

A Comprehensive Survey of Explainable Artificial Intelligence And Machine Learning Techniques For Heart Disease Diagnosis And Prediction

M. Dhinesh Kumar¹, S. Malolan², R. Sarveshwaran³, J. Barwesh⁴, A. Sivaramakrishnan⁵

^{1, 2, 3, 4}Dept of Artificial Intelligence and Data Science

⁵Assistant Professor, Dept of Computer Science and Engineering

^{1, 2, 3, 4, 5}Chettinad College of Engineering and Technology, Karur, India

Abstract- Cardiovascular disorders are still causing morbidity and mortality, including coronary heart disease, structural heart disease with congenital heart defects as well as atrial fibrillation. Such measures would be critical for effective early diagnosis and risk stratification that could impact on mortality and long-term complications. Traditional diagnosis methodologies, including electrocardiography (ECG), echocardiography and clinical risk scoring systems, are subject to the experience of clinicians with the underpinning knowledge that human expert may not be sufficient to model complex nonlinear interactions as they exist between many of the aforementioned variables. In recent years, Artificial Intelligence (AI), and in specific Machine Learning (ML), Deep Learning (DL) and Explainable AI (XAI) have shown promising results in providing diagnosis and prognosis of the pathologies of cardiovascular diseases. Ensemble learning and hybrid optimization methods, as well as ECG-based deep learning models provided high-predictive performance. In addition, explainable systems based on SHAP and LIME increase interpretability and clinical trust. Nevertheless, several challenges must be addressed such as data imbalance, poor generalization ability, little multi-center validation and non-transparency. This review offers an extensive overview of existing AI approaches for predicting Heartdisease covering their techniques, datasets, performance metrics, comparative studies, challenges and future work needed for enabling clinically commercialized AI models.

Keywords- Heart Disease Prediction, Structural Heart Disease, Machine Learning, Explainable AI, ECG Analysis

I. INTRODUCTION

Cardiac disorders CVDs are one of the leading global causes of mortality, causing millions death every year. The most common subsets of cardiovascular disease are CHD, SHD, congenital heart defects and AF. Early diagnosis leads to greater patient survival rate and lower health costs.

Conventional diagnostics include the use of electrocardiography (ECG), echocardiography, angiography and risk-scoring models like Framingham Risk Score. Although useful, such methods can be limited either by the subjective nature of interpretation, invasiveness of the procedure or an incapability to model complex nonlinear associations between risk factors.

AI has evolved to be a disruptive paradigm in healthcare analytics. Structured clinical data can be analysed using machine learning algorithms, and high-dimensional ECG and imaging features are considered by deep learning models. The state-of-the-art deep learning models based on ECG have shown great ability for detecting structural heart disease [5], such as large-scale ECG-based deep learning models. Ensemble methods also enhance the predictive stability in structured data [11]. Moreover, interpretable AI models improve transparency and medical trust of the cardiovascular predictive models [1], [3].

In this paper, we describe an organised review of AI-based systems for diagnosing and predicting heart disease, aggregating recent works including methodology, type of data, evaluated performance metrics and the futuristic work in field.

II. PROBLEM FORMULATION AND TASK DEFINITION

AI for heart disease systems mainly fall into two categories: diagnosis and prognosis prediction.

A. Disease Diagnosis

Diagnostic models are used to separate subjects into diseased or non-diseased, or amongst different types of heart disease.

Demir et al. built ML and DL models for CHD diagnosis on UCI and Framingham datasets, achieving high accuracy rate for SVM and XGBoost models [2]. Ansari et al.

proposed a double explainability approach using LIME and permutation feature importance for heart disease prediction with high accuracy of diagnosis [3].

Also, hybrid ML-DL systems, for example XAI-HD allows to include explainable mechanisms besides predictive modeling to increase transparency [1].

B. Severity Prediction and Prognosis

AI not only classifies but also forecasts disease progression, recidivism rate and severity.

Deep learning ECG models also have been shown to be effective in detecting structural abnormalities from big datasets [5]. XGBoost models have been applied to forecast tissue ablation recurrence for atrial fibrillation [14].

Adaptive deep SVM models have been utilized to promote early detection and severity assessment as well [6].

These platforms move beyond binary classifications in the support for treatment planning and risk stratification.

III. DATASETS AND DATA MODALITIES

AI-based cardiovascular systems rely on diverse data sources.

A. Clinical and Laboratory Data

Structured datasets, the UCI Cleveland dataset and Framingham dataset are examples which contain features such as age, cholesterol, blood pressure, chest pain type and maximum heart rate [2], [3].

The stability and robustness of the method can be further improved when using clinical data by applying ensemble learning methods like boosting [11].

B. ECG Signal Data

Electrocardiogram (ECG) signal recordings capture the physiological electrical signals of the heart. Databases of ECGs have been used to train deep learning models for structural heart disease [5].

In addition, hybrid CNN-based ECG image classification frameworks also improve the feature extraction [10].

C. Multi-Institutional and Imaging Data

Others have combined image and patient electronic health record (EHR) data [4]. Nevertheless, most of these studies are based on single-center data and lack generalizability across populations.

IV. FEATURE REPRESENTATION TECHNIQUES

A. Handcrafted and Structured Features

Most conventional ML-based approaches rely on statistical features, such as ST depression, chest pain type, and heart rate variability [3]. The use of feature selection methods and metaheuristic optimization enhances prediction performance [12].

B. Deep Feature Learning

The deep learning based models can learn the hierarchical representation of ECG waveforms and clinical features automatically. CNN and LSTM models are well suited for learning the temporal and spatial patterns in cardiac activities [2], [10].

Explainable frameworks integrate SHAP and LIME for feature attribution and transparency [1].

V. RECOGNITION AND PREDICTION MODELS

A. Classical Machine Learning

The most popular ones are still Logistic Regression, Support Vector Machines, Random Forest and XGBoost [2], [12]. Ensemble methods provide robustness and prevent overfitting [11].

B. Deep Learning and Mixture of Models

Deep learning models learnt from ECG signals have better diagnostic efficiency [5]. Adaptive deep SVM-based methods combine neural embeddings with margin-based classification [6].

Quantum-enhanced ML has also been investigated for predictive improvement: this includes the recent work [9].

C. Explainable AI Frameworks

XAI-HD combines SHAP and LIME explanations for improving clinical interpretability [1]. Algorithm based: Type of machine learning algorithms that offer a mechanism to explain the local and global interpretation [3]. Explainable ensemble models could further enhance clinician trust [11].

VI. EVALUATION PROTOCOLS

Several performance measurements such as accuracy, precision, recall and F1-score are also taken into account in the evaluation of performance for heart diseases prediction. The AUC–ROC allows to determine the ability of a model to separate diseased from non-diseased cases at various thresholds. The cross-validation is employed to avoid overfitting and the Wilcoxon signed-rank test is used for testing statistical significance of the results [1]. Models based on ECG apply external validation in independent cohorts to enhance reliability [5].

VII. COMPARATIVE ANALYSIS

Author(s)	Year	Disease Focus	Dataset	Method	Performance
Talukder et al. [1]	2025	Heart Disease	CHD, FHD, SHD	XAI-HD (ML + DL + SHAP + LIME)	Error reduced 20–25%
Demir et al. [2]	2025	Coronary Heart Disease	UCI & Framingham	ML & DL (SVM, CNN, LSTM)	SVM Accuracy = 92.42%
Ansari et al. [3]	2025	Heart Disease	UCI Cleveland	RF + LIME + PFI	Accuracy = 99%
Poterucha et al. [5]	2025	Structural Heart Disease	>1M ECG records	Deep Neural Network	High internal & external validation
Dhandapani et al. [10]	2025	Heart Disease	ECG Images	CNN Hybrid	Improved ECG feature learning

VIII. CHALLENGES AND OPEN ISSUES

Despite substantial progress in AI-based prediction of heart disease, a few key challenges still exist that constrain the broad clinical application. A fundamental issue is the lack of multi-center validation studies as the majority of models are created and validated on single-institution datasets making it impossible to generalize across populations. Dataset imbalance, and intrinsic bias are two other influencing factors which challenge the reliability of prediction in small

populations or the minority population. Also, we lack standardize benchmarking setup which make it difficult to fairly compare with this models. A timeless trade-off between explicability and predictive power also exists, since very complex deep learning models often behave as black box systems. Furthermore, regulatory issues, moral concerns and practical limitations of real world hospital information system interfacing are major barriers to clinical implementation. Addressing these challenges is critical for moving AI systems from being experimental research tools to reliable clinical decision-support tools.

IX. FUTURE RESEARCH DIRECTIONS

For future AI-based cardiovascular diagnostic research, there is a need to focus on the design of multimodal learning strategies, such as fusing ECG signals, imaging characteristics, genetic markers and valuable clinical structures data for increasing the robustness of prediction. There are privacy preserving methods, such as federated learning that can allow joint multi-institutional model training while keeping patient anonymity. Another interesting avenue is to design (lightweight, explainable) AI architecture that can be applied for real-time monitoring and edge deployment. Prospective validation study on a large scale in different population demographics is needed to ensure its clinical reliability and regulatory approval. In addition, fairness-sensitive modeling paradigms need to be integrated to reduce demographic bias and ensure fair access to healthcare services. Seamless connecting with wearable health monitoring systems and IoT landscapes might provide the possibility for better early diagnoses and continuous risk assessment. All these developments together will enhance the scalability, transparency and trustworthiness of AI-enabled cardiovascular health care systems.

IX. CONCLUSION

This survey reviewed AI-based heart disease diagnostic and predictive models, focusing on the development from traditional machine learning to deep learning and explainable hybrid networks. On the one hand, deep learning is found to have high detection power in ECG based method, and on the other hand ensemble methods of classifiers develop robustness on structured datasets. Explainable AI is an important medium that connects predictive power and clinical confidence. While we have made considerable achievements, issues such as generalizability, validation and interpretability remain to be solved for efficiently transforming AI from research settings to the clinic. Next-generation intelligent cardiovascular systems must balance the quadruple aim of accuracy, transparency,

scalability and ethical fitness to achieve substantive healthcare value.

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