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Pulse Oximetry Screening for Detection of Critical Congenital Heart Defects: Why to Bother?

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Editorial

Congenital heart defects (CHD) are the commonest form of congenital malformations affecting 6-8 infants per 1,000 live births [1]. Although congenital heart disease remains an important cause of death in infancy, not all forms of major congenital heart disease will be evident at birth or in the early neonatal period [2,3]. The current screening tools for detection of CHD are: fetal anomaly screening ultrasound and routine examination of newborn prior to discharge from the hospital. Both of these screening tools have a low detection rate and the studies report that up a third of infants with moderate to severe CHD may be discharged home undiagnosed [2,4,5].

Critical congenital heart defects (CCHD) are the most serious form of CHD and defined as conditions requiring surgery or intervention within the first 28 days after birth or result in death. The incidence of CCHD is 2-3 per 1,000 live births [6,7]. Infants with a CCHD are at risk of sudden deterioration following closure of the ductus arteriosus and can result in cardiovascular collapse, acidosis and death. Early diagnosis of such babies would improve survival, as well as reducing the morbidity associated with circulatory collapse prior to recognition of a problem and administration of appropriate treatment.

The infants with CCHD remain well while in-utero because of the fetal circulation. However, majority of infants with CCHD are likely to have some degree of hypoxaemia soon after birth. The infants with moderate or severe hypoxia can be detected clinically, but hypoxia in infants with CCHD is often mild soon after birth making it difficult to detect clinically. Interestingly this can be detected by a very simple test – pulse oximetry. Pulse oximetry is a well-established, simple, visible, fast, accurate and non-invasive method of detecting hypoxaemia. This can objectively quantify the degree of hypoxaemia in asymptomatic infants.

Pulse oximetry screening has been well-studied in over 230,000 infants enrolled in many well designed research studies and randomised controlled trials [6-12]. In 2012, a systematic review and meta-analysis published reported that pulse oximetry screening has a high specificity, moderate sensitivity and a low false-positive rate. This meta-analysis identified 13 studies, of over quarter of a million babies, investigating the use of pulse oximetry screening in detection of CCHDs. The overall sensitivity for pulse oximetry screening was 76.5%, the specificity 99.9% and false positive rate 0.14%. This review concluded that pulse oximetry meets criteria for universal screening for detection of CCHDs6. The addition of pulse oximetry screening to exiting screening tools may increase detection rates for CCHD to over 90% [6-8]. Other studies have established its cost-effectiveness, acceptability to parents and staff and feasibility of implementing screening outside the research context [13-16].

A. Critical congenital heart defects (CCHD)detected by pulse oximetry screening	
CCHD most likely to be detected	CCHD less likely to be detected
D-Transposition of the great arteries (TGA)	Coarctation of the aorta
Hypoplastic left heart syndrome (HLHS)	Interrupted aortic arch
Pulmonary atresia (with intact septum)	Mild Ebstein anomaly
Tetralogy of Fallot	Double-outlet right ventricle
Total anomalous pulmonary venous return	Single ventricle
Tricuspid atresia	Pink Fallot's tetralogy
Truncus arteriosus	
B. Non-cardiac conditions likely to be detected on pulse oximetry screening include: sepsis, pneumonia, persistent pulmonary hypertension of newborn (PPHN) and pneumothorax	

Table 1: Cardiac and non-cardiac conditions likely to be detected on pulse oximetry screening.

In addition, the detection of non-critical CHDs and significant non-cardiac conditions such as respiratory problems or early-onset sepsis is reported as an additional benefit of pulse oximetry screening if performed early [5,6,9,17]. These clinically important conditions contributed to up to two third of the reported false positive groups which reduces the clinically non-significant false positivity to half.

In 2011, the United States Health and Human Services recommended adding pulse oximetry screening to the uniform newborn panel and since then pulse oximetry screening is being established as a national US government policy. In recent years, pulse oximetry screening has been recommended by several European countries including Switzerland, Ireland and Poland. The UK national Screening Committee (UK NSC) has recently completed a pilot study on feasibility and implications of pulse oximetry screening [18].

However, it is noteworthy to understand that not all CCHDs are likely to be detected on pulse oximetry screening (Table 1).

In summary, pulse oximetry screening is feasible, cost-effective, and acceptable to staff and parents. It qualifies the criteria for universal screening programme for detection of CCHD, and recently has been recommended by many countries to include in their national screening programme. Adding pulse oximetry screening as an adjunct to the existing screening tools can help in detecting up to 90% CCHDs. However, parents and health care professionals need to aware that not infants with CCHD can be detected with pulse oximetry screening and this test has its own limitations.

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