistent test accuracy with a sensitivity of 76.3% and specificity of 99.9% for detection of CCHD. Studies suggest that overall detection of CCHD rises to over 93% with the addition of POS to routine clinical examination.

The low prevalence of CCHD means very large implementation studies are needed to show statistically significant improvements in outcome. POS is now mandatory for all babies in the USA and in a birth cohort of over 26 million infants; Abouket al clearly
demonstrated that overall mortality from CCHD was reduced by 33%, following the introduction of POS in individual states.\textsuperscript{10}

POS does generate false positives (FP), but these occur ten times less frequently than with clinical examination alone.\textsuperscript{9} The rate of FPs with POS varies according to the time of screening.\textsuperscript{5} Later screening (after 24 hours of age), leads to fewer FPs but up to half of cases of CCHD may present prior to screening.\textsuperscript{4} Early discharge is commonplace in the UK and other countries, so screening in the first 24 hours is pragmatic and reduces the risk of acute collapse prior to screening\textsuperscript{4} (the outcome screening aims to prevent).

In UK studies,\textsuperscript{2, 6, 11} including the 2015 NSC pilot study\textsuperscript{8} the test positive rate is consistently between 0.7% and 0.8%. Importantly, up to 80% of babies who are admitted to a Neonatal Unit (NNU) following a positive test have a non-cardiac condition (such as pneumonia or sepsis) which required treatment\textsuperscript{8, 11} 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 one-third of test positive cases undergoing this investigation.\textsuperscript{8, 11}

Data from the NSC UK pilot\textsuperscript{8} suggest that 70 in every 10 000 babies screened with POS will test positive and 35 will be admitted to a NNU for further investigations. Of these, 28 will have a condition that requires treatment and only 7 will be healthy (true false positive).\textsuperscript{8}

Despite these reassuring figures the NSC remains concerned about potential over-diagnosis and overtreatment of infants with FP screening tests and therefore convened a workgroup of senior UK neonatologists and other health professionals to consider the balance between benefit and risk of POS for these babies. The group concluded that the majority of infants admitted to NNU following a positive test would benefit and there would be modest harms (delayed discharge and unnecessary investigations and treatment) in a minority.\textsuperscript{8} The question of whether
parental anxiety is unnecessarily increased in screen positive infants admitted to neonatal units is important. In a rigorous psychometric analysis of mothers of all false positive cases there was no significant increase in anxiety compared with true negatives.\(^2\)

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.

In summary, 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.\(^2,5,8,11\) Universal screening is recommended in North America and some European countries\(^12\) and already practiced in over 40% of UK hospitals.\(^7\) In the face of this, the UK NSC decision is difficult to understand.

We think that routine POS should be recommended in the UK. We urge parents, patients and health professional to voice their views on this crucial consultation.

References

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