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RETROSPECTIVE STUDY OF NEONATAL SCREENING WITH PULSE OXIMETRY IN A SECONDARY CARE MATERNITY HOSPITAL

ESTUDO RETROSPECTIVO DE TRIAGEM NEONATAL DO TESTE DO CORAÇÃOZINHO EM UMA MATERNIDADE DE ATENÇÃO SECUNDÁRIA

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ABSTRACT

Objectives**:** To assess the incidence of abnormal pulse oximetry screening (CCHD screening) results in newborns and analyze its usefulness for the early detection of congenital heart diseases. Methods**:** This was a descriptive, quantitative, and retrospective study, conducted through the review of 3,282 medical records of newborns admitted between 2020 and 2022 in a secondary care maternity hospital in the countryside of Rio Grande do Sul, Brazil. Data were collected by a trained multidisciplinary team using a standardized form extracted from electronic medical records. Information was organized in Microsoft Excel® spreadsheets and analyzed using descriptive statistics, with results presented as absolute and relative frequencies. Newborns who were transferred within 24 hours of life or who required oxygen therapy in the first 48 hours were excluded. Results**:** Of the 3,282 neonates evaluated, 10 (0.3 %) presented abnormal results on pulse oximetry screening. Of these, four cases were confirmed as congenital heart diseases, including patent foramen ovale, tricuspid regurgitation, and ventricular septal defect. The other six cases did not show relevant cardiac alterations after further examinations. Conclusion**:** Pulse oximetry screening proved to be a useful tool for neonatal screening and early detection of congenital heart diseases. Its implementation enables timely referral of newborns with abnormal findings to specialized services, which may contribute to reducing neonatal morbidity and mortality. The findings of this study reinforce the importance of systematic pulse oximetry screening in secondary care maternity hospitals.

Keywords: Critical congenital heart disease; Pulse oximetry screening; Neonatal health; Cardiovascular screening.

RESUMO

Objetivos: Avaliar a incidência de testes do coraçãozinho alterados em recém-nascidos e analisar sua utilidade na detecção precoce de cardiopatias congênitas. Metodologia: Estudo descritivo, quantitativo e retrospectivo, realizado por meio da revisão de 3.282 prontuários de recém-nascidos internados entre 2020 e 2022 em uma maternidade de atenção secundária no interior do Rio Grande do Sul. Os dados foram coletados por equipe multiprofissional treinada, utilizando formulário padronizado com dados extraídos dos prontuários eletrônicos. As informações foram organizadas em planilha do Microsoft Excel® e analisadas

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por estatística descritiva, com apresentação em frequências absolutas e relativas. Foram excluídos recém-nascidos transferidos antes de 24 horas de vida ou em uso de oxigenoterapia nas primeiras 48 horas. Resultados: Dos 3.282 neonatos avaliados, 10 (0,3%) apresentaram alterações no teste do coraçãozinho. Destes, quatro casos confirmaram diagnóstico de cardiopatias congênitas, incluindo forame oval patente, regurgitação tricúspide e comunicação interventricular. Os outros seis casos não evidenciaram alterações cardíacas relevantes após exames complementares. Conclusão: O teste do coraçãozinho demonstrou ser uma ferramenta útil como triagem neonatal para detecção precoce de cardiopatias congênitas. Sua aplicação possibilita o encaminhamento oportuno de neonatos com alterações para serviços especializados, o que pode contribuir para a redução da morbimortalidade neonatal. Os achados deste estudo reforçam a importância da realização sistemática do teste em maternidades de atenção secundária.

Palavras-chave: *Cardiopatias críticas; Oximetria de pulso; Saúde neonatal; Triagem cardiovascular.*

INTRODUCTION

During fetal life, oxygenation occurs in the placenta, with oxygenated blood being transported to the fetus through the umbilical vein. Part of this flow follows the ductus venosus toward the inferior vena cava (IVC), while another part mixes with the portal circulation, supplying oxygen to the liver. In the IVC, oxygenated blood mixes with less oxygenated blood from the lower limbs, entering the right atrium (RA). A significant portion is diverted through the foramen ovale to the left atrium (LA), while the remaining blood stays in the RA and mixes with blood from the head and upper limbs. Blood from the LA is directed to the left ventricle (LV) and ascending aorta, supplying the brain and heart. Less oxygenated blood from the right ventricle (RV) flows into the pulmonary trunk, but due to high pulmonary resistance, part of it is diverted through the ductus arteriosus to the descending aorta, returning to the placenta via the umbilical arteries (MOORE; PERSAUD, 2008).

After birth, placental circulation is interrupted and pulmonary respiration begins, resulting in the functional closure of the ductus arteriosus mediated by bradykinin, followed by the closure of the foramen ovale due to pressure changes between the atria (MOORE; PERSAUD, 2008). These changes make the early identification of critical congenital heart defects (CCHD) essential for proper neonatal management.

Critical congenital heart defects (CCHD) are structural malformations present from fetal life, and their clinical presentation depends on the persistence of the ductus arteriosus after birth. They are classified based on ductal dependency into defects with duct-dependent pulmonary flow (e.g., pulmonary atresia), duct-dependent systemic flow (e.g., hypoplastic left heart syndrome, critical coarctation of the aorta), or parallel circulation (e.g., transposition of the great arteries) (FERREIRA *et al.*, 2023).

Within the first 36 to 48 hours of life, a typical timeframe for hospital discharge of full-term newborns - CCHD signs often have not yet manifested, and cardiac auscultation may be unremarkable. Without early diagnosis, these conditions can quickly progress to shock, metabolic acidosis, cardiopulmonary arrest, or neurological damage (BRASIL, 2014).

Globally, congenital heart diseases account for about 30% of congenital malformations, with an incidence of 8 to 10 cases per 1,000 live births and are responsible for up to 15 % of infant deaths (FERREIRA *et al.*, 2023; VAN DER LINDE *et al.*, 2011). In Brazil, the prevalence and epidemiological profile of CCHD are similar to those in developed countries but face greater challenges regarding access to diagnosis and specialized treatment, contributing to significant neonatal morbidity and mortality (SELIG, 2020).

CCHD management requires early specialized interventions, such as surgery or catheterization, typically performed at cardiac referral centers that are often far from lower-complexity hospitals, such as secondary care maternity units (SOCIEDADE BRASILEIRA DE CARDIOLOGIA, 2009).

Despite the relevance of prenatal morphological ultrasound and fetal echocardiography, only about 30 % of congenital heart defects are diagnosed prenatally in Brazil, due to limited access and high costs (FERREIRA *et al.*, 2023; SELIG, 2020). This reinforces the need for simple, accessible, and universal screening methods to support early diagnosis and treatment.

The "heart test", internationally known as pulse oximetry screening for critical congenital heart disease (CCHD screening), involves measuring peripheral oxygen saturation in one upper limb (pre-ductal) and one lower limb (post-ductal) between 24 and 48 hours of life. The test is considered abnormal if saturation is < 90 % in either limb or if there is a ≥ 3 % difference between measurements. These cases are referred to for cardiological evaluation. Countries such as the United States, the United Kingdom, and Sweden have incorporated CCHD screening into mandatory neonatal screening protocols, resulting in proven reductions in CCHD-related morbidity and mortality and earlier detection before hospital discharge (PLANA *et al.*, 2018; THANGARATINAM *et al.*, 2012; MAHLE *et al.*, 2009).

In Brazil, the test became mandatory through Ordinance No. 13/2014 of the Ministry of Health (BRASIL, 2014). However, challenges persist regarding standardization, coverage, and real impact in secondary care maternity settings - defined in national guidelines as hospitals providing care to pregnant women and newborns without high-risk factors and offering medium-complexity obstetric and neonatal care (SOCIEDADE BRASILEIRA DE PEDIATRIA, 2022).

Given this context, this study aims to determine the incidence of altered pulse oximetry screening results in newborns admitted to a secondary care maternity hospital in southern Brazil between 2020 and 2022. Additionally, it seeks to analyze confirmed CCHD diagnoses among abnormal cases and discuss the test's utility as an initial screening tool and its potential impact on neonatal morbidity and mortality.

METHODOLOGY

This is a retrospective, descriptive, and quantitative study based on the review of electronic medical records of newborns admitted between January 2020 and December 2022. This period was

chosen because it marks the beginning of the systematic and universal documentation of pulse oximetry screening at the studied maternity hospital, classified as a secondary care center located in Santa Maria, in the countryside of Rio Grande do Sul (RS), Brazil.

The studied institution is a public hospital serving exclusively through the Brazilian Unified Health System (SUS).

Pulse oximetry screening was performed by trained nursing professionals, following the Ministry of Health's protocol (BRASIL, 2014). The exam consisted of measuring peripheral oxygen saturation (SpO_2) at two sites: the right upper limb (pre-ductal) and one of the lower limbs (post-ductal), between 24 and 48 hours of life. Normal results were considered as $SpO_2 \geq 95$ % in both limbs with a difference of less than 3 % between them.

Inconclusive results were defined as SpO_2 between 90 % and 94 % or a ≥ 3 % difference; abnormal results were $SpO_2 < 90$ % in either extremity. Inconclusive cases were retested after one hour. If the alteration persisted, the newborn was referred to as transthoracic echocardiography, following institutional protocol.

The study population included all live newborns who underwent pulse oximetry screening during the period. Exclusion criteria included newborns transferred to another facility before 24 hours of life or those who were on oxygen therapy during the first 48 hours after birth.

Collected variables included: gestational age, birth weight, sex, result of the pulse oximetry test (normal, inconclusive, abnormal), need for test repetition, performance of additional exams (such as echocardiography), and final diagnosis of congenital heart disease.

Data collection was carried out by a multidisciplinary team from the neonatology service using a standardized form. Chart reviews were independently conducted by two researchers, and discrepancies were resolved by consensus. Missing data were categorized as "not reported" and excluded from statistical analyses.

Data were organized in Microsoft Excel™ spreadsheets and analyzed using descriptive statistics, with absolute and relative frequencies presented.

The study was approved by the Research Ethics Committee of Universidade Franciscana (approval number 7.018.484) in accordance with Resolution No. 466/2012 of the Brazilian National Health Council.

RESULTS

From January 2020 to December 2022, there were 3,282 live births recorded in a secondary care maternity hospital in the countryside of Rio Grande do Sul, which serves as a referral center for nearby municipalities. All newborns underwent pulse oximetry screening per institutional protocol. Two cases were excluded from the final analysis because they received oxygen

therapy during the first 48 hours of life, according to the established exclusion criteria. Thus, the final sample included 3,280 newborns.

Of the total sample, 3,270 (99.7 %) had normal screening results, and 10 (0.3 %) had altered results, including both positive and inconclusive cases. Among these 10 altered tests, two were classified as positive (SpO_2 < 90 % in any limb or persistent discrepancy after retesting), and eight were initially classified as inconclusive (SpO_2 between 90 % and 94 % or ≥ 3 % difference between upper and lower limbs).

In the group of two positive tests, the first newborn presented a 3+/6+ holosystolic murmur and central cyanosis on physical exam and was referred to an intensive care unit at a higher-complexity hospital. Despite the initial clinical suspicion, no CCHD diagnosis was confirmed. The second newborn underwent transthoracic echocardiography, which revealed a patent foramen ovale, a condition often considered physiological in neonates.

Among the eight inconclusive cases, four normalized oxygen saturation levels after retesting within the first 48 hours and were reclassified as normal. The remaining four cases remained inconclusive and were referred to in echocardiography. One exam showed no cardiac abnormalities, two revealed a patent foramen ovale, and one revealed tricuspid regurgitation associated with an apical ventricular septal defect, compatible with mild pulmonary hypertension.

The detection of a patent foramen ovale in three of the ten altered cases (one positive and two inconclusive) raises discussion about the potential for false positives, particularly given the absence of routine echocardiography in cases with normal screening results. This limitation was addressed in the discussion.

Pulse oximetry screening was repeated after one hour in all initially inconclusive cases, per Ministry of Health protocol. Persistence of findings led to specialized evaluation.

DISCUSSION

Pulse oximetry screening is recognized as an essential tool for CCHD screening in newborns, particularly in secondary care maternity hospitals such as the one analyzed in this study. Various national and international studies show that routine application of the test between 24 and 48 hours of life allows for early identification of oxygenation abnormalities even in the absence of clinical symptoms, enabling timely referral for specialized assessment (MAHLE *et al.*, 2009; PLANA *et al.*, 2018; KOPPEL *et al.*, 2003).

Large-scale studies in countries like Sweden and the UK have reported similar prevalence of abnormal test results. In Sweden, Plana *et al.* (2018) analyzed 38,429 newborns and found 0.23 % of positive or inconclusive results, with a 20 % confirmation rate of heart disease among initially inconclusive cases. In the UK, among 19,860 neonates, 0.9 % had altered results, and 13 % were later confirmed as congenital heart disease (THANGARATINAM *et al.*, 2012; PLANA *et al.*, 2018).

In this study, among 3,280 newborns, only 10 (0.3 %) had abnormal results, consistent with international findings. Of these, four (40 %) had confirmed congenital heart defects after further testing. Unlike many international studies that consider only moderate to severe defects, this study also included mild findings such as patent foramen ovale, often physiological in the early neonatal period (VAN DER LINDE *et al.*, 2011; PINTO JÚNIOR *et al.*, 2015).

The higher confirmation rate (40 %) in this study may be partly due to including mild cases that do not require immediate intervention. If only moderate to severe defects are considered, the confirmation rate would be closer to the international average of around 10 %. This underscores the importance of standardized criteria for defining screening outcomes, a topic still debated in the literature (MAHLE *et al.*, 2009; THANGARATINAM *et al.*, 2012).

Another important factor is that in the studied institution, prenatal screening coverage with morphological ultrasound and fetal echocardiography remains limited due to resource availability and regional logistics. Although most pregnant women undergo at least one obstetric ultrasound, few have access to routine fetal echocardiography, typically only in high-risk situations. Thus, pulse oximetry screening acts as a complementary tool to prenatal screening, enhancing detection of CCHD that might otherwise go unnoticed until hospital discharge (PINTO JÚNIOR *et al.*, 2015; THANGARATINAM *et al.*, 2012; MAHLE *et al.*, 2009).

Despite its benefits, the universal implementation of pulse oximetry screening also presents challenges. Studies suggest it may lead to more diagnostic tests (such as echocardiography), higher healthcare costs, and additional hospitalizations, especially in false-positive or uncertain cases like patent foramen ovale (NARAYEN *et al.*, 2019; THANGARATINAM *et al.*, 2012). In this study, for example, no systematic echocardiographic investigation was performed in all newborns with normal results, limiting assessment of the true false-negative rate.

Key limitations of this study include its retrospective design, reliance on hospital records, absence of echocardiography in all normal cases, and restriction to a single secondary care public maternity hospital, which may limit generalizability.

Nevertheless, results reinforce that combining prenatal screening strategies with pulse oximetry testing at birth can improve outcomes by identifying conditions requiring specialized care early (FERREIRA *et al.*, 2023; THANGARATINAM *et al.*, 2012; PLANA *et al.*, 2018; MAHLE *et al.*, 2009). Brazil's national experience, though still unequal among regions, is moving toward broader access and standardization of pulse oximetry as a neonatal screening tool, in line with international efforts to reduce CCHD-related morbidity and mortality (PINTO JÚNIOR *et al.*, 2015; BRASIL, 2014).

CONCLUSION

Pulse oximetry screening proved to be an effective tool for early diagnosis of congenital heart diseases in the neonatal period, identifying relevant alterations in a small proportion of newborns and enabling timely referral for further evaluation and specialized care.

The findings of this study, aligned with results from countries like Sweden and the UK, highlight the test's potential in secondary care settings, emphasizing its utility, accuracy, and low cost as a non-invasive neonatal screening strategy. Although the incidence of altered results was low - possibly due to the integration of good prenatal practices - maintaining and expanding systematic implementation of the test remains essential to ensure early detection of congenital heart defects and contribute to reducing neonatal morbidity and mortality.

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