Advances in the Application of Artificial Intelligence in Heart Disease Detection

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Abstract: Heart disease is one of the leading causes of death and disability worldwide. Early detection and accurate diagnosis are crucial for improving clinical outcomes. In recent years, the application of artificial intelligence (AI) in medical imaging and clinical data analysis has provided new strategies for the automatic detection of cardiac diseases. This review summarizes recent progress in AI-based approaches for detecting aortic stenosis, congenital heart disease, rheumatic heart disease, left ventricular hypertrophy, ventricular function and wall motion abnormalities, and heart failure.
***Keywords*:** Heart disease detection; Echocardiography; Artificial intelligence

1. Introduction

Cardiovascular diseases are the leading cause of mortality and disability worldwide, and heart disease accounts for the majority of these cases. According to the World Health Organization, approximately 18 million people die from cardiovascular diseases each year, representing over one-third of global deaths. Early detection and precise diagnosis are therefore essential to improve prognosis and reduce disease burden.

Conventional diagnostic approaches—including electrocardiography (ECG), echocardiography, cardiac magnetic resonance imaging (MRI), and laboratory testing—are widely used but remain limited by subjectivity, high cost, and dependency on operator experience. Among them, echocardiography is the preferred imaging technique for cardiac evaluation, yet its diagnostic accuracy heavily depends on image quality and clinician expertise. In recent years, artificial intelligence (AI) has emerged as a transformative tool in medical imaging and clinical decision-making. By leveraging deep learning and pattern recognition, AI systems can automatically extract and analyze complex features from echocardiographic images and multimodal data, improving the consistency and efficiency of diagnosis. Studies have demonstrated that AI achieves high accuracy in detecting aortic stenosis, congenital heart disease, rheumatic heart disease, left ventricular hypertrophy, ventricular wall motion abnormalities, and heart failure. However, most current models are trained on small or single-center datasets, and their clinical interpretability and generalizability remain limited.

Therefore, this study aims to review recent progress in AI-based cardiac disease detection, summarize its advantages and current challenges, and discuss future directions toward standardized, explainable, and clinically applicable intelligent diagnostic systems.

2. Research Progress

2.1 Aortic Stenosis

Aortic stenosis (AS) is one of the most common acquired valvular heart diseases, and its early identification is crucial to prevent heart failure and sudden cardiac death. Transthoracic echocardiography (TTE) remains the diagnostic gold standard but is highly dependent on operator skill and therefore difficult to standardize across clinical settings [1]. In recent years, artificial intelligence (AI) techniques have shown significant potential in AS detection. A multiple instance learning (MIL) model based on attention mechanisms can automatically select key echocardiographic views for AS grading, achieving an AUC of 0.923 and markedly improving diagnostic consistency [2]. Furthermore, a hybrid convolutional neural network–long short-term memory (CNN–LSTM) framework has been developed to capture dynamic valvular motion, attaining an AUC of 0.91 in valvular disease detection [3]. Review studies also indicate that AI has made breakthroughs in view recognition, valve segmentation, and disease classification, laying a foundation for an integrated intelligent diagnostic system for valvular diseases [4].

2.2 Congenital Heart Disease

Congenital heart disease (CHD) is the most prevalent structural cardiac abnormality in children, and delayed diagnosis often leads to high infant mortality. Echocardiography remains the first-line diagnostic method, but its complexity and operator dependence limit accuracy. AI provides a promising alternative for automatic CHD detection. A ResNet50-based deep learning model achieved an AUC of 0.91 in classifying pediatric echocardiographic images [5], while a stacked CNN–LSTM model for fetal cardiac videos reached an AUC of 0.945 in distinguishing normal fetuses from those with hypoplastic left heart syndrome (HLHS) [6]. Multiview and multimodal fusion models have further enhanced diagnostic precision: combining two-dimensional echocardiography with Doppler data improved accuracy by 10%–20%, with multimodal fusion yielding an additional 7%–12% gain [7]. Nonetheless, current AI studies are limited by small sample sizes and insufficient multicenter validation [8]. Future work should emphasize the integration of multimodal temporal features and the development of explainable, clinically deployable CHD screening systems.

2.3 Rheumatic Heart Disease

Rheumatic heart disease (RHD) remains a major cause of cardiovascular morbidity in low- and middle-income countries. Echocardiography is the diagnostic gold standard [9], but its widespread application is limited by high cost and professional training requirements. AI combined with portable ultrasound devices provides a feasible solution for large-scale screening. A ResNet-50–based deep learning model achieved an AUC of 0.84 in identifying mitral regurgitation from standard echocardiographic images [10], and a 3D convolutional neural network (C3D) reached 72.77% accuracy, surpassing traditional two-dimensional models [11]. In resource-limited areas, handheld AI-assisted echocardiography achieved an AUC of 0.90 when used by non-specialists [12]. Studies have also demonstrated that simplified screening protocols combined with remote expert review can greatly improve diagnostic accuracy [13]. International guidelines now advocate for integrating AI and handheld ultrasound to promote early RHD detection and community-level screening [14-15].

2.4 Left Ventricular Hypertrophy

Left ventricular hypertrophy (LVH) is a common manifestation of cardiac structural remodeling, often resulting from chronic pressure overload due to hypertension or valvular disease. Early detection is essential to prevent adverse outcomes such as heart failure and arrhythmia. Traditional diagnostic methods rely on ECG and echocardiography but are limited by subjectivity and measurement variability. AI offers an objective and scalable alternative. In a population-based study, AI-assisted point-of-care ultrasound operated by briefly trained non-professionals successfully detected LVH, significantly enhancing community-level screening efficiency [16]. CNN-based models have also demonstrated strong capability in automatically quantifying ventricular wall thickness and chamber size from echocardiograms, improving accuracy and consistency [17]. Furthermore, deep learning applied to chest X-rays has achieved an AUC of 0.80 in detecting LV structural abnormalities [18]. These results highlight AI's ability to perform both quantitative measurements and automatic recognition in LVH assessment, supporting its future integration into multimodal and multicenter diagnostic workflows.

2.5 Ventricular Function and Wall Motion Abnormalities

Assessment of ventricular function and regional wall motion abnormalities (RWMA) is essential for evaluating ischemic heart disease and myocardial performance. Conventional echocardiographic evaluation relies on manual estimation of left ventricular ejection fraction (LVEF) and subjective wall motion scoring, which is prone to inter-observer variability. AI offers a pathway toward standardized and quantitative analysis. Deep convolutional neural networks (DCNN) have been shown to significantly improve measurement accuracy and consistency in LVEF and RWMA evaluation [17]. A three-stage pipeline combining U-Net segmentation, optical flow estimation, and temporal convolutional networks achieved an SVM classification AUC of 0.95, demonstrating robust performance in RWMA detection [19]. In addition, deep learning analysis of chest X-rays has enabled identification of severe LV dilation or hypertrophy with an AUC of 0.80 [20]. Collectively, these studies suggest that AI enables quantitative, automated assessment of ventricular geometry and function, accelerating the transition from subjective interpretation to objective, reproducible cardiac function evaluation.

2.6 Heart Failure

Heart failure (HF) is the terminal stage of various cardiac diseases, characterized by high morbidity and mortality. Accurate early identification and risk prediction are key to improving patient survival. Traditional diagnosis relies on clinical parameters and echocardiography but lacks predictive precision for complex disease trajectories. Recent studies have employed AI-based feature selection and ensemble learning methods to enhance predictive accuracy. An XGBoost model using seven optimized clinical and echocardiographic variables achieved an AUC of 0.89 for one-year all-cause mortality prediction [21]. Similarly, CNN-based models analyzing echocardiographic images demonstrated comparable accuracy (AUC = 0.89) for automated HF detection [22]. Review articles have highlighted AI's advantages in improving diagnostic precision and prognostic stratification but emphasized limitations in generalizability and dataset size [23]. Future research should focus on large-scale, multicenter data integration and interpretable multimodal frameworks combining Echo, ECG, and EHR data to achieve clinically deployable, individualized HF management systems.

3. Limitations and Prospects

3.1 Problems Partially Addressed

In recent years, artificial intelligence has achieved remarkable progress in cardiac disease detection. Deep convolutional neural networks (DCNN), long short-term memory networks (LSTM), and multimodal fusion frameworks have demonstrated high accuracy (AUC > 0.90) in detecting conditions such as aortic stenosis, congenital heart disease, and heart failure. These models significantly enhance diagnostic precision and efficiency compared with conventional methods. Moreover, AI enables automated image segmentation, view recognition, and feature extraction, reducing human intervention and improving consistency across operators. The integration of multimodal datasets—including echocardiography, electrocardiography, and electronic health records—has also shown great potential in risk stratification and prognostic prediction, marking a transition from experience-based to data-driven precision cardiology.

3.2 Unresolved Challenges

Despite these achievements, several challenges remain before AI can be fully integrated into clinical cardiology. The interpretability of deep learning models remains limited; their “black-box” nature makes it difficult for clinicians to understand and trust the underlying decision process [24]. Additionally, most studies are based on small-scale or single-center datasets, lacking multicenter and multiracial validation, which restricts generalizability. Integration of AI tools into clinical workflows is still at an early stage, and the lack of standardized data collection and annotation further complicates clinical translation. Furthermore, strict data privacy regulations and limited data sharing hinder large-scale model training and external validation. Overcoming these barriers will be essential for transforming AI from a research tool into a clinically reliable diagnostic system.

3.3 Future Perspectives

Future developments in AI-based cardiac diagnostics will focus on multimodal integration, interpretability enhancement, and clinical implementation. Establishing large-scale models that combine echocardiography, electrocardiography, laboratory tests, and genomic data will allow for more precise disease classification and individualized risk prediction. Explainable AI (XAI) methods—such as attention visualization and causal reasoning—should be applied to make diagnostic outputs more transparent and clinically interpretable. Furthermore, integrating AI into hospital information systems could enable real-time decision support and dynamic disease monitoring. Low-cost AI-assisted portable ultrasound devices are expected to expand access to early screening in primary care and under-resourced regions. Through multicenter collaboration, data standardization, and interdisciplinary research, AI is poised to become a cornerstone technology in the early detection and precision management of cardiovascular diseases.

4. Conclusion

Artificial intelligence is reshaping the diagnostic paradigm of cardiovascular medicine. By integrating deep learning, temporal modeling, and multimodal data fusion, AI systems have demonstrated expert-level or superior performance in detecting aortic stenosis, congenital heart disease, rheumatic heart disease, left ventricular hypertrophy, ventricular wall motion abnormalities, and heart failure. These technologies significantly improve diagnostic accuracy, automation, and efficiency, providing a powerful foundation for precision cardiology.

However, key barriers such as limited interpretability, small and non-diverse datasets, and insufficient clinical integration continue to restrict large-scale adoption. To address these limitations, future efforts should focus on building explainable and transparent AI frameworks, promoting data standardization and sharing, and conducting multicenter validation studies. With continued technological innovation and interdisciplinary collaboration, AI-assisted diagnostic systems are expected to play an essential role in early screening, accurate diagnosis, and personalized management of heart diseases, ultimately contributing to the reduction of the global cardiovascular disease burden.

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