Screening for Critical Congenital Heart Defects: One Positive Case Over Eight Years. What Does This Mean?

Ana Ferraz[1], Patrícia Silva[2], Gabriela Mimoso[1], Sofia Morais[1]

Port J Pediatr 2020;51:208-11
DOI: https://doi.org/10.25754/pjp.2020.18527

Abstract

Neonatal screening for congenital heart defects is a standard procedure. However, its low efficiency is being questioned particularly due to the robust prenatal screening programs. Nevertheless, some cardiac defects, e.g. total anomalous pulmonary venous return, have a low rate of prenatal diagnosis. In this context, pulse oximetry has an important role in its diagnosis. We report the clinical case of a neonate with a total anomalous pulmonary venous return and normal prenatal scans in which the pulse oximetry positively contributed to the diagnosis. In eight years of screening with pulse oximetry, this was the only case diagnosed at our center.

Keywords: Heart Defects, Congenital/diagnosis; Infant, Newborn; Neonatal Screening/methods; Oximetry/methods; Portugal; Sensitivity and Specificity

Introduction

Congenital heart disease is the most common group of malformations, accounting for 1% of all congenital malformations. Approximately 25% are critical congenital heart diseases.[1-3] Neonates with critical congenital heart diseases, defined as defects requiring surgical or catheter-based interventions within the first year of life, are at high risk for early mortality.[1-4] The risk especially increases when the diagnosis is not established after birth,[3,5] but mortality rates show extensive variability worldwide depending on many factors, e.g. prenatal care, improvements in neonatal cardiac surgery, anesthetic technique, and neonatal and pediatric intensive care.[2] Prenatal detection enables adequate prenatal and preoperative planning as well as improves surgical outcomes and neonatal mortality and morbidity.[6-8]

In the past, screening for congenital heart defects relied on two strategies, prenatal scans and the postnatal physical examination. Despite the advances in fetal scans, there is a great asymmetry in the sensitivity of this tool in the detection of cardiac anomalies between different maternity settings.[9,10] Studies have shown a prenatal critical congenital heart diseases detection rate ranging from 2.4% to 54%.[9,11,12] This variability has been related to the quality of prenatal care, socioeconomic and geographical factors as well as defect type.[9,11] In general, tertiary centers have the highest fetal detection rates. Despite this, some critical congenital heart diseases are difficult to diagnose prenatally, as coarctation of the aorta, transposition of the great arteries, and total anomalous pulmonary venous return, which has detection rates of less than 10%, regardless of the prenatal diagnosis quality.[9,11,13] Physical examination fails to detect more than 50% of the critical congenital heart diseases.[14,15] Hypoxia with pulse oximetry > 80% may not be noticeable on physical examination, especially in dark skin children.[2] Pulse oximetry screening is recommended in order to increase the detection of major heart defects (Table 1) prior to hospital discharge. It is superior to physical examination alone and is increasingly being implemented worldwide.[1,3,16,17] It has a high specificity (99.9%) and moderately high sensitivity (75.6%).[1]

Pulse oximetry screening for critical congenital heart diseases has been used routinely in our tertiary center since 2011. We report the only true positive case in eight years. Some authors have questioned the contribution of neonatal screening in tertiary centers with high prenatal diagnostic rates. Our case report highlights this issue.

Case Report

A female neonate was born from a 26-year-old healthy mother at 38 weeks gestation, after a spontaneous

1. Neonatology B, Maternidade Bissaya Barreto, Coimbra Hospital and University Center, Coimbra, Portugal
2. Pediatric Cardiology Department, Pediatric Hospital, Coimbra Hospital and University Center, Coimbra, Portugal
Corresponding Author
Ana Ferraz,
http://orcid.org/0000-0001-6868-331X
anaibferraz@hotmail.com
Neonatologia B, Maternidade Bissaya Barreto, Centro Hospitalar e Universidade de Coimbra, Rua Dr. Afonso Romão, 3000-602 Coimbra, Portugal
Received: 03/09/2019 | Accepted: 09/02/2020 | Published: 01/07/2020

vaginal delivery with an Apgar score of 9/10/10. The pregnancy was uneventful and the prenatal scans, which were performed at our tertiary center, were described as normal. There was no family history of consanguinity, heart disease, or early deaths.

The physical examination was normal, and the baby was clinically asymptomatic during the entire hospital stay.

The pulse oximetry performed at 48 hours of life revealed a peripheral oxygen saturation of 88% that was unresponsive to oxygen therapy. The neonate was admitted to our intensive care unit for clinical surveillance and cardiac evaluation. Physical examination on admission was normal and at room air the pre- and post-ductal pulse oximetry were 81%-85% (fraction of inspired oxygen, FiO_2, 21%), with a pulse oximetry differential of 5%.

The echocardiogram revealed dilated right heart cavities, a non-obstructed total anomalous pulmonary venous return, with a non-restrictive interatrial communication (*ostium secundum* type) with a right-to-left shunt.

The computed tomography angiography detailed the anomalous drainage, namely the superior and inferior left pulmonary veins and inferior right pulmonary vein draining to the right atrium via a common trunk and the superior right pulmonary vein draining to the superior vena cava. She awaits surgical correction.

Discussion

Pulse oximetry screening has been associated with an increase in the diagnosis rate of critical congenital heart diseases.[1,3,4,17] It is a non-invasive test, easy to perform, and widely available. In addition to the fetal scanning and the standard neonatal clinical examination, it is a useful tool to aid in the detection of critical congenital heart diseases.

Several countries have implemented pulse oximetry screening in their clinical practice and the true impact of the screening has also been studied.[18,19] In Portugal, pulse oximetry screening is performed in many centers, but there are no national guidelines and the impact of its use (*e.g.* algorithms used, true positive rates) is unknown.

At our center, over the past eight years, the present case has been the only one detected by pulse oximetry. This may reflect our relatively high prenatal diagnosis rate (68%). As described in the literature, neonatal critical congenital heart diseases screening will identify more cases in centers where prenatal detection rates are lower.[9,11] However, some congenital heart defects whose prenatal diagnosis can easily be missed, such as total anomalous pulmonary venous return, may be detected with pulse oximetry, thus avoiding a less favorable postnatal course when undiagnosed.[9,11,13]

A point of discussion has been the false positive cases (0.3%-2%).[3,20,21] A positive result in pulse oximetry screening leads to an echocardiogram, consuming time and resources. However, the costs derived from additional medical and surgical care and the reduced survival from a late diagnosis is compensatory. It also leads to parental anxiety, but a systematic review concludes that parents accept the screening rather well, even in the presence of a false positive result.[22] At the same time, pulse oximetry screening allows the detection of other hypoxemic conditions, such as hemoglobin variants as well as respiratory and infectious diseases, which would otherwise go undetected after physical examination (Table 1).[23,24]

Although the positive efficiency is not significant, the low cost of pulse oximetry counterbalances the high cost, both financially and clinically related to undiagnosed critical congenital heart diseases.

In conclusion, the low costs of pulse oximetry screening dictate its maintenance in clinical practice, even in tertiary centers where the prenatal diagnosis of critical congenital heart diseases is standard.

Table 1. Conditions detected through screening for critical congenital heart diseases using pulse oximetry

Primary critical congenital heart diseases screening targets
Hypoplastic left heart syndrome
Pulmonary atresia with intact ventricular septum
Total anomalous pulmonary venous return
Tetralogy of Fallot
Transposition of the great arteries
Tricuspid atresia
Truncus arteriosus
Secondary critical congenital heart diseases screening targets
Coarctation of the aorta
Double outlet right ventricle
Ebstein's anomaly
Interrupted aortic arch
Other single ventricles
Secondary non-critical congenital heart diseases screening targets
Hemoglobinopathy
Hypothermia
Infection/sepsis
Lung disease
Non-critical congenital heart disease
Persistent pulmonary hypertension
Other hypoxemic condition not otherwise specified

WHAT THIS CASE REPORT ADDS

- Pulse oximetry screening increases the diagnostic rate of critical congenital heart diseases and is cost-effective even in tertiary centers with a high prenatal diagnostic rate of cardiac defects.
- Some critical congenital heart diseases, as total anomalous pulmonary venous return and coarctation of the aorta, have a low prenatal detection rate, and they might be detected with pulse oximetry screening.
- False positive cases can be an advantage, allowing for the detection of other diseases, such as infections, respiratory problems, and hemoglobin variants, and they are well tolerated by the parents.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

Funding Sources

There were no external funding sources for the realization of this paper.

Provenance and peer review

Not commissioned; externally peer reviewed

Consent for publication

Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

References

1. Wong KK, Fournier A, Fruitman DS, Graves L, Human DG, Narvey M, et al. Canadian Cardiovascular Society / Canadian Pediatric Cardiology Association position statement on pulse oximetry screening in newborns to enhance detection of critical congenital heart disease. Can J Card 2017;33:199-208. doi: 10.1016/j.cjca.2016.10.006.
2. Olney R, Ailes EC, Sontag MK. Detection of critical congenital heart defects: Review of the contributions from prenatal and newborn screening. Semin Pernatol 2015;39:230-7. doi: 10.1053/j.semperi.2015.03.007.
3. Aranguren Bello H, Londoño Trujillo D, Troncoso Moreno G, Dominguez Torres M, Taborda Restrepo A, Fonseca A, et al. Oximetry and neonatal examination for the detection of congenital heart disease: A systematic review and meta-analysis. F1000Res 2019;8:242 doi: 10.12688/f1000research.17989.1.
4. Mahle WT, Newburger JW, Matherne GP, Smith FC, Hoke TR, Koppel R, et al. Role of pulse oximetry in examining newborns for congenital heart disease: A scientific statement from AHA and AAP. Pediatrics 2009;124:823-36. doi: 10.1542/peds.2009-1397.
5. Fixler DE, Xu P, Nembhard WN, Ethen MK, Candield MA. Age at referral and mortality from critical congenital heart disease. Pediatrics 2014;134:e98-105. doi: 10.1542/peds.2013-2895.
6. van Velzen CL, Haak MC, Reijnders G, Rijlaarsdam ME, Bax CJ, Pajkrt E, et al. Prenatal detection of transposition of the great arteries reduces mortality and morbidity. Ultrasound Obstet Gynecol 2015;45:320-5. doi: 10.1002/uog.14689.
7. Oster ME, Kim CH, Kusano AS, Cragan JD, Dressler P, Hales AR, et al. A population-based study of the association of prenatal diagnosis with survival rate for infants with congenital heart defects. Am J Cardiol 2014;113:1036-40. doi: 10.1016/j.amjcard.2013.11.066.
8. Wright LK, Ehrlich A, Stauffer N, Samai C, Kogon B, Oster ME. Relation of prenatal diagnosis with one-year survival rate for infants with congenital heart disease. Am J Cardiol 2014;113:1041-4. doi: 10.1016/j.amjcard.2013.11.065.
9. Quartermain MD, Pasquali SK, Hill KD, Goldberg DJ, Huhta JC, Jacobs JP, et al. Variation in prenatal diagnosis of congenital heart disease in infants. Pediatrics 2015;136:e378-85. doi: 10.1542/peds.2014-3783.
10. Gardiner HM, Kovacevic A, van der Heijden LB, Pfeiffer PW, Franklin RC, Gibbs JL, et al. Prenatal screening for major congenital heart disease: Assessing performance by combining national cardiac audit with maternity data. Heart 2014;100:375-82. doi: 10.1136/heartjnl-2013-304640.
11. Trimes J, Fruitman D, Zuo KJ, Smallhorn JF, Hornberger LK, Mackie AS. Effectiveness of prenatal screening for congenital heart disease: Assessment in a jurisdiction with universal access to healthcare. Can J Cardiol 2013;29:879-85. doi: 10.1016/j.cjca.2013.04.028.
12. Hill GD, Block JB, Frommelt MA. Disparities in the prenatal detection of critical congenital heart disease. Prenat Diagn 2015;35:859-63. doi: 10.1002/pd.4622.
13. Ailes E, Gilboa S, Honein MA, Oster ME. Estimated number of infants detected and missed by critical congenital heart defect screening. Pediatrics 2015;135:1000-8. doi: 10.1542/peds.2014-3662.
14. Wren C, Richmond S, Donaldson L. Presentation of congenital heart disease in infancy: Implications for routine examination. Arch Dis Child Fetal Neonatal Ed 1999;80:F49-53. doi: 10.1136/fn.80.1.f49.
15. Ainsworth S, Wyllie JP, Wren C. Prevalence and clinical significance of cardiac murmurs in neonates. Arch Dis Child Fetal Neonatal Ed 1999;80:F43-45. doi: 10.1136/fn.80.1.f43.
16. Plana Mn, Zamora J, Suresh G, Fernandez-Pineda L, Thangaratinam S, Ewer AK. Pulse oximetry screening for critical congenital heart defects. Cochrane Database Syst Rev 2018;3:CD011912. doi: 10.1002/14651858.CD011912.pub2.
17. Ewer AK, Middleton LJ, Furmston AT, Bhoyar A, Daniels JP, Thangaratinam S, et al. Pulse oximetry screening for congenital heart defects in newborn infants (PulseOx): A test accuracy study. Lancet 2011;378:785-94. doi: 10.1016/S0140-6736(11)60753-8.
18. Grosse SD, Peterson C, Abouk R, Glidewell J, Oster ME. Cost and cost-effectiveness assessments of newborn screening for critical congenital heart disease using pulse oximetry: A review. Int J Neonatal Screen 2017;3:34. doi:10.3390/ijns3040034.
19. Mukerji A, Shafey A, Shah PS, Sander B, Shah V, et al. Cost-effectiveness of pulse oximetry screening for critical congenital

heart defects in Ontario. J Paediat Child Health 2018;23:e17-8.
20. Hu XJ, Ma XJ, Zhao QM, Yan WL, Ge XL, Jia B, et al. Pulse oximetry and auscultation for congenital heart disease detection. Pediatrics 2017;140:e20171154. doi: https://doi.org/10.1542/peds.2017-1154.
21. Zhao QM, Ma XJ, Ge XL, Liu F, Yan WL, Wu L, et al. Pulse oximetry with clinical assessment to screen for congenital heart disease in neonates in China: A prospective study. Lancet 2014;384:747-54. doi: 10.1016/S0140-6736(14)60198-7.
22. Roberts TE, Barton PM, Auguste PE, Middleton LJ, Furmston AT, Ewer AL. Pulse oximetry as a screening test for congenital heart defects in newborn infants. A cost effectiveness analysis. Arch Dis Child 2012;97:221-6. doi: 10.1136/archdischild-2011-300564.
23. Oster ME, Aucott SW, Glidewell J, Hackell J, Kochilas L, Martin GR, et al. Lessons learned from newborn screening for critical congenital heart defects. Pediatrics 2016;137:e20154573. doi: 10.1542/peds.2015-4573.
24. Jawin V, Ang HL, Omar A, Thong MK. Beyond critical congenital heart disease: Newborn screening using pulse oximetry for neonatal sepsis and respiratory disease in middle-income country. PLoS One 2015;10:e0137580. doi: 10.1371/journal.pone.0137580.

Um Caso Positivo em Oito Anos no Rastreio de Cardiopatias Congénitas Críticas: Que Significado?

Resumo

O rastreio de cardiopatias congénitas críticas complementa as ecografias fetais e o exame clínico do recém-nascido no diagnóstico de algumas anomalias cardíacas congénitas. A importância do rastreio em centros terciários, com altas taxas de diagnóstico pré-natal de cardiopatias, tem sido questionada. No entanto, é também reconhecido que algumas anomalias cardíacas, tais como o retorno venoso pulmonar anómalo total, apresentam uma baixa taxa de diagnóstico pré-natal. O rastreio com oximetria de pulso pode conduzir ao diagnóstico. Relatamos um caso clínico de retorno venoso pulmonar anómalo total diagnosticado após o rastreio positivo das cardiopatias congénitas críticas e cujas ecografias obstétricas foram realizadas no nosso centro. Em oito anos de rastreio com oximetria de pulso, este foi o único caso verdadeiro positivo no nosso centro.

Palavras-Chave: Cardiopatias Congénitas/diagnostico; Oximetria/métodos; Portugal; Recém-Nascido; Sensibilidade e Especificidade; Triagem Neonatal/métodos