

Pulse oximetry as a screening tool for congenital heart diseases

Thzuska Pico Mawyin^{1,a}; Ramón Miguel Vargas-Vera^{1,2,b}; Giomar Viteri Gómez^{3,c}; Jonathan Jaramillo Valarezo^{2,d}; Aischa Castro Gaviño^{2,d}; Edison Burgos Cox^{2,d}

1 Universidad Católica de Santiago de Guayaquil. Guayaquil, Ecuador.

2 Universidad de Guayaquil. Guayaquil, Ecuador.

3 Hospital Universitario de Guayaquil. Guayaquil, Ecuador.

^a Neonatologist; ^b Geneticist; ^c Pediatrician; ^d General practitioner.

ABSTRACT

Objective: To report the results of pulse oximetry screening for congenital heart diseases in healthy newborns during the transitional period between birth and discharge. Those with positive screening results were referred for echocardiographic evaluation aimed at a specific diagnosis.

Materials and methods: A retrospective, descriptive, observational, cross-sectional study. Data were obtained from screening 4,897 newborns who were admitted to the rooming-in area of the Obstetrics and Gynecology Unit at Hospital Universitario de Guayaquil, within the first 24 hours of birth, all diagnosed as healthy newborns. As part of the protocol, cardiac screening was performed on all newborns at admission and discharge. Newborns with saturation levels below 95 % on pulse oximetry underwent consecutive measurements at one-hour intervals, with a total of three readings. Additionally, those with more than a 3 % difference in saturation between preductal and postductal readings were also monitored. Newborns with positive screening results underwent transthoracic echocardiography to evaluate for heart defects. **Results:** Among the 4,897 newborns, 626 (12.8 %) had a positive neonatal cardiac screening, out of which 497 (79.4 %) were diagnosed with some form of structural heart defect. Patent ductus arteriosus was the most frequent finding, with 127 cases (25.55 %). Critical malformations such as aortic aneurysms and coarctation of the aorta were reported in 0.8 % and 1.6 %, respectively. **Conclusions:** Screening for congenital heart diseases using pulse oximetry during the transitional period prior to discharge is essential for the early detection of congenital heart diseases in healthy newborns.

Keywords: Congenital Abnormalities; Heart Defects, Congenital; Neonatal Screening; Oxygen Saturation; Oximetry (Source: MeSH NLM).

INTRODUCTION

Congenital heart diseases (CHDs) are the most common type of congenital malformation. Structural defects occur in 1 % of newborns and are associated with high morbidity and mortality rates after birth, mainly due to delayed diagnosis, which adversely affects neonatal outcomes ⁽¹⁾.

In 2019, Armas López et al. reported a global incidence of 8 per 1,000 live births with CHD and a high associated mortality rate ⁽²⁾. According to Instituto Nacional de Estadísticas y Censos (INEC - Ecuador's National Institute of Statistics and Census), the birth rate in 2020 was 15.2 per 1,000 inhabitants; moreover, fetal deaths due to unspecified congenital malformations ranked seventh, a category that includes complex or critical CHDs (CCHDs) ⁽²⁻⁴⁾.

Multiple studies underscore the importance of screening for CHDs ⁽⁵⁾. Clinical manifestations of critical CHDs—such as cyanosis, tachypnea,

and murmurs—may develop after discharge, even within 48 hours of birth. Consequently, performing pulse oximetry screening before discharge increases the detection rate of cardiac defects, including potentially complex abnormalities ^(6,7). A positive pulse oximetry result—defined as preductal oxygen saturation < 90 % and/or a 3 % difference between preductal and postductal saturation—and prenatal ultrasound diagnosis, although generally effective, identify only about 30 % of structural defects. This underscores the critical role of pulse oximetry at birth ^(8,9).

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommends pulse oximetry screening for CCHDs, in conjunction with laboratory tests during the first 24 hours after birth, to rule out major cardiovascular diseases such as patent ductus arteriosus, transposition of the great arteries, hypoplastic left heart syndrome, and tetralogy of Fallot. These procedures are endorsed by several organizations, including the ACHDNC, the American Academy of Pediatrics (AAP), the

Corresponding author:

Thzuska Pico Mawyin
thzuska@hotmail.com

Received: August 21, 2024

Reviewed: September 17, 2024

Accepted: September 19, 2024



This is an open access article distributed under the terms of the Creative Commons Attribution 4.0 International License (<http://creativecommons.org/licenses/by/4.0/>).

Copyright © 2025, Revista Horizonte Médico (Lima). A publication of Universidad de San Martín de Porres, Peru.

American Heart Association (AHA), and the American College of Medical Genetics (ACMG), prior to hospital discharge ⁽¹⁰⁻¹³⁾.

In Ecuador, no studies have evaluated CHD diagnosis using pulse oximetry, nor has a national algorithm for cardiac screening been established. This gap increases the risk of discharging apparently healthy newborns with asymptomatic cardiovascular diseases. The objective of the present study is to report the results of CHD screening evaluated by echocardiography for definitive diagnosis.

MATERIALS AND METHODS

Study design and population

This was a retrospective, descriptive, observational, cross-sectional study. The study population comprised 4,897 full-term newborns admitted to the Neonatology Department of Hospital Universitario de Guayaquil (HUG), Ecuador, between July 1 and November 31, 2020. Data were obtained from medical records and the vital signs logbook, where screening results and cardiology assessments are documented.

Inclusion criteria were live, full-term newborns hospitalized in the rooming-in area from June to November 2020. Exclusion criteria were newborns with prenatally diagnosed cardiovascular diseases or congenital disorders requiring intensive or intermediate care.

Table 1. Interpretation of preductal and postductal oxygen saturation in newborns

Oxygen saturation criteria	Interpretation
Saturation ≤ 94 % on the right hand and left foot; measurement repeated once or if a 4 % difference between extremities was observed.	Positive
Saturation ≥ 95 % on the right hand and left foot; measurement repeated once.	Negative

Source: AAP, 2014.

Ethical considerations

As this was a retrospective study with no manipulation of variables and no collection of personally identifiable data, informed consent was not required. HUG Ethics and Research Committee approved this research on September 19, 2021.

Table 2. Pulse oximetry screening results

Screening results	Number of cases	%
Positive pulse oximetry screening result	626	12.78
Negative pulse oximetry screening result	4,271	87.21
Total	4,897	100.00

Source: HUG Statistics Department, 2022

Variables and measurements

Healthcare staff were trained in the correct procedure for obtaining vital signs. Cardiac screening was performed upon admission and again prior to discharge from the rooming-in area. Newborns with positive screening results were evaluated by a cardiologist and underwent echocardiography.

Pulse oximeters use an infrared light beam directed at the skin to measure oxygen saturation. These battery-operated devices—from the brands PushMed, ChoiceMMed, and Hylogy—were calibrated by trained technicians and equipped with a neonatal sensor for measurement. Waveform stability was verified to ensure accurate readings of both oxygen saturation and heart rate, thereby confirming the plethysmographic waveform displayed on the monitor.

Statistical analysis

Screening was conducted according to the AAP algorithm for the early detection of CHDs. Pulse oximetry measurements were obtained from the right hand and either foot. An oxygen saturation ≤ 94 % or a > 4 % difference between hand and foot readings on repeated measurements at one-hour intervals was considered a positive screening result. Conversely, an oxygen saturation > 94 % or a < 3 % difference between hand and foot readings was classified as a negative result (Table 1) ⁽¹⁰⁾.

RESULTS

In this study, pulse oximetry was performed according to the AAP algorithm. A total of 626 newborns (12.78 %) had oxygen saturation values < 94 % within the first 24-48 hours prior to discharge, whereas 4,271 newborns (87.21 %) had values > 94 % (Table 2).

Pulse oximetry as a screening tool for congenital heart diseases

Echocardiography results for positive pulse oximetry screenings are presented in Table 3. Among the 626 positive screenings, 497 cases (79.40 %) had a positive echocardiogram, while the remaining 129 (20.60 %) showed negative findings.

Table 3. Echocardiogram results

Echocardiogram result	Number of cases	% of positive cases (n = 626)
Positive echocardiogram	497	79.40
Negative echocardiogram	129	20.60
Total	626	100.00

Source: HUG Statistics Department

Table 4 shows the distribution of echocardiographic diagnoses. Patent foramen ovale (54.70 %) and patent ductus arteriosus (25.50 %) were the most common findings.

Table 4. Echocardiographic diagnoses

Diagnosis	Number of cases	%
Patent ductus arteriosus	127 (*)	25.50
Ventricular septal defect	34	6.84
Atrial septal defect	25	5.00
Secondary pulmonary hypertension	23	4.60
Patent foramen ovale	272	54.70
Aortic aneurysm	4	0.80
Coarctation of the aorta	8	1.60
Heart rhythm disorders	4	0.80
Total	497	100.00

Source: HUG Statistics Department

Note (*): Newborns were discharged early, between 24 and 48 hours.

DISCUSSION

CHDs occur in 0.8-1 % of newborns and are frequently associated with comorbidities that can worsen the clinical course, such as low birth weight and extracardiac anomalies ⁽¹⁴⁾. Although some centers provide prenatal diagnosis, postnatal assessment remains essential for detecting these malformations ⁽¹⁵⁻¹⁷⁾.

Structural defects of the heart and great vessels represent the most common congenital malformations, with an incidence of 2.5-3 per 1,000 live births ⁽¹⁸⁾. This group includes conditions characterized by decreased pulmonary blood flow or right-sided heart obstruction, which allow admixture of oxygenated and deoxygenated blood, as well as conditions that prevent normal mixing between the systemic and pulmonary circulations ^(13,19). These features correspond to the typical presentation of right-to-left shunts caused by septal defects, in contrast to acyanotic CHDs, where the shunt is left-to-right as a result of left-sided obstructions ^(20,21). Notably, our screening detected structural heart defects in 12.78 % of newborns.

The most frequent CHD in our study was patent ductus arteriosus, identified in 127 patients (25.60 %), which is

consistent with the findings of Mendoza et al., who reported this malformation in 19.00 % of cases ⁽¹¹⁾. We also identified 25 cases (5.0 %) of atrial septal defect and 34 cases (6.84 %) of ventricular septal defect, results comparable to those reported by Herrera Morban et al. ⁽²²⁾. Some atrial and ventricular septal defect cases manifested after hospital discharge and were therefore not included in our dataset. Although asymptomatic CHDs may appear clinically insignificant at birth, they often have significant repercussions during the first years of life ^(22,23).

Patent foramen ovale was among the most frequently detected abnormalities by echocardiography (127 cases) and represented one of the most common findings in this study. Early diagnosis is crucial, as some defects, although not classified as critical, are clinically relevant because of their association with pulmonary hypertension ^(24,25). Although atrial and ventricular septal defects were not the most prevalent findings, their detection was nonetheless conclusive in our study.

With regard to the ChoiceMMed oximeter, a higher number of false positives was observed compared with other brands, such as Masimo SET, which has been reported to achieve a sensitivity of 100 % and a specificity of 93.90 %; however, both devices

demonstrated a negative predictive value of 100 % ^(26,27). In this study, the quality of saturation readings was assessed every 8-hour shift, alongside evaluation of the patient's clinical condition and existing risk factors. Pulse oximetry and screening together enabled the identification of critical cardiovascular diseases, including four cases of aortic aneurysm and eight cases of coarctation of the aorta. Nevertheless, sensitivity and specificity could not be specifically assessed, consistent with the conclusions of Riede et al. ^(12,28).

It is also noteworthy that early fetal echocardiography has a high rate of both false negatives and false positives because cardiac structures are not yet fully developed at that stage. Furthermore, its use is limited in routine prenatal care due to constraints in first-line equipment and trained personnel ^(29,30).

In conclusion, pulse oximetry screening for CHD is essential for the early diagnosis of newborns.

Acknowledgments: We thank the staff of HUG for providing the data used in this study.

Author contributions: TPM and JJV contributed to the conceptualization and research; RMV was responsible for data curation, formal analysis, and methodology; GVG and ACG drafted the manuscript; and GVG and EBC revised and edited the manuscript.

Funding sources: The article was funded by the authors.

Conflicts of interest: The authors declare no conflicts of interest.

BIBLIOGRAPHIC REFERENCES

1. Meller CH, Grinenco S, Aiello H, Córdoba A, Sáenz-Tejeira MM, Marantz P, et al. Congenital heart disease, prenatal diagnosis and management. *Arch Argent Pediatr* [Internet]. 2020;118(2):149-61.
2. Armas López M, Sierra RE, Rodríguez Collado Y, Elías Armas KS. Morbilidad y mortalidad neonatal por cardiopatías congénitas. *Rev Cubana Pediatr* [Internet]. 2019;91(1):661.
3. Ministerio de Salud Pública. Gacetas Epidemiológicas: Gaceta General 2020 [Internet]. Ecuador: Ministerio de Salud Pública; 2020. Available from: <https://www.salud.gob.ec/gacetas-epidemiologicas-gaceta-general-2020/>.
4. Ramírez-Escobar M, Betancurt-Serrano J, Ramírez-Cheyne J, Torres-Muñoz J, Madrid-Pinilla AJ. La pulsioximetría como herramienta para la tamización de cardiopatías congénitas críticas. Una revisión narrativa *Revista Colomb Cardiología* [Internet]. 2019;26(1):33-42.
5. Berlanga-Bolado O, Rivera-Vásquez P, Martínez Padrón HY. Sensitivity and specificity of pulse oximetry to detect congenital heart disease in newborns. *Horiz Sanitario* [Internet]. 2023;22(2):271-78.
6. Jiménez Carvajal MG, López Pérez D, Fernández Luna CP. Relevancia de la detección de cardiopatías congénitas complejas mediante cribado con oximetría de pulso en recién nacidos aparentemente sanos en los establecimientos de salud. *Arch Cardiol Méx* [Internet]. 2018;88(4):298-305.
7. Flórez-Muñoz Sandra L, Rubiano-Pedroza José A, Molina-Medina Clara N, Lozada-Muñoz Adriana, Rocha-Pacheco Lila M. Tamizaje con oximetría de pulso en el diagnóstico de cardiopatías congénitas críticas en recién nacidos. *Revista Colomb Cardiología* [Internet]. 2021(6):583-89.
8. Huang Y, Zhong S, Zhang X, Kong L, Wu W, Yue S, et al. Large scale application of pulse oximeter and auscultation in screening of neonatal congenital heart disease. *BMC Pediatr* [Internet]. 2022;22(483).
9. Suárez-Ayala D, Morcillo-Bastidas K, Vallejo-Mondragón EL, Valencia-Salazar A, Madrid-Pinilla A. Conocimiento y aplicación del tamizaje neonatal de cardiopatías congénitas críticas mediante el uso de oximetría de pulso. *Revista Colomb Cardiología* [Internet]. 2016;23(6):553-59.
10. Cullen PJ, Guzmán CB. Tamiz de Cardiopatías congénitas Críticas. Recomendaciones Actuales. *Acta Med Grupo Ángeles* [Internet]. 2014;12(1):24-9.
11. De Rubens Figueroa J, Mier Martínez M, Jiménez Carvajal MG, García Aguilar H. Tamizaje neonatal cardíaco en México, una herramienta para el diagnóstico temprano de cardiopatías críticas. *Gac Med Méx* [Internet]. 2022;158(2):67-71.
12. Riede F, Worner C, Dahnert I, Mockel 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* [Internet]. 2010;169(8):975-81.
13. 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* [Internet]. 2018;3:CD011912.
14. Cárdenas L, Enriquez G, Haecker S. Recién nacido portador de cardiopatía congénita compleja. Análisis de riesgo, toma de decisiones y nuevas posibilidades terapéuticas. *Rev Med Clin Las Condes* [Internet]. 2016;27(4):476-84.
15. Copado Mendoza DY, Martínez-Acevedo, S. Importancia del diagnóstico prenatal de las cardiopatías congénitas. *Perinatol Reprod Hum* [Internet]. 2018;32(3):127-30.
16. Janjua D, Singh J, Agrawal A. Pulse oximetry as a screening test for congenital heart disease in newborns. *J Mother Child* [Internet]. 2022;26(1):1-9.
17. Singh Y, Chen SE. Impact of pulse oximetry screening to detect congenital heart defects: 5 years' experience in a UK regional neonatal unit. *Eur J Pediatr* [Internet]. 2022;181(2):813-21.
18. Martínez I, Buendía F, Pijuán T, Abelleira C, Sánchez I, Bautista V. Selección de lo mejor del año 2018 en cardiopatías congénitas. *CardioClinics* [Internet]. 2018;54(1):23-9.
19. Peña R, Corona C, Medina M, Garrido L, Gutierrez C, Mier M. Presentación y manejo de las cardiopatías congénitas en el primer año de edad. *Arch Cardiol Méx* [Internet]. 2021;91(3):337-46.
20. Quiroz Conforme JF, Regalado Muñoz LS, Quiroz Conforme NV, Mendoza Gutiérrez AJ. Causas y consecuencias de cardiopatías congénitas en recién nacido. *RECIAMUC* [Internet]. 2021;5(2):131-39.
21. Giraldo-Grueso M, Zarante I, Mejía-Grueso A, Gracia G. Factors for congenital heart disease: a case control study. *Rev Colomb Cardiología* [Internet]. 2020;27(4):324-9.
22. Herrera Morban DA, Colomé-Hidalgo M, Méndez R, Torres Z, Jiménez S, Alcántara Tiburcio T, et al. Epidemiología de cardiopatías congénitas en un hospital de tercer nivel, Santo Domingo Norte, República Dominicana. *Cienc y Salud* [Internet]. 2020;4(2):37-44.
23. Barreto T. Pulse oximetry screening for critical congenital heart defects in newborns. *Am Fam Physician* [Internet]. 2019;99(7):421-2.
24. Mohammad N, Haikh S, Memon S, Das H. Spectrum of heart disease in children under 5 years of age at Liaquat University Hospital, Hyderabad, Pakistan. *Indian Heart J* [Internet]. 2014;66(1):145-9.
25. Méndez-Duran L, Echeverría-Consuegra R, Pérez O, Barbosa V, Contreras L, Cañón K. Prevalencia de cardiopatías congénitas diagnosticadas o tratadas por cateterismo cardíaco en pediatría. *Rev Colomb Cardiología* [Internet]. 2021;28(2):146-52.
26. Peña-Juarez R, Chávez-Saenz JA, García-Canales A, Medina-Andrade MA, Martínez-González MT, Gutiérrez-Cobián L, et al. Comparación de Oxímetros para detección de cardiopatías congénitas críticas. *Arch Cardio Mex* [Internet]. 2019;89(2):172-80.
27. Narayen IC, Te Pas AB, Blom NA, Van den Akker-van Marle ME. Cost-effectiveness analysis of pulse oximetry screening for critical congenital heart defects following homebirth and early discharge. *Eur J Pediatr* [Internet]. 2019;178(1):97-103.

Pulse oximetry as a screening tool for congenital heart diseases

28. 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 Cardiol* [Internet]. 2017;33(2):199-208.
29. Abdala D, Di Cicco V. Diagnóstico prenatal de cardiopatías congénitas. *Salud mil* [Internet]. 2020;39(1):14-9.
30. González-Andrade F, Echeverría D, López V, Arellano M. Is pulse oximetry helpful for the early detection of critical congenital heart disease at high altitude? *Congenit Heart Dis* [Internet]. 2018;13(6):911-18.