Pulse Oximetry-Based Critical Congenital Heart Disease Screening and Its Differential Performance in Rural America

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Critical congenital heart disease (CCHD) is defined as "any potentially life-threatening, ductal dependent cardiac lesions which requires surgical or cardiac catheterization-based intervention or results in death in the first 28 days of life."[1] Since 2018, CCHD screening is available for all births in the US.[2] Pulse oximetry is a highly specific and a moderately sensitive test for detection of CCHD with very low false-positive rates. CCHD screening with pulse oximetry is used to detect 12 CCHD conditions and 6 non-CCHD, secondary conditions.[3]

In the article entitled "Postnatal Diagnosis of Critical Congenital Heart Disease Is More Common in Rural Settings" by Marcus et al,[4] the authors assessed the relationship between rural geographical status and the postnatal diagnosis of congenital heart disease using oxygen saturation-based CCHD screening from retrospective data at two major cardiac surgery centers in the state of Washington over a 5-year period, with an additional focus on the mode of diagnosis for specific lesions. The authors report that both the incidence of undiagnosed CCHD and the postnatal diagnosis rate (5.5 per 10 000 live births and 48%, respectively) in rural areas was significantly higher compared with urban areas (2.1 per 10 000 live births and 32%). Pulse oximetry screening identified 7.5% of all CCHD and 22% of all postnatally diagnosed CCHD; however, the authors also noted that there were 15% false-negative screens.

The authors attribute the low rate of prenatal diagnosis and thereby a higher rate of postnatal CCHD diagnosis by pulse oximetry screening in rural areas to inadequate prenatal care, which can lead to missed diagnosis before birth. The challenge of access to care likely extends beyond traditional prenatal care. Because this is a cross-sectional study, there are naturally some limitations to the results. For example, there is no information on the mothers; we do not know whether they were able to access prenatal care near home and how many visits they completed. We also do not have information about the availability of more advanced imaging and testing in these areas. We know that there are approximately 3000 board-certified pediatric cardiologists in the US, which translates to 4.0 per 100 000 children aged 0-17 years, with significant geographic variation.[5] Although there are no data on the number of fetal cardiologists in the US, we would expect that number to be low overall and much lower in rural areas, because these physicians are concentrated mostly at or near large academic centers in major cities. According to the American Heart Association, 60 million people live in rural areas, yet only 9% of US physicians practice there.[6] Hence, the lack of access to a maternal-fetal medicine specialist or fetal cardiologist artificially inflates the overall postnatal diagnostic yield for patients living in a rural setting.

The article provides insight on the performance of the cost effective universal CCHD screening program in rural areas. This work can be crucial in understanding CCHD screening impact in resource-limited settings, where immediate specialist and high-acuity care is not available. In addition, this insight can be applicable to other similar settings, both within the US and worldwide, where there is a dire need to improve the quality and access to comprehensive prenatal and neonatal care.

Even with adequate access, there are limitations to prenatal screening. Both total anomalous pulmonary venous return and coarctation of the aorta (CoA) are known to have low prenatal detection rates owing to the limitations of fetal echocardiogram. In this study, total anomalous pulmonary venous return was the most common diagnosis after a failed CCHD screen, thereby stressing the importance of CCHD screening in diagnosing a potentially life-threatening cardiac lesion if missed prenatally. At the same time, it was noted that 73% of all false-negative screens were CoA, making it even more difficult to diagnose CoA, even after birth.

It is important to understand the impact of other confounding variables on these results which include race, ethnicity, and socioeconomic status, which were not addressed in this study. Also, no maternal data were coupled with the available data to study maternal factors, prenatal access to care, or maternal comorbidities and their effect on the difference in incidence of CCHD. These data are important because they have shown previously that maternal health, diabetes, socioeconomic factors, and access to ultrasound imaging impacts prenatal diagnosis of CCHD.[7]

There is still evidence to suggest that there is room for improvement in compliance with universal CCHD screening. Further, as suggested by the authors, many cases of CoA continue to be missed by both prenatal and postnatal CCHD pulse oximetry-based screening. CoA can be life

CCHD	Critical congenital heart disease
CoA	Coarctation of the aorta

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threatening in early infancy, but may not be associated with hypoxemia, stressing the importance of going back to the bedside and performing a thorough clinical examination including palpation of femoral pulses, and if suspicious, a 4-limb blood pressure measurement. It is important to remember that CCHD screen is only a supplement to physical examination and not a replacement in diagnosing congenital heart disease.

Additional diagnostic techniques, such as photoplethysmography, which adds data on perfusion and radiofemoral pulse delay, might add some value and help to increase the accuracy of diagnosis of CCHD, especially in those lesions where hypoxemia is not apparently seen in the first few days of life.[8] However, this strategy needs further large-scale testing. Other interventions can include expanded use of telehealth to support prenatal and neonatal care in the rural areas where millions of Americans live. Future studies will need to continue to evaluate the impact of healthcare access and technology on the diagnostic rates of CCHD and the care provided to these children living in rural areas. ■

CRediT authorship contribution statement

Krishna Kishore Umapathi: Writing – review & editing, Writing – original draft, Conceptualization. **John G. Frohna:** Writing – review & editing, Supervision, Conceptualization.

Declaration of Competing Interest

The authors declare no conflicts of interest.

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References

1. 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.
2. Glidewell J, Grosse SD, Riehle-Colarusso T, Pinto N, Hudson J, Daskalov R, et al. State actions in support of newborn screening for critical congenital heart disease — United States, 2011–2018. MMWR Morb Mortal Wkly Rep 2019;68:107-11.
3. 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. https://doi.org/10.1542/peds.2015-4573
4. Marcus BS, Perez-Kersey P, Lee AB, Jensen RA, Dullanty BS, Parrish PR, et al. Postnatal diagnosis of critical congenital heart disease is more common in rural settings. J Pediatr Clin Pract 2024. https://doi.org/10.1016/j.jpedcp.2024.200120
5. American Board of Pediatrics. Pediatric subspecialty state and county maps. Accessed August 26, 2024. https://www.abp.org/dashboards/pediatric-subspecialty-us-state-and-county-maps
6. Harrington RA, Califf RM, Balamurugan A, Brown N, Benjamin RM, Braund WE, et al. Call to action: rural health: a presidential advisory from the American Heart Association and American Stroke Association. Circulation 2020;141:e615-44. https://doi.org/10.1161/CIR.0000000000000753
7. Campbell MJ, Lorch S, Rychik J, Quartermain MD, Passarella M, Groeneveld PW. Socioeconomic barriers to prenatal diagnosis of critical congenital heart disease. Prenat Diagn 2021;41:341-6. https://doi.org/10.1002/pd.5864
8. Siefkes H, Oliveira LC, Koppel R, Hogan W, Garg M, Manalo E, et al. Machine learning-based critical congenital heart disease screening using dual-site pulse oximetry measurements. J Am Heart Assoc 2024;13:e033786. https://doi.org/10.1161/JAHA.123.033786