Machine Learning-Based for Congenital Heart Disease

Subjects: Computer Science, Interdisciplinary Applications

Contributor: Prabu Pachiyannan, Musleh Alsulami, Deafallah Alsadie, Abdul Khader Jilani Saudagar, Mohammed AlKhathami, Ramesh Chandra Poonia

Congenital heart disease (CHD) represents a multifaceted medical condition that requires early detection and diagnosis for effective management, given its diverse presentations and subtle symptoms that manifest from birth. IoMT addresses the crucial challenge of timely identification and detection of cardiovascular diseases by utilizing IoMT data, aiming to enhance the accuracy and efficiency of diagnosis, which in turn enables prompt intervention and improves patient outcomes.

Keywords: healthcare ; internet of medical things ; congenital heart disease ; classification ; prediction

1. Introduction

Cardiovascular disease is a significant medical condition that affects heart performance and leads to complications such as coronary artery disease and impaired vascular function [1]. These difficulties can result in myocardial infarction and cerebrovascular accidents. According to a survey, heart disease annually impacts an estimated 620,000 individuals in the United States [2]. While heart disease can affect both genders, males are more vulnerable. Statistics from 2010 show that a quarter of all fatalities were attributed to heart disease. In the United States, there are approximately 738,000 cases of heart attacks, with 528,000 of these cases being initial occurrences. The remaining 220,000 individuals experience subsequent episodes. Symptoms of heart disease include chest tightness, pain and discomfort, shortness of breath, ankle swelling, neck and abdominal pain, rapid heartbeat, dizziness, cardiac arrest, fainting, changes in skin color, ankle irritation, weight loss, and fatigue. The manifestation of symptoms depends on the type of cardiovascular ailment, which include but are not limited to arrhythmia, myocardial infarction, heart failure, congenital coronary artery disease, mitral valve insufficiency, and dilated cardiomyopathy.

Congenital heart disease (CHD) refers to a group of structural abnormalities in the heart that occur during the prenatal stage of development [3]. These congenital abnormalities manifest during the prenatal period and affect the morphology and physiology of the heart, leading to various cardiovascular complications. CHD is a common congenital anomaly around the world, imposing a significant health burden on affected individuals and medical systems alike. The global incidence of CHD shows significant variation, with an estimated occurrence of approximately 1% of all live births [4]. In the United States, it is believed that around 40,000 newborns are affected by CHD each year. The severity of the condition can vary, ranging from individuals who experience minimal to no symptoms to those who require immediate medical attention. CHD encompasses a diverse range of anomalies, including structural malformations in the heart valves, walls, and vasculature. Examples of frequently encountered instances of CHD include atrial septal defects, ventricular septal defects, and Tetralogy of Fallot [5].

It is widely recommended that all pregnant women worldwide undergo fetal evaluation and ultrasound between 18 and 24 weeks of gestation [6]. This procedure involves detailed imaging, including ultrasound scans of the heart, which has the potential to identify over 90% of severe congenital heart conditions. However, despite the widespread use of fetal ultrasound technology, the prevalence of fetal detection for genetic cardiovascular diseases within the community ranges from 30 to 50% [7]. The hypothesis suggests that the main reason for the significant disparity in diagnoses is insufficient and inconsistent proficiency in analyzing fetal cardiac images [8]. This is primarily due to the complexity of detecting a small and rapidly beating fetal heart as well as the relatively low awareness of congenital coronary artery disease among healthcare providers, given its low incidence. Although clinical quality assurance efforts focused on a single center and conducted on a small scale have shown promising results in improving CHD detection rates by up to 100%, the sustainability and scalability of such programs present significant challenges. To address this, experiments were conducted to determine if utilizing machine learning (ML) analysis of images could improve the evaluation rates typically observed in community medicine [9]. This was achieved by training the ML model on data from a limited number of clinically relevant imaging studies.

Machine learning has been demonstrated to be proficient in detecting intricate image patterns and successfully applied in adult cardiovascular ultrasound technology [10]. It has even surpassed the performance of physicians in tasks involving the classification of views, using small and downsampled databases. However, despite its widespread usage across various domains, the application of machine learning in the context of CHD or fetal ultrasound still requires further refinement. The use of deep learning in medical scenarios that are inherently rare presents inherent limitations, irrespective of the volume of training data available.

2. Machine Learning in Coronary Heart Disease

Various conventional techniques in machine learning have been employed to address the challenges associated with manually analyzing electrocardiogram (ECG) signals in Coronary Heart Disease (CHD). The conventional machine learning approach involves several steps, including preprocessing, feature extraction, feature selection, and categorization processes. Differentiating between normal and CHD signals based on their distinctive characteristics is a time and resource-intensive task. The robustness of the features obtained is significantly impacted by the quality of the underlying data. Preprocessing steps, such as noise elimination and R-peak identification, are essential to extract crucial attributes needed for effective categorization. This research suggests leveraging machine learning to improve the efficiency of an automated CHD diagnosis method, aiming to overcome the limitations associated with traditional machine learning approaches. Machine learning algorithms play a crucial role in acquiring and recognizing unique features from input ECG signals. The goal is to enhance the accuracy and effectiveness of the diagnostic process for CHD through the utilization of advanced machine learning techniques.

In their study, Xu et al. [11] presented a novel methodology for the automated classification of pediatric Congenital Heart Disease (CHD) through the analysis of heartbeats. The researchers conducted an extensive extraction of diverse features from normal heart signals, encompassing characteristics derived from the time domain, frequency domain, and wavelets. Employing machine learning methodologies, particularly random forest and support vector machines, the proposed approach demonstrated promising outcomes. The results revealed a commendable accuracy rate of 87.5% in effectively categorizing CHD cases. Additionally, the specificity and sensitivity values were noteworthy, standing at 89.7% and 85.2%, respectively. These findings underscore the efficacy of the devised method in reliably identifying pediatric CHD through the analysis of heartbeats, showcasing its potential as a valuable diagnostic tool in this medical context.

Ng et al. [12] designed an automated framework aimed at classifying perioperative hazards in patients with complex Congenital Heart Disease (CHD) by leveraging retinal images. The authors introduced an innovative feature extraction method that harnessed both color-based and texture-based characteristics obtained from retinal images. Subsequently, these extracted features were employed in risk classification through the application of machine learning, specifically utilizing a support vector machine. Results from the implemented framework demonstrated a notable predictive accuracy, achieving an impressive rate of 84.9% in effectively identifying perioperative risks in patients diagnosed with complex congenital heart disease. This research highlights the potential of utilizing retinal images and advanced machine learning techniques as a valuable tool for automating the identification of perioperative hazards, thereby contributing to enhanced patient care and risk management in the context of complex CHD cases.

Kobel et al. [13] conducted a thorough assessment of the Apple Watch iECG's effectiveness in detecting Congenital Heart Disease (CHD) in children. The study involved obtaining iECG measurements from pediatric patients, including those with and without CHD. A meticulous comparative analysis was performed by juxtaposing the iECG data against conventional ECG records. The outcomes of this investigation suggest a promising role for the Apple Watch iECG as a potential screening tool for CHD in children, revealing a sensitivity of 92% and an accuracy of 93% in identifying the condition. In a separate study led by van Genuchten and colleagues [14], the physical capacity of children diagnosed with CHD was evaluated. A cohort of pediatric patients with CHD underwent exercise tests to assess their peak oxygen uptake (VO_2peak). The research findings unveiled a significant revelation—children with CHD exhibited diminished exercise capacity compared to their healthy counterparts, as evidenced by lower VO_2peak measurements. These results underscore the considerable impact of CHD on the ability of pediatric populations to partake in physical activities, shedding light on the broader implications of the condition on the overall well-being of affected individuals.

Kavitha et al. [15] introduced an innovative approach termed Multilayer Deep Detection Perceptron (MLDDP) for the identification of testicular deviations, both with and without Congenital Heart Disease (CHD). This practical method utilized a Multilayer Deep Learning framework that incorporated multiple layers of perceptrons to discern anomalies associated with CHD. Upon evaluating the proposed MLDDP on a provided dataset, it demonstrated an exceptional detection accuracy of 95.4% in precisely identifying testicular deviations, irrespective of the presence of CHD. The success of MLDDP underscores the potential of machine learning techniques in advancing the diagnosis of CHD-related conditions.

In a distinct study, Liu et al. [16] concentrated on the computer-aided analysis of heart sounds in pediatric patients diagnosed with left-to-right shunt CHD. The researchers introduced a methodology leveraging machine learning techniques, specifically employing a Convolutional Neural Network (CNN), to extract pertinent features from heart sound signals and accurately classify the presence of left-to-right shunt CHD. The outcomes were noteworthy, with a precision rate of 90.8% and a region under the receiver operating characteristics curve of 0.935, indicating the method's efficacy in identifying and categorizing CHD with left-to-right shunt. These findings underscore the potential of machine learning-based analyses in supporting medical professionals in diagnosing specific types of CHD, showcasing the promising intersection of technology and healthcare.

Ge et al. proposed an innovative method for identifying Pulmonary Hypertension (PH) associated with Congenital Heart Disease (CHD) by incorporating time–frequency domain analysis and machine learning (ML) characteristics [17]. The researchers integrated time–frequency analysis techniques into an ML framework to extract relevant features from echocardiographic data. Through their approach, the method achieved an impressive precision level of 91.6% in accurately identifying pulmonary hypertension linked to congenital heart disease. This study demonstrates the efficacy of combining advanced signal processing techniques with machine learning approaches to enhance the identification and characterization of specific cardiac conditions, specifically focusing on the challenging context of pulmonary hypertension in the presence of congenital heart disease.

Steeden et al. [18] delved into the exploration of utilizing artificial intelligence (AI) in the assessment of Congenital Heart Disease (CHD). The authors undertook a thorough examination of AI-based methodologies, specifically machine learning (ML), applied to tasks such as image analysis, risk estimation, and detection within the realm of CHD. Their comprehensive analysis provided insightful perspectives on the potential of AI in augmenting the assessment and treatment of individuals with CHD. By shedding light on the various applications of AI in the context of CHD, the study contributes to the evolving landscape of medical technology and its role in advancing cardiac care and diagnostics.

Alici-Karaca et al. introduced a Convolutional Neural Network (CNN) with a lightweight architecture designed to precisely classify cases of radiation-induced liver disease, as detailed in their research publication [19]. While their study does not directly focus on congenital heart disease, it underscores the application of machine learning in medical image analysis. The featured lightweight CNN successfully achieved an impressive classification accuracy rate of 93.1% when tasked with identifying radiation-induced liver disease. This result highlights the versatile capacity of machine learning to be applied across diverse medical conditions, showcasing its potential in aiding accurate diagnoses beyond the specific context of congenital heart disease.

Qiao et al. presented the Residual Learning based Diagnostic System (RLDS), an innovative diagnostic system employing residual learning, designed for cases of fetal Congenital Heart Disease (CHD) [20]. This system utilized residual learning, a type of machine learning, to extract distinctive features from images of fetal echocardiography for discrimination. Remarkably, the diagnostic accuracy of the RLDS for fetal CHD reached an impressive 96.5%. Additionally, the system offered interpretability by generating attention maps and assigning importance scores to features, enhancing the understanding of the diagnostic process for medical professionals. This research underscores the potential of incorporating machine learning, specifically residual learning, in creating advanced diagnostic tools for fetal CHD with the added benefit of interpretability.

The following **Table 1** summarizes key findings from various studies investigating the use of machine learning (ML) in cardiovascular health, focusing on congenital heart disease (CHD) and related conditions. Each entry includes the reference number, author, study objective, and identified limitations. This compilation offers a succinct overview of the objectives pursued by researchers, shedding light on both the potential and challenges associated with ML applications in the diagnosis and assessment of cardiovascular health.

Table 1. Key Findings.

S.No	Author Information	Objective of the Work	Limitations
1	Xu et al. [11]	Automated classification of pediatric CHD through heartbeat analysis using ML	Time and resource-intensive differentiation between normal and CHD signals; Impact of data quality on feature robustness
2	Ng et al. [12]	Automated framework for classifying perioperative hazards in complex CHD using retinal images and ML	Limited generalizability; Dependency on image quality

S.No	Author Information	Objective of the Work	Limitations
3	Kobel et al. [13]	Assessment of Apple Watch iECG in detecting CHD in children	Small sample size; Limited diversity in patient population
4	van Genuchten et al. [14]	Evaluation of physical capacity in children diagnosed with CHD	Small sample size; Limited to exercise capacity assessment
5	Kavitha et al. [15]	Introduction of Multilayer Deep Detection Perceptron (MLDDP) for identifying testicular deviations with or without CHD	Limited validation on diverse datasets; Dependency on data quality
6	Liu et al. [16]	Computer-aided analysis of heart sounds in pediatric patients with left-to-right shunt CHD using CNN	Limited to specific type of CHD; Dependency on quality of input heart sound data
7	Ge et al. [17]	Identification of Pulmonary Hypertension associated with CHD using time–frequency domain analysis and ML	Limited to Pulmonary Hypertension; Generalization to other types of CHD
8	Steeden et al. [18]	Exploration of AI-based methodologies for CHD assessment	Limited discussion on specific limitations; Generalizability to different AI methodologies
9	Alici-Karaca et al. [19]	CNN for classifying radiation-induced liver disease	Limited to radiation-induced liver disease; Dependency on image quality
10	Qiao et al. [20]	Introduction of RLDS for diagnosing fetal CHD using residual learning	Limited to fetal CHD; Dependency on the quality of fetal echocardiography images

References

1. Laggerbauer, B.; Engelhardt, S. MicroRNAs as therapeutic targets in cardiovascular disease. J. Clin. Investig. 2022, 132, e159179.
2. O’Sullivan, J.W.; Raghavan, S.; Marquez-Luna, C.; Luzum, J.A.; Damrauer, S.M.; Ashley, E.A.; O’Donnell, C.J.; Willer, C.J.; Natarajan, P. Polygenic risk scores for cardiovascular disease: A scientific statement from the American Heart Association. Circulation 2022, 146, e93–e118.
3. Sudheer, K.R.; Mohammad Koya, P.K.; Prakash, A.J.; Prakash, A.M.; Manoj Kumar, R.; Shyni, S.; Jagadeesan, C.K.; Jaikrishan, G.; Das, B. Evaluation of risk due to chronic low dose ionizing radiation exposure on the birth prevalence of congenital heart diseases (CHD) among the newborns from high-level natural radiation areas of Kerala coast, India. Genes Environ. 2022, 44, 1.
4. Lin, H.; Ye, S.Q.; Xu, Z.H.; Penaloza, J.S.; Aljuhani, M.; Vetter, T.; Zhao, M.T.; McBride, K.L. Generation and characterization of a human induced pluripotent stem cell (iPSC) line from a patient with congenital heart disease (CHD). Stem Cell Res. 2022, 65, 102958.
5. Xu, W.; Yu, K.; Ye, J.; Li, H.; Chen, J.; Yin, F.; Xu, J.; Zhu, J.; Li, D.; Shu, Q. Automatic pediatric congenital heart disease classification based on heart sound signal. Artif. Intell. Med. 2022, 126, 102257.
6. Wong, J.; Kohari, K.; Bahtiyar, M.O.; Copel, J. Impact of prenatally diagnosed congenital heart defects on outcomes and management. J. Clin. Ultrasound 2022, 50, 646–654.
7. Bottelli, L.; Franzè, V.; Tuo, G.; Buffelli, F.; Paladini, D. Prenatal detection of congenital heart disease at 12–13 gestational weeks: Detailed analysis of false-negative cases. Ultrasound Obstet. Gynecol. 2023, 61, 577–586.
8. Gearhart, A.; Dwork, N.; Jone, P.N. Artificial intelligence in echocardiography to diagnose congenital heart disease and fetal echocardiography. Intell.-Based Med. 2022, 6, 100082.
9. Truong, V.T.; Nguyen, B.P.; Nguyen-Vo, T.H.; Mazur, W.; Chung, E.S.; Palmer, C.; Tretter, J.T.; Alsaied, T.; Pham, V.T.; Do, H.Q.; et al. Application of machine learning in screening for congenital heart diseases using fetal echocardiography. Int. J. Cardiovasc. Imaging 2022, 38, 1007–1015.
10. Ge, B.; Yang, H.; Ma, P.; Guo, T.; Pan, J.; Wang, W. Detection of pulmonary hypertension associated with congenital heart disease based on time-frequency domain and deep learning features. Biomed. Signal Process. Control 2023, 81, 104316.
11. Suvorov, V.; Loboda, O.; Balakina, M.; Kulczycki, I. A New Three-Dimensional (3D) Printing Prepress Algorithm for Simulation of Planned Surgery for Congenital Heart Disease. Congenit. Heart Dis. 2023, 18, 491–505.

12. Ng, W.W.; Liang, H.; Peng, Q.; Zhong, C.; Dong, X.; Huang, Z.; Zhong, P.; Li, C.; Xu, M.; Sun, Y.; et al. An automatic framework for perioperative risks classification from retinal images of complex congenital heart disease patients. Int. J. Mach. Learn. Cybern. 2022, 13, 471–483.
13. Kobel, M.; Kalden, P.; Michaelis, A.; Markel, F.; Mensch, S.; Weidenbach, M.; Riede, F.T.; Löffelbein, F.; Bollmann, A.; Shamloo, A.S.; et al. Accuracy of the Apple Watch iECG in children with and without congenital heart disease. Pediatr. Cardiol. 2022, 43, 191–196.
14. van Genuchten, W.J.; Helbing, W.A.; Ten Harkel, A.D.; Fejzic, Z.; Md, I.M.K.; Slieker, M.G.; van der Ven, J.P.; Boersma, E.; Takken, T.; Bartelds, B. Exercise capacity in a cohort of children with congenital heart disease. Eur. J. Pediatr. 2023, 182, 295–306.
15. Kavitha, D.; Renumadhavi, C.H. An efficient multilayer deep detection perceptron (MLDDP) methodology for detecting testicular anomalies with or without congenital heart disease (TACHD). J. Supercomput. 2022, 78, 4057–4072.
16. Liu, J.; Wang, H.; Yang, Z.; Quan, J.; Liu, L.; Tian, J. Deep learning-based computer-aided heart sound analysis in children with left-to-right shunt congenital heart disease. Int. J. Cardiol. 2022, 348, 58–64.
17. Solvin, H.B.; Diab, S.G.; Elle, O.J.; Holmstrøm, H.; Brun, H. Real-Time Remote-Mentored Echocardiography in Management of Newborns with Critical Congenital Heart Defects. Congenit. Heart Dis. 2023, 18, 551–559.
18. Steeden, J.A.; Muthurangu, V.; Secinaro, A. Artificial Intelligence-Based Evaluation of Congenital Heart Disease. In Artificial Intelligence in Cardiothoracic Imaging; Springer International Publishing: Cham, Switzerland, 2022; pp. 365–376.
19. Alici-Karaca, D.; Akay, B.; Yay, A.; Suna, P.; Nalbantoglu, O.U.; Karaboga, D.; Basturk, A.; Balcioglu, E.; Baran, M. A new lightweight convolutional neural network for radiation-induced liver disease classification. Biomed. Signal Process. Control 2022, 73, 103463.
20. Qiao, S.; Pang, S.; Luo, G.; Pan, S.; Yu, Z.; Chen, T.; Lv, Z. RLDS: An explainable residual learning diagnosis system for fetal congenital heart disease. Future Gener. Comput. Syst. 2022, 128, 205–218.