Int J Cardiovasc Sci. 2024; 37:e20240030

SHORT EDITORIAL

Is Pulse Oximetry a Reliable Method for Screening for Critical Congenital Heart Disease in Asymptomatic Neonates at 35 Weeks or More of Gestation?

Thais Salim,[1,2] Gabriel Porto Soares[1,2]

Universidade Federal do Rio de Janeiro, Instituto do Coração Edson Abdala Saad,[1] Vassouras, RJ – Brazil
Universidade de Vassouras, Curso de Medicina,[2] Vassouras, RJ – Brazil

Short Editorial referring to the article: Prevalence of Critical Congenital Heart Disease Detected in the Pulse Oximetry Test in Asymptomatic Newborns, ≥35 Gestational Weeks, in a Maternity in Southern Brazil

Congenital cardiovascular malformations (CCVMs) are structural abnormalities of the heart or blood vessels present during fetal life and at birth.[1] From 2010 to 2017, the incidence of CCVMs was estimated at 9.4 cases per 1,000 live births in countries in Southeast Asia and the Northern Hemisphere.[2] During the same period, rates in Brazil ranged from 9.7 (in the northern region) to 62.8 (in the southeastern region) per 1,000 live births,[3] and similar values have been reported by the Global Burden of Disease.[4] Variability in the incidence of CCVMs is explained by diagnostic challenges arising from its broad clinical spectrum, which ranges from asymptomatic to symptomatic heart disease with associated risk of mortality.[4] Approximately 25% of all patients with CCVMs are considered critical and require intervention within the first year of life because of the life-threatening nature of CCVMs and the significant dependence on hospital-based medical care.[3,5]

Congenital malformations are the second leading cause of death in children under 1 year of age and the third leading cause of death in people under 20 years of age in the last decade in Brazil.[3,4,6] Among these malformations, CCVMs account for nearly 50% of malformation-related deaths.[3,4,6,7] Despite their high incidence and significant impact on mortality, CCVMs are deemed preventable causes of death since early therapeutic intervention could alter their outcome.[5] The high mortality rate of CCVMs is due to the lack of prenatal and perinatal diagnoses, which hinders or delays effective treatment and results in death.[1,3,7-9]

During pregnancy, examinations such as second trimester morphology ultrasound and fetal echocardiography aid in diagnosis. However, the sensitivity of these methods may be as low as 80% because of the transition from fetal to neonatal circulation at birth.[7-9] Furthermore, the Brazilian Unified Health System restricts the use of fetal echocardiography to high-risk pregnancies.[9-12]

In 2014, the Brazilian Ministry of Health began recommending routine neonatal screening with pulse oximetry for all asymptomatic newborns (i.e., those without heart murmurs, central cyanosis, and dyspnea as well as those with semiotically normal pulses) at 35 weeks of gestation or older.[5,12] Pulse oximetry should be performed between 24 and 48 hours after birth as a screening method for critical congenital heart disease (CCHD). It has a sensitivity of 70% and specificity of 99.9%.[1,5,12] Due to variability in neonatal oxygen saturation, pulse oximetry might produce false-positive results if performed before 24 hours of life.[5] An abnormal pulse oximetry result warrants continued hospitalization of the newborn and the performance of transthoracic echocardiography for diagnostic confirmation of CCVMs.[5,11,12]

Is pulse oximetry a reliable screening method for CCHD in asymptomatic newborns aged 35 weeks or older? It is a low-cost, noninvasive screening method, but what about its reliability? Witkowski et al. conducted an observational study of 5,667 asymptomatic newborns born at or after 35 weeks of gestation. This cohort was followed for 18 months, from at least 24 hours after

Keywords

Congenital Heart Defects; Newborn Infant; Echocardiography; Oximetry.

Mailing Address: Thais Salim
Universidade Federal do Rio de Janeiro, Instituto do Coração Edson Abdala Saad. Rua Professor Rodolpho Paulo Rocco, 255, 8º andar. Postal code: 27700-000. Cidade Universitária, Campus do Fundão, Vassouras, RJ – Brazil
E-mail: thais.salim@yahoo.com.br

DOI: https://doi.org/10.36660/ijcs.20240030

birth until hospital discharge, within one Brazilian municipality. In this study, pulse oximetry revealed abnormalities in 0.17% of the newborns. Transthoracic echocardiography was performed on all neonates with abnormal pulse oximetry results. Of these, 20% showed no abnormalities and 70% had a patent foramen ovale, an adaptive change from fetal to neonatal circulation that typically resolves spontaneously within the first 3 months of life. Atrial septal defects were found in the remaining 10% of newborns. None of the abnormalities identified were classified as CCHD. Transthoracic echocardiography has proven to be important for the early detection of cardiac morphologic abnormalities in asymptomatic newborns.[1]

According to a meta-analysis of 46,965 neonates, pulse oximetry screening had a sensitivity and specificity of 0.82 (95% confidence interval [CI], 0.53 to 0.95) and 0.97 (95% CI, 0.57 to 1.00), respectively, and transthoracic echocardiography had a concordance of 0.92 (95% CI, 0.89 to 0.94) for CCHD.[13] In a United Kingdom cohort study of 20,055 newborns followed up to 1 year of age, the accuracy of pulse oximetry testing compared with transthoracic echocardiography was estimated to be 13.33% (95% CI, 8.9% to 18.92%) for critical malformation. Pulse oximetry had a sensitivity of 75.0% (95% CI, 53.3% to 90.2%) for identifying critical conditions in the entire cohort. Pulse oximetry showed a sensitivity of 58.3% (95% CI, 27.7% to 84.8%) in the cohort without suspected critical cardiac malformations after prenatal ultrasound.[14]

In brief, pulse oximetry has emerged as an important method for screening asymptomatic newborns. It helps identify candidates for further evaluation with transthoracic echocardiography, allowing early detection of cardiovascular malformations and ensuring prompt access to treatment to reduce mortality in the pediatric population. Pulse oximetry also offers a cost-effective alternative to other fetal screening methods, such as ultrasound and echocardiography. In addition, it is noninvasive, more readily available to the population, and relatively easy to implement, improving accessibility and simplicity in health care settings.

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