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EFFECTIVENESS OF ECHOCARDIOGRAPHIC SCREENING IN DETECTING COMPLEX CONGENITAL HEART DEFECTS IN CHILDREN: AN OVERVIEW

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Аннотация

Врожденные пороки сердца (ВПС) остаются одной из ведущих причин младенческой заболеваемости и смертности. Своевременная диагностика сложных ВПС необходима для снижения смертности и улучшения результатов лечения. Эхокардиографический скрининг считается «золотым стандартом» благодаря высокой чувствительности и специфичности. В обзоре проанализированы современные данные (2020–2024) об эффективности эхокардиографического скрининга при выявлении сложных ВПС у новорождённых и детей. По сравнению с физикальным обследованием и пульсоксиметрией эхокардиография демонстрирует более высокую диагностическую точность, достигая чувствительности 95–100%, особенно при цианотических пороках и аномалиях, зависящих от открытого артериального протока. Однако к ограничениям относятся высокая стоимость, зависимость от квалификации оператора и риск гипердиагностики незначимых дефектов. Интеграция таргетированного эхокардиографического скрининга в неонатальную практику в сочетании с пульсоксиметрией и клинической оценкой обеспечивает оптимальную раннюю диагностику и улучшение исходов лечения.

Ключевые слова: *эхокардиографический скрининг, врожденные пороки сердца, ранняя диагностика, диагностическая точность, неонатальная кардиология*

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Abstract

Congenital heart defects (CHDs) remain one of the leading causes of infant morbidity and mortality. Timely detection of complex CHDs is essential to reduce mortality and improve outcomes. Echocardiographic screening is considered the gold standard due to its high sensitivity and specificity. This review analyzes the current evidence (2020–2024) on the effectiveness of echocardiographic screening in detecting complex CHDs among neonates and children. Compared to physical examination and pulse oximetry, echocardiography demonstrates superior diagnostic accuracy, with sensitivity reaching 95–100%, especially for cyanotic and duct-dependent lesions. However, limitations include high costs, operator dependency, and potential overdiagnosis of minor anomalies. The integration of targeted echocardiographic screening into neonatal care, combined with pulse oximetry and clinical evaluation, ensures optimal early detection and better patient outcomes.

Keywords: *echocardiographic screening, congenital heart defects, early detection, diagnostic accuracy, neonatal cardiology*

Introduction

Complex congenital heart defects (CHDs) – often termed “critical” CHDs – are severe cardiac malformations present at birth that typically require surgery or catheter intervention in early life[1][2]. These defects (e.g. hypoplastic left heart syndrome, transposition of the great arteries, tetralogy of Fallot, truncus arteriosus) can lead to acute cardiovascular collapse or death if not recognized promptly[1]. Early detection is therefore crucial: delayed diagnosis of critical CHD is associated with higher risk of infant mortality and worse outcomes after intervention[1]. Traditionally, CHD screening has relied on prenatal ultrasound and postnatal clinical examination, but these have relatively low detection rates (on the order of 25–50%)[3]. In the past decade, routine **newborn pulse oximetry** screening has been implemented in many countries to improve early identification of critical CHDs. Pulse oximetry is a simple bedside test to detect hypoxemia (low oxygen saturation) in apparently healthy newborns, which is a common feature of many critical CHDs[4][5]. However, some serious defects do not cause early hypoxemia and may evade pulse ox screening[6][7]. **Echocardiography** – an ultrasound of the heart – is the diagnostic gold standard for CHD and can visualize structural anomalies directly[8]. This raises the question of using echocardiography as a *screening* tool to detect complex CHDs early. Below, we examine the effectiveness of echocardiographic screening in children, including its accuracy (sensitivity, specificity, predictive value), impact on early diagnosis and outcomes, the types of CHD it detects best, comparisons with other screening methods, and criteria for its use at different pediatric ages.

Materials and Methods

This review analyzed data from PubMed, Scopus, and Cochrane Library databases (2012–2024). Priority was given to systematic reviews, meta-analyses, and large-scale cohort studies comparing echocardiographic screening with physical examination and pulse oximetry in neonates and infants. Keywords included: “echocardiographic screening,” “congenital heart defects,” “neonatal diagnosis,” and

"early detection".

Results

Echocardiography shows 90–100% sensitivity and ~99% specificity for detecting CHDs, making it the gold standard for confirmation after abnormal screening [9][10][11][12]. However, universal use yields ~5% false positives versus ~1.3% for pulse oximetry, with a PPV of only 1–2% for critical CHDs [13][14][15][16].

Modeling studies report detection of 68–85% of critical CHDs via universal echocardiography, slightly higher than pulse oximetry (~68%) [17][18]. Some lesions, like coarctation of the aorta, may still be missed in early life [7].

Thus, echocardiography is highly accurate but better suited for targeted screening rather than universal application due to cost, resource demands, and risk of overdiagnosis.

Table 1: Comparision of the performance of echocardiography with other screening methods for critical CHD:

Screening Method	**Sensitivity for Critical CHD**	**Specificity**	**False-Positive Rate**
Newborn physical exam	~30–50%[17][3]	~99.5%[15]	~0.05% (≈1 in 2,000)[16]
Newborn pulse oximetry	~75%[18][19]	~99.9%[18]	~0.1% (≈1 in 1,000)[18]
Newborn echo screening (all infants)	~69%[17] (practical)*	~94–95%[15]	~5% (≈1 in 20)[15]

**In ideal conditions with expert cardiologists, echocardiography can approach*

nearly 100% sensitivity for structural CHD[20]. The ~69% figure reflects actual detection rates of critical CHD in a population screening model, recognizing that some lesions (e.g. coarctation) may not be apparent on the first days' echo or could be missed due to operator limitations[17][7].

Systematic CHD screening significantly improves **early detection** and outcomes in infants with critical CHDs. Mandatory newborn screening in the US reduced early CHD-related deaths by **~33%**, saving **~120 lives annually** [23][24][25][26], and lowered emergency hospitalizations for undiagnosed cases [27].

Early detection enables timely surgery and reduces preoperative complications, neurologic injury, and mortality, whereas delayed diagnosis increases risk of shock and organ damage [1][28]. Adding **echocardiography** to clinical exams nearly **doubles detection rates** before discharge (69% vs. 32%) [17], though rapid referral remains essential [29].

Pulse oximetry effectively detects cyanotic lesions — d-TGA, TAPVR, truncus arteriosus, pulmonary atresia, HLHS — but misses many non-cyanotic defects such as coarctation, with sensitivity as low as **~21%** [7][30]. **Clinical examination** identifies murmurs in **93.6%** of CHD cases by six weeks [31] but still misses half of critical CHDs [3][32].

Echocardiography detects nearly all structural anomalies, including asymptomatic and non-cyanotic lesions [17], but universal screening risks overdiagnosis of minor defects [33][34][14]. Combining **pulse oximetry + physical exam + targeted echocardiography** yields the highest diagnostic accuracy and optimizes early intervention [6][30].

Table 2: Summ of effectiveness various screening methods detect certain major CHD types:

CHD Lesion	**Detected by Pulse Ox?**	**Detected by Physical Exam?**	**Detected by Echo?**
Transposition of	Yes (usually)[30]	Often *no murmur*;	Yes – anatomy

great arteries	– causes hypoxemia	cyanosis may be missed visually	clearly abnormal on echo
Tetralogy of Fallot	Yes – cyanotic spells or low O_2[30]	Possible harsh murmur (VSD/PS); cyanosis if severe	Yes – VSD and RV outflow obstruction seen
Total anomalous pulm. venous return	Yes – severe hypoxemia[35]	Cyanosis (if noted); no specific murmur	Yes – anomalous connections visible
Hypoplastic left heart syndrome	Yes – ductal closure causes low O_2[35]	Maybe gray color, weak pulses; no murmur	Yes – small left heart structures seen
Coarctation of the aorta	**Often NO** (O_2 can be normal)[6][7]	Possibly differential pulses or mild late murmur	Yes – arch narrowing detectable if carefully evaluated
Large VSD (isolated)	No (fully oxygenated blood)	**Yes – loud murmur** after a few days of life	Yes – defect visualized, shunt flow on Doppler
Truncus arteriosus	Yes – causes cyanosis	Possible single S2, systolic murmur; cyanosis	Yes – common arterial trunk easily seen
Tricuspid atresia	Yes – cyanosis	Maybe murmur of VSD; cyanosis evident if severe	Yes – atretic tricuspid and ASD/VSD visualized
Ebstein's anomaly	Often yes (if severe cyanosis)	Murmur of tricuspid	Yes – malformed tricuspid and

		regurgitation possible	atrialized RV seen
Critical pulmonary stenosis/atresia	Yes – cyanosis (ductal dependent)	Murmur if PS; cyanosis if duct closes	Yes – dysplastic valve or atresia visible
Atrioventricular canal (AVSD)	No (if balanced circulation)	Often **yes – loud murmur** by a few weeks old	Yes – septal defects and common AV valve seen

Detection of select CHDs by different screening methods. Pulse oximetry excels at cyanotic lesions; physical exam can catch those with murmurs or abnormal pulses; echocardiography can identify essentially all structural defects if performed early.

Echocardiography detects nearly all CHDs, including lesions missed by other methods such as coarctation [6][7], but universal screening is limited by cost, resource demands, and risk of overdiagnosis of minor, clinically insignificant defects [14].

Pulse oximetry efficiently identifies critical cyanotic CHDs with high specificity and minimal false positives but misses many acyanotic lesions [6][7]. Physical exam contributes to detecting murmurs but has low standalone sensitivity.

The highest detection rates are achieved through a tiered strategy: universal pulse oximetry + clinical exam, reserving echocardiography for confirmatory diagnostics and high-risk infants. This approach maximizes accuracy while minimizing unnecessary interventions [6][7][14].

Echocardiography provides the highest theoretical detection rate for congenital heart defects, identifying nearly all structural anomalies, including non-cyanotic lesions [17][38]. **Pulse oximetry** detects ~70–76% of critical CHDs [18], while physical exam alone captures only ~30–50% [36]. Combining exam + pulse oximetry improves sensitivity to **80–90%** [37][32]. However, routine universal echo screening

offers minimal incremental benefit for critical CHDs (69% vs. 68% with pulse ox alone) but detects more non-critical lesions [17][38].

Pulse oximetry has excellent specificity (~99.9%), with false positives in only **0.1–0.2%** of newborns [18]. Echocardiography, if used universally, generates **~5%** false positives [15], leading to unnecessary follow-ups, while physical exams fall in between (~0.5%) [15]. The positive predictive value (PPV) for failed pulse ox ranges **20–50%** [39], versus only **2–3%** for abnormal echo screens [16].

Pulse oximetry is inexpensive ($14 per infant) [40][41] and cost-effective ($12,000 per life-year saved) [41]. Universal echo screening is far costlier (~£3.5M/100,000 infants vs. £480K for pulse ox) [42], with high marginal costs per additional diagnosis [38]. Moreover, echo requires specialized pediatric cardiology expertise, limiting scalability [43][44][45], whereas pulse oximetry is quick, nurse-performed, and universally implemented [46].

Echocardiography is targeted for:

- Newborns with failed pulse ox or abnormal physical exams [6][7][50][11]
- Infants with **high-risk syndromes** (e.g., Down, Turner, DiGeorge) [51][52] [53]
- NICU patients, persistent hypoxemia, or complex anomalies [54][55]
- Older infants or children with new murmurs, cyanosis, poor growth, or suspected CHD [56][57][61]

Discussion

Echocardiography remains the **gold-standard** diagnostic tool but is not suited for universal newborn screening due to cost, feasibility, and false positives [15][38] [15][38]. **Pulse oximetry + clinical exam** offers the optimal balance: high specificity, moderate sensitivity, low cost, and practical scalability [18][7] [18][7]. Echo serves as a **targeted follow-up** and is routinely applied in high-risk groups, maximizing detection while minimizing unnecessary interventions [48][52][54] [48][52][54].

This review evaluates echocardiographic screening for complex congenital heart defects (CHDs) within contemporary newborn and pediatric pathways.

Echocardiography remains the diagnostic gold standard, capable of visualizing virtually all structural anomalies and crucial as an immediate follow-up after any abnormal newborn screen or suspicious examination. As a **universal** screening tool, however, echo yields a higher false-positive rate and significant resource demands, making it less suitable for population-wide deployment. By contrast, **pulse oximetry plus clinical examination** provides the best balance of feasibility, specificity, and public-health impact, with substantial reductions in early infant CHD mortality where implemented. The most effective and scalable strategy is a **tiered approach**: universal pulse oximetry and careful physical examination for all newborns, **rapid referral** pathways, and **targeted echocardiography** for screen-positive infants, high-risk syndromes, NICU graduates, or later clinical suspicion. Program performance hinges on timely access to pediatric cardiology expertise, standardized algorithms, and robust systems for confirmatory testing and transfer. Future gains will likely come from tele-echocardiography networks, pediatric-focused training, handheld/point-of-care devices, and AI-assisted acquisition/interpretation, particularly for lesions under-detected by oximetry (e.g., left-sided obstructive lesions).

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