Review paper on Enhancing Pediatric Cardiology in Infants with Artificial Intelligence: Early Detection of Congenital Heart Disease

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Abstract:

Congenital heart disease (CHD) is a part of the common worldwide and primary causes of infant morbidity and mortalityif not detected at an early stage. Traditional diagnostic approaches such as Pulse oximetry and postnatal screening are heavily rely on the expertise of cardiac, which may result in delayed or missed diagnoses due to limited healthcare resources. Congenital heart diseases a group of heart disorder that are known to be some of the most critical cardiac conditions present at birth time. Common Congenital heart disease (CHD) defects include Atrial Septal Defect (ASD), Ventricular Septal Defect (VSD), Patent Ductus Arteriosus (PDA), Tetralogy of Fallot (TOF), and Transposition of Great Arteries (TGA). With the help of Artificial Intelligence (AI) incudes Machine Learning and Deep Learning techniques involve automated congenital heart disease (CHD) detection in infants by analysis of echocardiographic Images, Electrocardiogram (ECG) signals, and pulse oximetry data. Artificial Intelligence approaches can help in improving the accuracy and efficiency of diagnostic performance. AI based systems can used to achieve high levels of sensitivity and specificity in diagnosing congenital heart diseases. AI can help optimize task of healthcare in critical aspects of patient care. AI detecting congenital heart disease, there are still some difficulties remain, such as the need for large amounts of high-quality data and the requirement for careful validation of the algorithms and analysis.

Introduction:

Congenital heart disease (CHD) is the most common birth time abnormality, affecting about one in every hundred babies born worldwide. Globally, congenital heart disease (CHD) seems to be a major cause of infant's morbidity and mortality. Everyone with congenital heart disease (CHD) needs ongoing medical care for their whole life. Because these heart diseases can be so serious, a quick diagnosis and intervention are essential to ensure a new-born's well-being. After birth and the transition from fetal to new born circulation, CHDs symptoms are marked by the closure of the fetal shunt, making the existence of this condition evident [2, 3] The incidence of CHDs in Asia is higher than the global average, reaching a ratio of 9.3 cases per 1000 live births [1]. However, in>50% of new born, CHDs are clinically undetectable during hospital discharge [4]. This condition accounts for 5% of all childhood deaths and 18% of deaths within the first year of life among live born infants [1, 2, 4].Traditional screening methods, including pulse oximetry and clinical assessment, though useful, often fall short especially in healthcare environments with limited resources. Artificial intelligence(AI) is rapidly changing how we detect and diagnose congenital heart disease (CHD) in infants. By using methods that range from machine learning to advanced deep learning models, AI can

analyse medical images, ECGs, and other patient data with greater accuracy and efficiency. This offers a significant improvement over traditional methods and makes early diagnosis more accessible.

Literature Review:

As per all references, here review some previous work with their functionality, merits and demerits are as follow.

Henning Clausen, Elisabeth Norén, SallaValtonen, et al. [9] The study evaluated levels of circulating cardiovascular biomarkers (including NT-proBNP and hs-cTnT) in Swedish new born to detect CHD at birth. Blood samples were analysed within 48 hours of delivery and compared between CHD-affected and healthy infants.

This review covers analysed the clinical outcomes of neonates diagnosed with critical CHD using a registry-based approach in South India. Data was collected prospectively on survival, intervention timing, and access to care. The study emphasized the challenges of late detection and limited access to specialized cardiac services[10].

Jan Pavlicek, Eva Klaskova, et al. [11] Evaluated the timing, diagnostic methods, and outcomes of major congenital heart defects (CHDs) diagnosed during the first year of life in infants across Czech regions. The study involved reviewing echocardiography findings, clinical symptoms, and delays in diagnosis.

The study proposed a structured strategy involving early postnatal screening using clinical examination, pulse oximetry, and confirmatory echocardiography for CHD detection. The authors emphasized timely evaluation within 48 hours of birth and integration of multidisciplinary care[12].

Jintai Chen, Shuai Huang, Ying Zhang, et al[13] Developed a deep learning model using pediatric ECG data to detect CHD, enhanced with domain knowledge (clinical concept embeddings). Improved interpretability and classification performance.Showed promising results using ECG-based AI models; highlighted integration of human expertise for model transparency and effectiveness.

The paper reviews the current applications of artificial intelligence in congenital heart disease, including image analysis, risk prediction, natural language processing (NLP), and

clinical decision support. It emphasizes the growing role of AI in precision medicine and highlights challenges in data availability and model generalization [14].

Methodology:

Imagine a world where it's easier and faster to find heart problems in infants right after they're born. AI can help doctors detect congenital heart disease (CHD) in infants.

Right now, finding these heart defects can be tricky. Doctors might use an echocardiogram, a special ultrasound for the heart, proximately Test, but it takes a lot of specialty, time, expert views to get a clear picture. In many places, there aren't enough specialists to do all the screenings, which means some babies' conditions aren't found until later when it's much harder to treat. AI offers a powerful solution by making these detections more reliable and accessible.

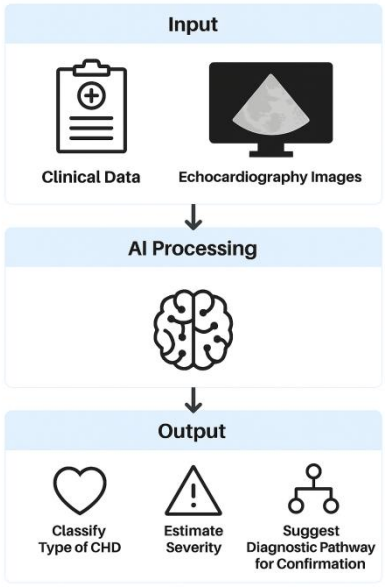

Figure 1 Early CHD Detection Process

CHD Diagnostics Method

Method	Utility	Limitations
Echocardiography	Gold standard for structural heart assessment. Enables prenatal and postnatal diagnosis.	Operator-dependent; limited access in rural/low-resource settings.
Pulse Oximetry	Quick, non-invasive screening for critical CHD. Widely used in newborn care.	May miss non-cyanotic CHDs; not diagnostic alone.
Auscultation	Accessible and simple. Can detect murmurs linked to CHD.	Low sensitivity; subtle murmurs often missed.

Electrocardiography (ECG)	Detects rhythm and electrical anomalies. Can suggest underlying defects.	Not definitive for structural CHD; used as a supplementary tool.
Additional Imaging (CT/MRI)	Provides detailed anatomical views in complex cases.	Requires sedation (MRI); radiation risk (CT); not routine for neonates.

How AI Is Helping Infants

AI isn't a replacement for doctors; it's a helpful assistant. AI can analyse various types of data to provide a comprehensive understanding of a baby's heart.

Result and Analysis

AI-based models, particularly those using deep learning and advanced machine learning, consistently demonstrate high sensitivity (up to 93%) and specificity (up to 96%) for detecting CHD in prenatal and neonatal populations, often surpassing traditional ultrasound and echocardiography in accuracy. These systems can analyse complex imaging data (ultrasound, echocardiography, CT, ECG) and clinical records, enabling early and precise identification of a wide range of CHD types, including critical and subtle defects. AI-assisted diagnosis also reduces the workload for healthcare professionals and can help bridge expertise gaps, especially in low-resource or remote settings.

Key Diagnostic Metrics for AI in CHD Screening

AI Application Area	Sensitivity	Specificity	Accuracy	Citations
Prenatal ultrasound/Echocardiography	0.89–0.96	0.91–0.96	93–96%	7,8,5
CT-based diagnosis	82.9%	—	86–97%	7
ECG/heart sound analysis	85–97%	89–98%	94–97%	5

Figure 4 Summary of AI diagnostic performance for early CHD detection.

Conclusion

AI applications show strong promise for smarter, earlier screening of infant CHD, with high diagnostic accuracy and potential to reduce disparities in care. Using AI analysis of medical data like ultrasounds and ECGs can detect congenital heart disease in infants earlier and more accurately than human eyes alone. This leads to timely interventions, better outcomes, and improved long-term health for affected babies. AI can help make heart disease

screening for babies more fair and accessible. It can be used in clinics and hospitals in rural areas, so all babies can get the care they need, no matter where they live. To make AI work well in healthcare, we need to keep improving it. This means making sure it's accurate, reliable, and fair for everyone.This way, doctors can use it to help babies get the best care possible, and make sure every baby has a healthy start.

Reference

1 Singh Y, McGeoch L. **Fetal anomaly screening for detection of congenital heart defects. J Neonatal** Biol. 2016;05(02):1–5. https://doi.org/10.4172/2167-0897.100e115.

2 Yoon SA, Hong WH, Cho HJ. **Congenital heart disease diagnosed with echocardiogram in newborns with asymptomatic cardiac murmurs: a systematic review**. BMC Pediatr. 2020;20(1):322. https://doi.org/10.1186/s12887-020-02212-8.

3 Puri K, Allen HD, Qureshi AM. **Congenital heart disease.**Pediatr Rev. 2017;38(10):471–86. https://doi.org/10.1542/pir.2017-0032.

4 Bordbar A**. Ascertaining the Prevalence of Heart Malformations in Neonates : A Novel Clinically Approved Solution**. pp. 1–9, 2021.

5 HarisEjaz, TarannumThyyib, Ahmed Ibrahim, AroobNishat, Jhancy Malay, Role of artificial intelligence in early detection of congenital heart diseases in neonates, Jan – 2024 - Frontiers in Digital Health

6 L. D. Liastuti, YosiliaNursakina, Diagnostic accuracy of artificial intelligence models in detecting congenital heart disease in the second-trimester fetus through prenatal cardiac screening: a systematic review and meta-analysis – Feb,2025

7 Xiaowei Xu, QianjunJia, Haiyun Yuan et. al, A clinically applicable AI system for diagnosis of congenital heart diseases based on computed tomography images – Sep,2023

8 KhadizaTunSuha, Hugh Lubenow et.al, The Artificial Intelligence-Enhanced Echocardiographic Detection of Congenital Heart Defects in the Fetus: A Mini-Review – Mar,2025

9 Henning Clausen, Elisabeth Norén, SallaValtonen, "**Evaluation of Circulating Cardiovascular Biomarker Levels for Early Detection of Congenital Heart Disease in Newborns in Sweden**" , Dec 2022 - JAMA Netw Open

10 Deepa Sasikumar, Mukund A Prabhu, RenuKurup, et al. "**Outcomes of neonatal critical congenital heart disease: results of a prospective registry-based study from South India**", 2022, Indian Pediatrics

11 Jan Pavlicek, Eva Klaskova, Sabina Kapralova, AlzbetaMoravovaPalatova, AlicjaPiegzova, Richard Spacek, Tomas Gruszka, "**Major heart defects: the diagnostic evaluations of first-year-olds**" Nov 2021, BMC

12 MedihaKardasevic, Ida Jovanovic, JelicaPredojevicSamardzic, "**Modern Strategy for Identification of Congenital Heart Defects in the Neonatal Period**", Oct 2016, Professional Paper

13 Jintai Chen, Shuai Huang, Ying Zhang, Qing Chang, Yixiao Zhang, Dantong Li, JiaQiu, Lianting Hu, Xiaoting Peng, Yunmei Du, Yunfei Gao, Danny Z. Chen, AbdelouahabBellou, Jian Wu &Huiying Liang, "**Congenital heart disease detection by pediatric electrocardiogram based deep learning integrated with human concepts**", Feb 2024, Nature Communication

14 Pei-Ni Jone a, Addison Gearhart b, Howard Lei c, Fuyong Xing d, Jai Nahar e, Francisco Lopez-Jimenez f, Gerhard-Paul Diller g,h,i, Ariane Marelli j, Laura Wilson k, ArwaSaidi k, David Cho l, Anthony C Chang, "**Artificial Intelligence in Congenital Heart Disease: Current State and Prospects**", Dec 2022, JACC Journal