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Enhancing Pediatric Congenital Heart Disease Outcomes: The Role of Machine Learning Models and AI-Driven Methodologies

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ABSTRACT

Congenital heart disease (CHD) presents a complex etiology involving multifaceted genetic and environmental interactions. The global prevalence of CHD approximates 8 per 1,000 live births, with elevated rates observed during prenatal periods, attributed to spontaneous pregnancy loss and elective terminations. Timely and precise diagnosis remains fundamental for optimal clinical outcomes, necessitating collaborative efforts among genetic counselors, obstetric practitioners, and pediatric cardiovascular specialists. While conventional diagnostic approaches such as electrocardiography and echocardiography continue to serve as cornerstone tools, sophisticated imaging techniques including cardiac computed tomography and magnetic resonance imaging are increasingly incorporated into clinical practice. Nevertheless, diagnostic challenges persist due to limited clinical recognition, inadequate healthcare infrastructure, and scarcity of specialized practitioners, potentially compromising diagnostic timeliness. Within this framework, artificial intelligence (AI)—specifically machine learning and deep learning technologies—has emerged as a transformative approach in pediatric cardiovascular medicine. AI systems demonstrate capability in identifying complex patterns within extensive datasets, thereby enhancing diagnostic precision, facilitating risk assessment, and enabling personalized therapeutic interventions. Contemporary AI implementations have demonstrated potential in optimizing cardiac imaging interpretation, supporting clinical decision-making processes, and forecasting patient outcomes. Despite promising developments, AI integration within pediatric CHD management remains constrained. Single-institutional studies and the relative rarity of CHD limit data accessibility, emphasizing the necessity for multi-center collaborative research initiatives. Additionally, AI-based systems can enhance postoperative surveillance, simulate therapeutic approaches, and identify complications through wearable monitoring technologies. Such innovations prove particularly valuable in resource-constrained environments where pediatric cardiovascular expertise remains limited. This comprehensive review examines the current state, existing challenges, and future prospects of AI implementation in pediatric cardiovascular medicine. Leveraging AI's comprehensive potential may revolutionize care delivery pathways, enhance prognostic outcomes, and optimize health management for children with CHD.

Keywords: Artificial intelligence, congenital heart disease, machine learning, pediatric cardiology

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Introduction

Congenital heart diseases (CHD) demonstrate multifactorial etiology, arising from sophisticated interactions between genetic predisposition and environmental influences. Literature examining neonatal populations reports CHD incidence at approximately 8 cases per 1,000 live births (1,2). Research evidence suggests elevated CHD incidence during prenatal periods, correlating with increased rates of spontaneous pregnancy loss and fetal demise, alongside elective pregnancy terminations following prenatal diagnosis (3). Within this clinical context, genetic specialists, gynecological and obstetric practitioners, and particularly pediatric cardiovascular specialists must possess a comprehensive understanding of CHD risk factors and employ timely diagnostic methodologies for effective clinical management (4).

Primary diagnostic approaches in pediatric cardiovascular medicine following comprehensive clinical evaluation encompass electrocardiographic (ECG) and echocardiographic (ECHO) assessments (5). However, cardiac computed tomography (CT) and cardiac magnetic resonance imaging (MRI) are progressively utilized as diagnostic tools in subsequent clinical phases (6). Regarding CHD, particularly for prenatal or postnatal monitoring, diagnoses are generally established through ECHO evaluation when clinical suspicion emerges. Nevertheless, timely diagnostic achievement depends upon clinical awareness, available hospital infrastructure, and accessibility of experienced specialists (7). Navigating these diagnostic processes can prove time-intensive for both patients and healthcare providers. Furthermore, the broad spectrum of conditions and limitations in clinical decision support systems for preoperative and postoperative care contributes to increased clinician burden.

Through technological advancement, artificial intelligence (AI) tools, particularly machine learning (ML) and deep learning, have gained increasing prominence in medical practice, reflecting their adoption across various scientific disciplines. ML utilizes statistical modeling to identify patterns within historical data, enabling computational systems to predict future scenarios under comparable conditions.

The expanding implementation of AI has been enhanced by workforce growth and accumulated expertise within this domain. However, the application of by numerous physicians and healthcare professionals appears limited to conversational AI systems. Nevertheless, with appropriate data integration and system training, AI demonstrates substantial potential, particularly in areas such as CHD, characterized by significant disease variability and numerous diagnostic and therapeutic approaches.

Contemporary literature regarding AI applications primarily emphasizes developing diagnostic algorithms and postoperative monitoring in CHD. The primary challenge involves limited availability of large datasets, necessary for creating AI algorithms in single-center studies, particularly for rare conditions like CHD. Consequently, there is an urgent need for multicenter studies or extended data collection periods to accumulate sufficient information in this field.

AI is increasingly utilized in clinical applications, including diagnosis, monitoring, and treatment of CHDs, contributing to notable advances in pediatric cardiovascular medicine. The capacity to enhance diagnostic accuracy of imaging modalities such as cardiac MRI, echocardiography, cardiac CT angiography, and electrocardiography through AI algorithms enables more reliable and earlier detection of CHDs in both prenatal and postnatal stages.

Recently developed AI-based models are being implemented across various subspecialties within pediatric cardiology, including screening protocols, physical examination findings evaluation, diagnostic process support, medical image analysis, prognosis prediction, risk assessment, and personalized patient-specific medical approaches. ML techniques can also predict complication risk and progression, offering opportunities for preventive interventions (8).

However, AI technology integration into comprehensive care for children with CHD remains limited in the current literature, thereby hindering the full realization of its potential in this area. In complex diseases like CHD, the diversity of treatment options, especially postoperative approaches, can be simulated to create patient-specific treatment plans, potentially reducing mortality and morbidity. Furthermore, utilizing monitoring or wearable devices during postoperative periods can facilitate arrhythmia detection and the development of early warning systems for healthcare personnel. Additionally, AI can assist by providing relevant educational materials for early diagnosis and triage in rural areas where specialist healthcare personnel are scarce.

This survey aims to explore challenges and opportunities associated with integrating AI technologies into pediatric cardiology. Addressing these points will help improve healthcare quality and patient outcomes in pediatric cardiology.

Types of Machine Learning Models

ML, a computer science subdiscipline, simulates human cognitive processes through algorithms designed to learn from data inputs. As a fundamental component of big data analytics, ML finds application in diverse fields including pattern recognition, computer vision, and biomedical research. ML enables computational systems to derive insights from data, thereby providing predictive and decision-making capabilities without dependence on explicit programming protocols (9). This involves training algorithms on comprehensive datasets to identify significant patterns and iteratively improve performance metrics. Within medical applications, ML algorithms emerge as essential tools, capable of processing complex datasets to enhance diagnostic process precision, personalize therapeutic strategies, and predict patient prognoses (10,11).

ML is generally structured around three fundamental paradigms: supervised, unsupervised, and reinforcement learning models. Supervised learning involves training algorithms on datasets where each input is paired with a corresponding, predefined output label. This approach enables models to infer patterns between input features and expected outcomes, allowing for predictions on new, unobserved instances. It is widely applied in predictive tasks, particularly classification and regression problems. While classification models categorize data into distinct groups, regression models are used to predict values along a continuous spectrum.

Common classification techniques comprise logistic regression, decision trees, random forests, support vector machines (SVM), K-nearest neighbors (K-NN), naïve Bayes, and artificial neural networks. In the context of regression, frequently utilized models include linear and polynomial regression, ridge, and lasso regression, elastic net, support vector regression, and neural networks (12,13).

Unsupervised learning, by contrast, operates on data lacking explicit labels, with the goal of uncovering intrinsic structures or relationships within the dataset. Key methodologies in this category include clustering, dimensionality reduction, and mining for association rules. Clustering methods—such as K-means and hierarchical clustering—organize similar data points based on feature resemblance. Meanwhile, dimensionality reduction techniques like Principal Component Analysis minimize the feature set while preserving essential information content (14).

Reinforcement learning, the third paradigm, centers on training an agent to take a series of actions within an environment to optimize long-term cumulative rewards (15).

Figure 1 presents various types of ML models, and the subsequent section elucidates commonly used predictive models.

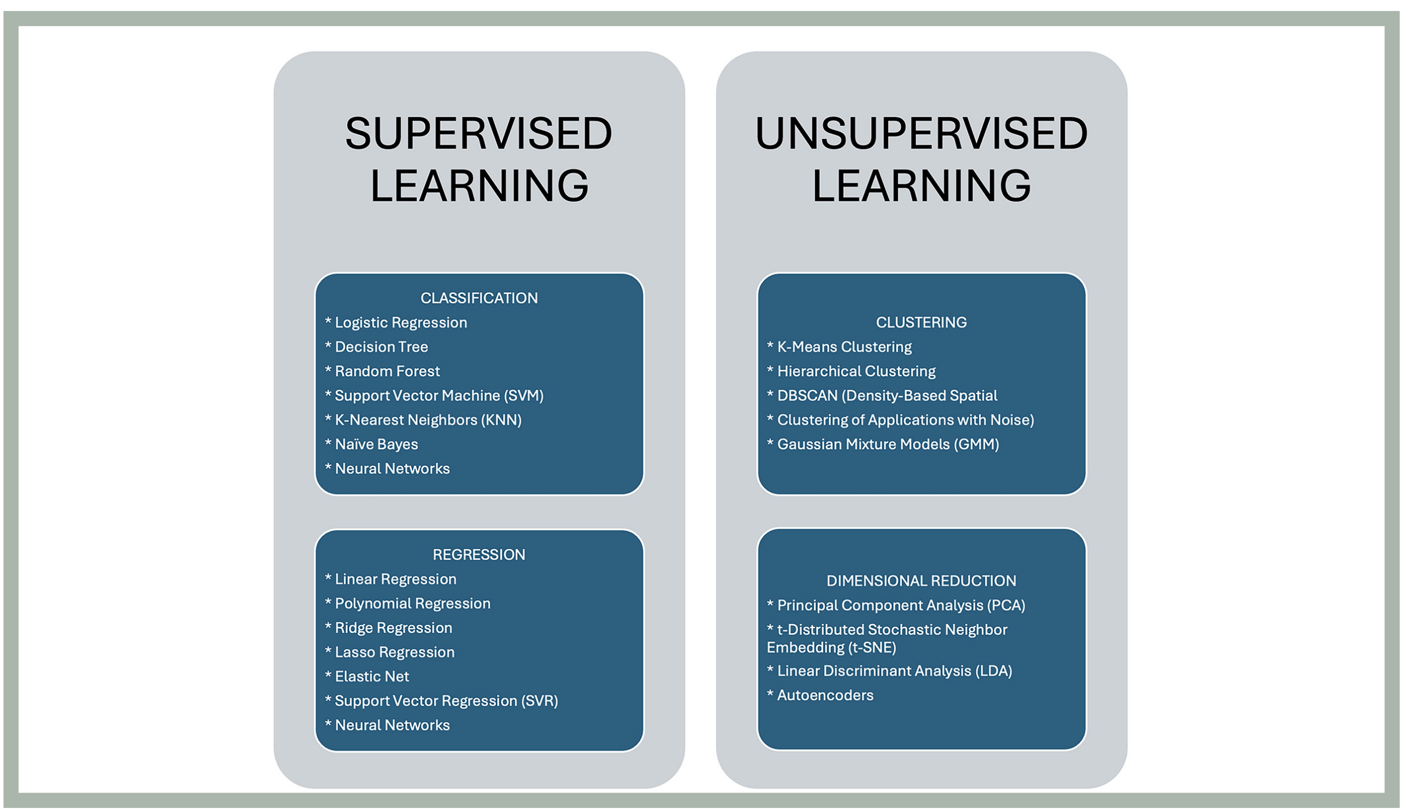

Figure 1. Types of machine learning

Data Classification and Predictive Models

Outcome prediction, risk assessment, and customization of treatment and follow-up for CHD patients now involve various ML models and AI tools, each with distinct advantages and limitations. A predictive model constitutes a mathematical equation designed to forecast outcomes based on one or more input variables. This tool employs data and algorithms to predict outcomes. Given a defined set of measured attribute values for an object x, the objective is to predict the unknown value of another attribute y. The attribute y is designated as the “output” or “response” variable, while the set $x = \{x_n, \cdots, x_1\}$ constitutes the “input” or “predictor” variables. The essence of predictive, also known as machine, learning is to construct a prediction function f(x) that approximates y with minimal error.

Data classification is a supervised learning method, that assigns new observations to one of several predefined categories, based on quantitative attribute Emser et al. (16). Data classification can be approached through two methods: one focuses on creating binary distinctions between two classes, assigning labels of 0 or 1 to data items, while the other aims to model P(y|x), providing both class labels and class membership probabilities. SVM exemplify the former approach, whereas logistic regression, decision trees, artificial neural networks, and K-NN represent the latter, differing in their data approximation methods.

SVM utilize statistical learning theory to estimate model performance on new data by considering the characteristics of the data and the performance on the training dataset performance. They create boundaries between datasets by solving complex optimization problems. Different kernel functions allow varying levels of model flexibility. Because they are founded on statistical principles, these machines have been studied extensively. They have demonstrated performance comparable to or better than other ML algorithms in medical studies. A disadvantage of SVM is that classification results only show simple divisions, without indicating class membership probabilities (17).

Logistic regression does not require a direct linear relationship between the independent and dependent variables themselves, but instead assumes linearity between the log-odds of the outcome and the predictor variables (18). This model is fundamentally based on the concept of odds in the context of binary outcomes. When focusing on a specific event, its odds are defined as the probability of occurrence relative to the probability of non-occurrence. These odds are often used to represent the likelihood of an event. The logistic regression framework applies the natural logarithm of the odds—referred to as the logit—as a linear function of the predictor variables.

In the case of a single predictor variable, denoted as XXX, the model can be written as:

ln^{f0}(odds)=β0+β1X\ln(\text{odds}) = \beta_0 + \beta_1Xln(odds)=β0+β1X

where ln^{f0}\lnln denotes the natural logarithm, β0\beta_0β0 is the intercept, and β1\beta_1β1 represents the coefficient for XXX. The coefficient β1\beta_1β1 indicates the change in the log-odds of the outcome for each one-unit increase in XXX. Since the difference between logarithms corresponds to the logarithm of a ratio, exponentiating β1\beta_1β1 provides the odds ratio, reflecting how the odds change with a one-unit increase in the predictor variable (19).

Decision trees represent a ML technique that creates models without requiring specific data requirements or assumptions (20). Before examining decision trees, it is essential to establish a foundation of terminologies. The root node serves as the starting point, initiating dataset division based on features or conditions. Decision nodes arise from subsequent root node splitting, representing intermediate decisions within the tree structure. Conversely, leaf nodes signify terminal points where further division is infeasible, denoting final classifications or outcomes. A sub-tree, analogous to a subgraph, constitutes a specific section of the overall decision tree. Pruning involves selective node removal to mitigate overfitting and enhance model simplicity. Branches or sub-trees represent distinct pathways of decisions and outcomes within the tree.

In hierarchical models such as decision trees, parent nodes represent decision criteria or conditions, while child nodes denote possible outcomes or subsequent decisions based on those criteria. These structures are used to derive solutions by analyzing previously resolved instances. The process typically begins by dividing the dataset into two subsets: one for training, where the tree is constructed, and one for testing, where the accuracy of the resulting decisions is validated. Each instance in the dataset is described by a set of attributes, one of which is selected to guide the decision-making process. All input attributes are assigned value categories—discrete attributes with limited unique values, form their own categories, while continuous or highly varied numeric attributes are grouped into defined intervals.

Within the tree, attributes are represented as internal or decision nodes, each branching into paths corresponding to different value categories. The terminal nodes, or leaves, of the tree indicate decision outcomes, effectively mapping the predicted class for the decision attribute. To classify a new, unseen instance, traversal begins at the root node and proceeds along branches that match the instance's attribute values, until a leaf node is reached, signifying the final decision (21).

While binary classification (e.g., positive vs. negative) is common, decision trees can also be extended to multi-class scenarios to accommodate more complex decision categories. Early theoretical foundations for decision trees were introduced by Clopper and Pearson (22) in 1934 through binary decision frameworks. However, practical applications in ML gained momentum later. In 1984, Leo Breiman proposed the Classification and Regression Tree algorithm, which popularized concepts like binary splits and the Gini impurity metric—both now standard in decision tree construction (23). Subsequently, Quinlan (24,25,26) introduced the ID3 algorithm in 1986, followed by the improved C4.5 model in 1993. These innovations paved the way for the integration of decision trees into ensemble learning approaches such as random forests and boosting techniques, cementing their role as foundational tools in modern ML.

The K-NN algorithm is another simple yet effective classification technique. It assigns a class to a new data point based on the majority class among its k closest neighbors in the feature space. Unlike some models, K-NN lacks a generalization phase, which may hinder interpretability. However, it offers transparency by presenting specific training instances that influenced a decision. This case-based reasoning is often viewed favorably in medical contexts, as it reflects clinical decision-making based on prior similar experiences (27).

Naïve and semi-naïve Bayes methods are simpler and faster than other classifiers (28). Although often outperformed by models like logistic regression or random forests, certain models remain popular for tasks such as text classification and spam filtering, especially where computational resources are limited (29). Naïve and semi-naïve Bayes methods utilize conditional probability tables. The decisions made by Bayesian classifiers, can be seen as aggregating information gains. The formula used to calculate the information needed to determine if something belongs to class C explains decisions by combining information gains that either support or oppose the class. This method works for semi-naïve Bayes as well, but it uses combined attribute/value pairs instead of simple values. This information gain can be arranged in a table to show the evidence for or against a decision (30).

The Application of Machine Learning Methods in Congenital Heart Disease

ML models, and AI-driven methods are transforming healthcare with innovative approaches to enhance diagnosis, treatment, and management of illnesses, particularly concerning pediatric cardiac conditions. This section explores how AI can contribute to addressing these complex cardiac issues. Furthermore, it examines how AI can improve diagnostic accuracy and personalize treatments, ultimately leading to enhanced outcomes for young patients.

Between 2011 and 2017, Qu et al. (31) conducted research at a prominent cardiac center in China, comprehensively examining unborn infants for potential cardiac defects via ultrasonography. Suspicious cases underwent confirmation through echocardiograms conducted by at least two pediatric cardiology specialists. The study scrutinized a total of 1,127 potential predictors of cardiac anomalies, employing an Explainable Boosting Machine to forecast defects and evaluating performance using Receiver Operating Characteristic curves. Predictors were prioritized based on their predictive contribution, and corresponding thresholds were established. The study encompassed 5,390 mother-child pairs, with the predictive model achieving a 76% accuracy rate. Predominantly, the top 35 predictors comprised laboratory test results, with only a single predictor originating from questionnaire data. The model exhibited an overall accuracy of 0.65, with sensitivity and specificity values of 0.74 and 0.65, respectively. Maternal uric acid levels, glucose levels, and blood clotting efficiency emerged as the most reliable and influential predictors of cardiac defects. Threshold analysis indicated that elevated uric acid levels, shortened activated partial thromboplastin time, and elevated glucose levels were the most salient predictors, correlating with 1.17-1.54 times, increased risk of cardiac defects. Based on these findings, the authors developed an online tool designed to facilitate screening and prevention of cardiac defects.

Owens et al. (32) used data from the Statewide Planning and Research Cooperative System spanning January 1, 2000, to December 31, 2014, to investigate maternal delivery hospitalizations and associated neonatal admissions among women diagnosed with cardiomyopathy, adult congenital heart disease (ACHD), pulmonary hypertension (PH), and valvular heart disease. The study employed the International Classification of Diseases, Ninth Revision, Clinical Modification, to identify cases and capture maternal major adverse cardiac events (MACE), neonatal clinical complications, and obstetric outcomes. Outcomes were analyzed using multivariate logistic regression.

Among the 2,284,044 delivery admissions reviewed, 3,871 involved women with cardiac conditions: 676 with cardiomyopathy, 1,528 with valvular heart disease, 1,367 with ACHD, and 300 with PH. Major cardiac events were reported in

16.1% of those with heart disease, with the highest incidence among patients with cardiomyopathy and PH. Neonatal complications were also more frequent among offspring of affected mothers, particularly in the cardiomyopathy and PH subgroups. Women with heart disease showed a significantly elevated risk of neonatal adverse outcomes, and this risk was most pronounced in the cardiomyopathy and PH cohorts. Independent predictors for neonatal complications included preeclampsia, MACE, preexisting diabetes mellitus, and other obstetric issues.

In another study, Xu et al. (33) retrospectively examined pediatric patients diagnosed with infective endocarditis between January 2010 and December 2021 at a single institution. Data collected included demographics, clinical symptoms, microbiological findings, ECHO data, pre-existing cardiac conditions, and outcomes. The study included 90 children, with 60% having a history of heart disease. *Staphylococcus aureus* emerged as the most commonly isolated pathogen, observed more frequently in patients without structural heart disease compared to those with underlying heart conditions.

ECHO analysis identified vegetations in 88 patients: 41 on the left side of the heart, 45 on the right, and 2 cases with bilateral involvement. Right-sided vegetations were more often seen in patients with heart disease, while left-sided vegetations predominated in those without. Embolic events were documented in 25 cases, with a higher incidence in the non-heart disease group. Spontaneous resolution of vegetations occurred in nine patients, and four patients died during hospitalization. Logistic regression indicated that the absence of structural heart disease and the presence of moderate to severe valvular dysfunction were independent risk factors for embolic complications.

Babič et al. (34) examined cardiovascular disease, encompassing a diverse array of conditions that impair heart and vascular function, including coronary artery disease, arrhythmias, and congenital or acquired structural abnormalities. The term is frequently associated with vascular obstructions or stenoses that may precipitate acute events such as heart attacks, angina, or cerebrovascular incidents. In their study, three datasets were analyzed: the Heart Disease Database, the South African Heart Disease Dataset, and the Z-Alizadeh Sani Dataset. The team applied various ML models, specifically Decision Trees, naïve Bayes, SVM, and Neural Networks, for predictive analytics, supplemented by descriptive methods based on rule-based association and decision logic.

Model performance was evaluated using data from multiple sources. For the Cleveland, Hungary, Switzerland, and Long Beach VA datasets, classification accuracies were reported as 88.09% for Decision Trees, 86.76% for naïve Bayes, 88.53% for SVM, and 89.93% for Neural Networks. Model accuracy rates on the South African dataset were slightly lower: Decision Trees achieved 73.87%, naïve Bayes 71.17%, SVM 73.70%, and Neural Networks 68.48%. With respect to the Z-Alizadeh Sani dataset, the results indicated accuracies of 85.38% for Decision Trees, 83.33% for naïve Bayes, 86.67% for SVM, and 86.32% for Neural Networks. Based on these findings, the researchers concluded that the implemented models yielded robust and consistent outcomes, often matching or surpassing those found in comparable studies.

In a separate investigation, Pachiyannan et al. (35) introduced a novel ML-based framework designed to reduce neonatal mortality in cases of CHD. The model processes infant medical records to pinpoint critical mortality risk factors, thereby supporting timely clinical intervention and individualized care strategies for neonates at elevated risk. By integrating maternal health history and prenatal indicators, the diagnostic tool facilitates accurate evaluation of newborns affected by CHD. The Cardiac Deep Learning Model yielded encouraging performance metrics, demonstrating a sensitivity of 91.74%, specificity of 92.65%, a positive predictive value of 90.85%, negative predictive value of 55.62%, and a miss rate of 91.03%. These findings imply that the model may serve as a valuable clinical resource, equipping healthcare providers with decision support capabilities to mitigate CHD-related neonatal mortality and enhance treatment outcomes.

Lee et al. (36) carried out a retrospective analysis involving ECG data from 1,035 pediatric patients aged under five at Chang Gung Memorial Hospital in Taoyuan, Taiwan. Based on ECG interpretations, patients were grouped into five diagnostic categories: normal cardiac anatomy, non-significant right heart disease, significant right heart disease, non-significant left heart disease, and significant left heart disease. ECG signals underwent preprocessing via continuous wavelet transformation and were then divided into two-second segments to augment the dataset. Following this, transfer learning was implemented using three pre-trained deep learning architectures: ResNet-18, InceptionResNet-V2, and NasNetMobile. These models were assessed using standard classification metrics, including accuracy, sensitivity, specificity, F1 score, and the area under the receiver operating characteristic curve (AUC-ROC).

Among the evaluated models, ResNet-18 achieved the best overall results for identifying clinically significant CHD, reaching an accuracy of 73.9%, F1 score of 75.8%, and AUC of 81.0%. While InceptionResNet-V2 demonstrated strong performance in detecting left-sided heart abnormalities, it required significantly more computational resources.

Notably, the AI models outperformed pediatric cardiologists' interpretations of conventional ECGs. The authors highlighted the potential of AI-enhanced ECG analysis as a valuable adjunct tool in CHD screening for young children, with ResNet-18 emerging as a particularly effective model.

Niyogi et al. (37) examined the transformative role of AI in the diagnosis, treatment, and lifelong management of CHD. The study addressed recent progress in prenatal detection, postnatal intervention, and chronic monitoring, while also recognizing limitations such as the lack of standardized datasets and ethical complexities. Notable advancements include the use of AI in fetal echocardiography and genetic screening, facilitating more precise prenatal risk assessment. Additionally, the integration of AI into imaging diagnostics has improved classification and severity evaluation of CHD subtypes. The authors also emphasized the role of AI-driven clinical decision support systems, which contribute to personalized care plans and better prognostic evaluations. Remote AI monitoring tools were also noted for their potential to detect complications early, supporting long-term patient management.

Chen et al. (38) introduced a deep learning–based diagnostic system, Congenital Heart Disease diagnosis via Electrocardiogram (CHDdECG), designed for the detection of congenital heart defects using pediatric ECGs. This approach combines automated feature extraction via wavelet transformation with selected expert-identified features. Trained on a dataset of 65,869 cases, CHDdECG achieved a ROC-AUC of 0.915 and a specificity of 0.881 on a real-world test set of 12,000 cases. On two external validation datasets (7,137 and 8,121 cases), the model yielded ROC-AUC scores of 0.917 and 0.907, and specificities of 0.937 and 0.907, respectively. The system outperformed cardiologists in CHD detection, with automatically extracted features contributing more significantly to model performance than manually selected ones. These findings highlight the promise of ECG-based deep learning for pediatric CHD screening, offering insights beyond conventional diagnostic approaches.

Conclusion

The studies reviewed underscore the significant potential of AI to revolutionize cardiovascular medicine by enhancing diagnostic precision, enabling personalized treatment approaches, and ultimately improving patient outcomes. AI algorithms have demonstrated their capability to analyze intricate datasets derived from electrocardiograms and advanced imaging modalities. This facilitates early and accurate detection of critical cardiovascular conditions, such as coronary heart disease and congenital heart defects, often surpassing the performance of conventional diagnostic techniques.

Moreover, decision support systems powered by AI offer considerable potential in personalizing treatment plans and enhancing the precision of outcome predictions. The integration of AI into remote patient monitoring facilitates the early identification of clinical complications, allowing for prompt intervention and better long-term disease management. Collectively, these technological advancements are poised to substantially influence healthcare delivery by providing clinicians with sophisticated tools to reduce cardiovascular-related mortality, improve patient outcomes, and support the timely and customized care of individuals at elevated risk.

Future research should focus on standardizing datasets, addressing ethical considerations, and validating AI models across diverse populations to fully realize the transformative potential of AI in cardiovascular care.

Authorship Contributions

İ.Z.G.: Supervision, Study planning, Methodology Planning, and Main manuscript editing.

E.K.: Main manuscript writing, Revision, Screening, Data extraction.

All authors have read and agreed to the published version of the manuscript.

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