



Congenital Heart Disease: Expanding Newborn Screening Guidelines

OVERVIEW

Congenital heart disease (CHD) is the most common birth defect in the United States, affecting approximately 40,000 infants annually.¹ Among these, 25% will have critical congenital heart disease (CCHD), conditions that require timely medical intervention to prevent severe complications or death.² Early detection through newborn screening is crucial, as some infants may appear healthy at birth and be discharged before a CHD is identified. There are many (dozens) of CHD subtypes, however no cure currently exists for any of them.³ In September 2011, critical congenital heart disease (CCHD) screening via pulse oximetry was added to the U.S. Recommended Uniform Screening Panel (RUSP).⁴ RUSP is a list of disorders that the Secretary of the Department of Health and Human Services recommendeds for states to screen among newborns. Previously, the recommendations were developed by the national Advisory Committee on Heritable Disorders in Newborns and Children, but the group was terminated in April 2025 as part of federal reorganization. As of 2025, 49 states and Washington D.C have enacted legislation, regulations or hospital guidelines requiring CCHD newborn screening.⁵

AAP 2025 Updated Recommendations

Linkage of newborn screening programs with birth defects monitoring programs and vital records

The use of pulse oximetry has led to advancements in CHD diagnosis, however, pediatricians should continue to use additional testing as needed

If weaning off oxygen is not possible prior to discharge, proceed only with echocardiogram unless state law requires pulse oximetry

Infants who have undergone echocardiogram prior to CHD screening may forgo formal CCHD screening

Use a simplified CCHD screening algorithm that eliminates a second rescreen if a newborn falls into the retest category, allowing for earlier intervention

Public health programs should look for ways to improve data collection, data sharing and improved access to care

Source: Oster, M. et al. (2024). Newborn Screening for Critical Congenital Heart Disease: A New Algorithm and Other Updated Recommendations: Clinical Report. *Pediatrics* 155(1): e2024069667.

California is the only outlier by mandating that screening be offered but not required.⁵ Along with the American Heart Association (AHA), the American Academy of Pediatrics (AAP) also endorsed the addition of CHD to RUSP, and has since provided updated recommendations for CHD screening.⁴

Mandated newborn screening using pulse oximetry has led to significant improvements in infant health outcomes.⁴ Pulse oximetry measures the level of oxygen in a newborn's blood and is a non-invasive, cost-effective screening costing roughly just \$15-22.⁶ Studies have shown that such screening reduces early infant deaths from CCHD by approximately 33% potentially saving 120 lives annually in the United States.⁷ Early detection enables healthcare providers to plan and administer appropriate treatments, such as surgery, in a timely manner, improving survival rates and long-term health for affected infants.

LIMITATIONS OF CURRENT GUIDELINES

While pulse oximetry is reliable and cost-effective, false positives and asymptomatic cases can occur, necessitating additional tests, in some cases, such as echocardiograms and electrocardiograms.⁴ A systematic review found that more than 50% of newborns with CHD are not diagnosed at birth.⁸ This is not due to lack of screening but rather false negatives during or delayed development of heart conditions. The same review found that echocardiograms are best used in cases of newborns with asymptomatic non-syndromic cardiac murmurs (ANCM).⁸ These tests are vital for confirming diagnoses in healthy newborns with cardiac murmurs and planning appropriate interventions. As a result, some states have introduced legislation to enhance CHD screening protocols. These bills expand the state's newborn screening requirements to include echocardiograms and electrocardiograms for infants at higher risk of CHD. The legislation mandates that newborns with a family history of CHD or those whose biological parents have specific cardiac health histories undergo these additional screenings. If a birthing facility lacks the necessary equipment or staff, it's required to refer the infant for the tests upon discharge.

ADVANTAGES TO ADDITIONAL SCREENING

- Physical exam detection of central cyanosis, one of the main markers for CHD, may be less reliable in newborns with darker skin tones.⁹
- Pulse oximetry tests are reported to overestimate oxygen levels up to 2.6% in newborns with darker skin, affecting diagnosis for non-white or African American newborns.⁹
- Newborns with darker skin may not be accurately diagnosed with CHD. Updating screening to include echocardiograms and electrocardiograms acts as a safety net for newborns that might not be correctly diagnosed using current screening guidelines.

DISADVANTAGES TO ADDITIONAL SCREENING

- Echocardiograms are more costly to patients than current newborn screening tests, ranging from ~\$500 to >\$3,000, depending on insurance status.¹⁰
- Electrocardiograms cost between \$30 to \$100 in copays for patients with insurance, while patients lacking insurance pay an average of \$1,500 with some hospitals charging up to \$3,000.¹¹
- Lack of access to cardio-diagnostic technologies such as electrocardiograms and echocardiograms is prevalent across the United States, as only 53% of hospitals offer hospital-based general cardiology services.¹² If states adopt legislation to expand CHD newborn screening to include electrocardiograms and echocardiograms, hospitals lacking cardiology services will have to provide referrals to specialists, further delaying official diagnosis.
- Additional mandated testing would place additional strain not only on pediatric cardiologists, but the specialized services required to perform electrocardiograms and echocardiograms therefore increasing wait-times for testing.
- Not all medical staff are trained to read and interpret newborn electrocardiograms and echocardiograms, requiring more specialists to keep up with an increase in testing demand.¹³
- Not all congenital heart conditions are detectable in the first 24 hours of life. Some lesions evolve or become symptomatic later—so a hospital-conducted echocardiogram/EKG before discharge might still require repeat testing later if clinically indicated, thereby limiting the value of a “single-screening” mandate.¹⁴
- The increase in cost for newborn screening, along with possible travel to a specialist, will further health disparities as legislation will mandate these services to be performed, leaving rural and low-income families at higher risk for an inability to access the services and financial insecurity due to increased medical bills, travel, and time away from work.

AHA POLICY GUIDANCE

Given the many concerns with mandating routine echocardiograms or electrocardiograms for newborn screening of congenital heart disease, the American Heart Association will continue to advocate for (1) evidence-based improvements to existing newborn screening protocols and (2) greater investment in prenatal detection and pediatric care infrastructure.

- **Preserve and strengthen pulse oximetry as the standard of care.** Pulse oximetry remains a cost-effective, evidence-based screening method for detecting critical CHD in newborns. Universal, mandated use of pulse oximetry should continue to serve as the foundation of newborn CHD screening. Improvements should focus on standardizing testing protocols and enhancing follow-up pathways for abnormal results.
- **Increase access to affordable, quality health care.** Support policies that expand Medicaid and Children’s Health Insurance Program (CHIP) coverage to increase access and reduce barriers to healthcare coverage to ensure families have access to diagnosis and treatment in the prenatal and pediatric period. This includes extending postpartum Medicaid coverage from 60 days to 12 months of continuous coverage, ensuring ongoing access to maternal and infant health services.
- **Improve the chain of survival.** Support implementation of Cardiac Emergency Response Plans (CERPs) in schools and communities, as well as pre-participation cardiovascular screening for student athletes, to reduce preventable sudden cardiac events in youth.
- **Support workplace accommodations for families.** Advocate for paid family and medical leave policies that allow parents and caregivers to care for infants or children with CHD without financial hardship. Ensuring adequate caregiving leave and workplace flexibility supports family well-being and continuity of care.

¹CDC. (2024). Congenital Heart Defects (CHDs): Data and Statistics. <https://www.cdc.gov/heart-defects/data/index.html>

²CDC. (2024). Congenital Heart Defects (CHDs): Screening for Critical Congenital Heart Defects. <https://www.cdc.gov/heart-defects/screening/index.html>

³Children’s Heart Foundation. (2025). CHD Facts and Statistics. <https://chfn.org/who-we-are/chd-facts-and-statistics.html>

⁴Oster, M., et al. (2024). Newborn Screening for Critical Congenital Heart Disease: A New Algorithm and Other Updated Recommendations: Clinical Report. *Pediatrics* 155(1).

⁵Jenco, M. (2019). Study: 49 states require newborn heart screening. *American Academy of Pediatrics*.

⁶Grosse 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. *International Journal of Neonatal Screening*. 2017; 3(4):34. <https://doi.org/10.3390/ijns3040034>

⁷CDC. (2025). Congenital Heart Defects (CHDs): Clinical Screening and Diagnosis for Critical Congenital Heart Defects. <https://www.cdc.gov/heart-defects/hcp/screening/index.html>

⁸Yoon, S. et al. (2020). Congenital heart disease diagnosed with echocardiogram in newborns with asymptomatic cardiac murmurs: a systematic review. *BMC Pediatrics* 20(1) 322.

⁹Sakai-Bizmark, R. et al. (2021). Effect of newborn screening for critical congenital heart disease on healthcare utilization. *Cardiology in the young* 30(8) 1157-1164.

¹⁰Reisman, S. (2025). Echocardiogram. *Spend on Health*. <https://spendonhealth.com/echocardiogram-cost/>

¹¹CostHelperHealth. (2024). EKG Cost. <https://health.costhelper.com/ecg.html>

¹²Naele, T. (2025). Access to Key Cardiac Services Remains Uneven Across the US. Cardiovascular Research Foundation. <https://www.tctmd.com/>

¹³Chen Y, Kunst E, Nasrawi D, et al. Nurses’ competency in electrocardiogram interpretation in acute care settings: A systematic review. *J Adv Nurs*. 2022;78(5):1245-1266. doi:10.1111/jan.15147

¹⁴William T. Mahle, Gerard R. Martin, Robert H. Beekman, W. Robert Morrow, Geoffrey L. Rosenthal, Christopher S. Snyder, L. LuAnn Minich, Seema Mital, Jeffrey A. Towbin, James S. Tweddell; Endorsement of Health and Human Services Recommendation for Pulse Oximetry Screening for Critical Congenital Heart Disease. *Pediatrics* January 2012; 129 (1): 190–192. 10.1542/peds.2011-3211