EDITORIAL

Clinical Screening for Congenital Heart Disease at Birth: A Long Way to Go

JONATHAN D REICH

Watson Clinic LLP, Lakeland, FL, USA.

There are many ways to express the problem of undiagnosed congenital heart disease (CHD) in infants. Vaidyanathan, *et al*. [1] describe the problem in a statistically detailed manner. I prefer this analysis: for every 700,000 live births, there will be 4,500 infants with CHD, 25% of these children will leave the hospital without a diagnosis, and roughly 30 infants will die from CHD before any cardiac diagnosis is made [2]. This exceeds the total operative mortality from the most complicated operation in CHD surgery, the Norwood [3,4]. If you search hypoplastic left heart syndrome on PubMed you'll get 1856 articles, if you search for undiagnosed CHD you'll get only 67 articles. This lack of research interest is despite, even in an era of extensive fetal diagnosis, the most common diagnosis made at autopsy for infants is CHD-nearly 40% [5].

Improving the diagnosis of CHD has received relatively little attention perhaps because there isn't much more we can do. There is nearly an inverse correlation between the ease of diagnosis and the morbidity of the cardiac lesion [6]. The cardiac diagnosis with the highest likelihood of being diagnosed is pulmonic stenosis which has a mortality rate near zero. The most difficult diagnosis to make is total anomalous pulmonary venous return, which, in one series, had a nursery diagnosis rate of 0% and an undiagnosed mortality rate of 30%. Hypoplastic left heart syndrome had an undiagnosed mortality rate of 100%, but a nursery detection rate of only 40% [7]. Fetal diagnosis has not really reduced the overall mortality rate from undiagnosed CHD, and in most studies really has a very small effect on rates of diagnosis [2,7,8]. It is likely if we spent more time emphasizing the physical exam we would just diagnose milder cases of pulmonic stenosis and other relatively benign forms of CHD; thus we would have little effect on the problem of morbidity and mortality from undiagnosed CHD.

For this reason, the use of pulse oximetry as a means of detecting CHD has become a popular topic. In the last ten years, nearly a sixth of all papers ever written on undiagnosed congenital heart disease have studied pulse oximetry to detect CHD. Like all fields of medicine, these studies have used varied methods rendering any conclusion subject to problems. The first problem is that some papers express the sensitivity of the test as the sensitivity of a combination of both the physical examination and pulse oximetry [1,8,9]. This is superfluous; all newborn infants should have a physical exam. No one writes papers assessing the effectiveness of MRIs when combined with the physical exam; it is assumed the patient had a physical exam before they had an MRI. The purpose of pulse oximetry is to *screen* patients who wouldn't be detected otherwise. An abnormal physical exam means the patient is not a candidate for a *screening* test. More importantly, it makes the study non-reproducible as no two physicians do the same physical exam. Unless the paper describes minute details of the physical exam, for example how many minutes the physician listen and with which stethoscope, no two researchers will obtain the same results.

Alternatively, there are studies which have demonstrated that routine pulse oximetry has

promising results. Riede, *et al*. [8] managed to reduce the presentation of undiagnosed congenital heart disease to 4.4% of CHD with routine pulse oximetry. This compares favorably to other studies which calculated that 25% of infants with CHD leave the nursery undiagnosed [2]. In order to achieve this reduction, Riede, *et al*. like Meberg, *et al*. had nearly 4 times as many false positive pulse oximeter readings as true positives as confirmed by echocardiography [7,8].

Vaidyanathan, *et al*. [1] arrived at similar conclusions, that we did in our study eight years earlier. A few cases of CHD were missed and in general the modality is limited in its ability to detect CHD [9]. The authors report a low sensitivity and a specificity below 90% [1]. This confirms our assessment that although pulse oximetry is inexpensive and without side effects, it is difficult to recommend universal adoption based on the available evidence. Pulse oximeters are designed for long term use in critically ill patients, not spot checks in healthy newborns. The software cycle lengths vary and various inputs are averaged by the machine which produces a number which may not reliably represent the patient's oxygenation [10]. The research frontier is waiting more for engineers to explore, than physicians. Pulse oximeters need to be designed with short cycle lengths, specifically for infants, and produce a paper trail for quality assurance. Otherwise, we're generating numbers from a black box and then reassuring parents their infants are healthy.

When it comes to screening newborns for congenital heart disease it would appear there is not much difference between the developed and developing world. It is difficult to resist the lure of a cheap, safe test which holds out the promise of saving the occasional baby which might be missed. Yet, screening populations is a complicated, expensive endeavor which requires a test with reliability. We have a long way to go to make a reality of the dream of diagnosing every baby with congenital heart disease before they leave the hospital. I am not sure the research on pulse oximetry has made the journey much shorter.

Funding: None.
Competing interests: None stated.

REFERENCES

1. Vaidyanathan B, Sathish G, Mohanan S, Sundaram KR, Warrier KKR, Kumar RK. Clinical screening for congenital heart disease at birth: A prospective study in a community hospital in Kerala. Indian Pediatr. 2011;48: 25-30.
2. Wren C, Reinhardt Z, Khawaja K. Twenty year trends in diagnosis of life-threatening neonatal cardiovascular malformations. Arch Dis Child Fetal Neonatal Ed. 2008;93:F33-5.
3. Ohye RG, Sleeper LA, Mahony L, *et al*. Comparison of shunt types in the Norwood procedure for single-ventricle lesions. New Engl J Med. 2010;362:1980-92.
4. Freedom RM, Black MD, Benson LN, Hypoplastic left heart syndrome. *In:* Allen HD, Gutgesell HP, Clark ED, Driscoll DJ, editors. Moss and Adams' Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult. Philadelphia: Lippincott, Williams, and Wilkins.2001.p.1011-26.
5. Weber MA, Ashworth MT, Risdon RA, *et al*. Sudden unexpected neonatal death in the first week of life: autopsy findings from a specialist centre. J Matern Fetal Neonatal Med. 2009;22:398-404.
6. Wren C. Presentation of CHD in infancy. Arch Dis Child. 1999;80:F50.
7. Meberg A, Andreassen A, Brunvand L, Markestad T, Moster D, Nietsch L, *et al*. Pulse oximetry screening as a complementary strategy to detect critical congenital heart defects. Acta Paediatr. 2009;98:682-6.
8. Riede FT, Wörner C, Dähnert I, Möckel A, Kostelka M, Schneider P. Effectiveness of neonatal pulse oximetry screening for detection of critical congenital heart disease in daily clinical routine-results from a prospective multicenter study. Eur J Pediatr. 2010;169:975-81.
9. Reich JD, Miller S, Brogdon B, Casatelli J, Gompf TC, Huhta JC, *et al*. The use of pulse oximetry to detect congenital heart disease. J Pediatr. 2003;142:268-72.
10. Reich JD, Connolly B, Bradley G, Littman S, Koeppel W, Lewycky P, *et al*. Reliability of a single pulse oximetry reading as a screening test for congenital heart disease in otherwise asymptomatic newborn infants: the importance of human factors. Pediatr Cardiol. 2008;29:371-6.