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A Comprehensive Study of Machine Learning and Deep Learning for Heart Disease Prediction

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Abstract. Cardiovascular diseases are the main reasons for death around the world at present, so early detection and intervention can be difficult. Review of Recent Applications of Machine Learning and Deep Learning in Cardiovascular Disease Prediction. Logistic regression, decision trees, random forests, support vector machines and gradient boosting have all been applied to the Cleveland and Kaggle cardiovascular datasets in previous studies. Based on research results, soft voting and stacking ensemble methods have been used to improve the prediction accuracy of a single classifier. Dense neural networks and hybrid Convolutional Neural Network - Long Short-Term Memory (CNN-LSTM) architectures are also deep learning models that have been researched and applied. Add to the above that SHapley Additive exPlanations (SHAP) and Local Interpretable Model-agnostic Explanations (LIME) are popular methods for explaining Artificial Intelligence (AI) models. However, there are still many problems, such as an abundance of small public datasets, class imbalance, a lack of external validation, and poor clinical interpretability. The above are the problems of this paper, and some future directions for constructing a reliable cardiac disease prediction system with clinical applications are proposed.

Keywords: heart disease prediction, machine learning, deep learning, explainable AI, ensemble learning

1. Introduction

Diseases of the cardiovascular system continue to be a major source of mortality around the world and are posing an enormous challenge for the public health system. Diseases in the above category include a wide range of disorders of the structure and functioning of the heart like coronary heart disease and heart failure among others [1, 2]. Otherwise, many patients will not experience the severe symptoms of heart disease, such as heart attack, chest pain or cardiac arrest, and may be harder to detect [3]. Although the above diagnostic methods are clinically feasible, they often have the following deficiencies: high cost, invasiveness, extended operating time, and reliance on specialized knowledge [1]. Therefore, accurate and timely early and automated identification of cardiac risk has gradually been one of the focus areas of research in recent years to reduce mortality, support preventive treatment and improve clinical decisions [2, 3].

Increasing use of Electronic Health Records (EHRs) has contributed to the increased use of machine learning algorithms for cardiac risk stratification. Machine learning models can analyze

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clinical data on many diseases for various patients, such as standard clinical and demographic indicators routinely collected in cardiovascular workups, including resting blood pressure, serum cholesterol, fasting blood sugar, chest pain type, and Electrocardiogram (ECG) characteristics, etc., to assess their risks [4]. Logistic Regression, Decision Tree, Support Vector Machine, K Nearest Neighbour, Random Forest, Naive Bayes, Gradient Boosting and AdaBoost are some of the popular algorithms which have been widely used in the previous research [5, 6]. The above are relatively high-efficiency, low-cost predictive models for organised clinical data. However, their performance can be affected by data quality, sample size, class imbalance, feature selection, validation strategies, and other reasons [7, 8]. As a result, although a high-accuracy rate has been achieved on a single publicly available dataset, the actual performance of the model in clinical practice is not guaranteed.

In recent years, attempts have been made to increase prediction accuracy in the models through feature selection, model optimization, ensembles, and deep learning. Feature selection is employed in minimizing the number of features, which may not be relevant to the process, in order to optimize computations; however, the degree of minimization varies with algorithms [8]. Ensemble methods (e.g., soft voting and stacking) are employed to improve the stability and address the deficiencies of single-model classifiers by combining multiple base classifiers [5, 9]. Recently, as far as meta-ensembles and methods for handling class imbalance go, approaches like Synthetic Minority Oversampling Technique (SMOTE) and Edited Nearest Neighbors (ENN) have been suggested to enhance the generalization performance of classifiers [10]. Moreover, many deep learning approaches have been used to predict heart diseases by several researchers, including the fully connected neural network, artificial neural network, convolutional neural network, long short-term memory networks and hybrid deep neural networks [11, 12]. Such models can accommodate the non-linear nature of the data, but they typically demand a larger size of data set and proper optimisation to avoid overfitting.

Interpretability of medical artificial intelligence is required at the same time as prediction accuracy. Explainable AI tools like SHAP, LIME, QLattice, and Anchor have been used to explain why a model gave a certain output with respect to clinical factors [9, 10]. Therefore, it is necessary to provide reasons for the classifications in terms of health and life for more users, as well as to improve their understanding and support. However, the present studies on cardiovascular disease prediction by artificial intelligence still have considerable deficiencies. Many AI predictive models have not been independently validated by external parties, and therefore they carry a high risk of bias; thus, their clinical applications are limited [13]. Hence, in the future, more research should be carried out to ensure that there is improved accuracy for the model and also external validation and clinical applicability for the model.

In consequence, the proposed study aims at discovering the works conducted by researchers in relation to machine and deep learning for predicting heart diseases. The areas that include traditional machine learning classification techniques, feature selection, ensemble learning, deep learning models, and explainable artificial intelligence are considered. In addition, the paper introduces some problems in this area, such as dataset bias, class imbalance, model generalization, a lack of external validation, and issues of clinical interpretability; it also puts forward some future research directions for building trustworthy clinical decision-support systems.

2. Overview of heart disease prediction: significance, challenges, and methods

Heart disease prediction is a typical application of medical data analysis that can help identify people with a higher risk in advance to take early preventative measures against severe health problems and improve the results of treatment. Cardiovascular diseases have various causes, such as

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coronary artery disease, other diseases of the heart, heart failure, arrhythmias and others that damage the structure or function of the heart [1]. Early warning is relatively important because people may not have any obvious symptoms until it is close to them suffering from a serious heart disease, such as a heart attack, chest pain or heart failure [2]. However, despite the above-mentioned traditional approaches to diagnosis, including (ECG), blood testing, exercise tests, echocardiogram, and angiography of the coronary arteries, which have already been used in clinical practice, they tend to be costly, usually conducted under conditions where the patient is acutely ill, and interpreted by experts in the field [1]. Thus, with the advancement of data, many people are also willing to purchase early warning devices for heart diseases.

Machine learning will be appropriate to investigate the database obtained in many cases regarding certain attributes of that illness. Commonly considered factors include the patient's age, sex, type of chest pain, resting blood pressure, cholesterol level in the blood, fasting blood sugar, results of the resting electrocardiogram, heart rate, angina, and ST depression [3]. Above mentioned characteristics may be utilized by a supervised classification model in order to predict whether an individual suffers from heart diseases. There are several traditional machine learning algorithms such as logistic regression, decision tree, support vector machines, K-nearest neighbors, Random Forests, Naive Bayes, gradient boosting, AdaBoost, XGBoost, and Multilayer Perceptron (MLP) that are traditionally used in previous studies [4, 5, 14].

The Prediction of the disease is not accurate at present. Many reasons can affect model performance, such as poor data quality, a small sample size, missing values, duplicate records, class imbalance, insufficient feature selection, and other reasons [6, 7]. For instance, a class imbalance problem exists in medical databases; the positive and negative samples do not exist in equal numbers. Failure to solve this problem results in a model that overly favours the majority class [6]. To reduce computation cost and maintain the classification accuracy of the model, non-informative or redundant variables may also be discarded by feature selection [7, 14]. At the same time, some suitable indices for the evaluation of models in practice include accuracy, sensitivity, specificity, F1-score and Area Under the Curve (AUC) [14]. Therefore, cardiac disease prediction should not be judged by a single index of accuracy. A good model should be based on reliable data preparation, feature selection, class balancing, cross-validation, interpretability and clinical applicability.

3. Machine learning and deep learning approaches for heart disease prediction

3.1. Machine learning approaches and representative studies

The original model is a typical machine learning model; it is relatively simple and understandable, so it is suitable for the structure of the clinical data. Logistic regression is a simple classifier that can compute the probability of a disease occurring, and its model coefficients are relatively straightforward. Decision trees are used to classify patients in a rule-based manner, and the decision-making process is thus more understandable. Nevertheless, a single decision tree is susceptible to overfitting, particularly when trained on small-scale datasets. In order to overcome this drawback, Random Forests employ ensembles of more than one decision tree. Other methods employed include support vector machines, k-nearest neighbor, naive Bayes, gradient boosting, and AdaBoost/XGBoost algorithms [4, 5].

Bhatt et al. have applied several machine learning algorithms like decision trees, random forest, multi-layer perceptron, and XGBoost to a large dataset containing 70,000 cardiovascular diseases cases on Kaggle. Among the tested models, (MLP) with cross-validation scored the best accuracy of 87.28%, followed by XGBoost and Random Forest which performed similarly [4]. This work is

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particularly noteworthy given its use of a substantially larger dataset compared to typical UCI-based benchmarks, and its results suggest that model performance on larger datasets may be more realistic than the extremely high accuracy reported in small benchmark datasets.

The authors Chandrasekhar and Peddakrishna tested the following methods: Random Forest, K Nearest Neighbors, Logistic Regression, Naïve Bayes, Gradient Boosting, and AdaBoost on the Cleveland and IEEE Dataport Datasets using GridSearchCV and fivefold Cross Validation to tune hyperparameters. As for individual models, logistic regression scored an accuracy rate of 90.16% on the Cleveland dataset, whereas AdaBoost gained an accuracy rate of 90% on the IEEE Dataport dataset. An additional soft voting ensemble classifier, combining all six classifiers, increased the accuracy rates to 93.44% for Cleveland data set and 95% for IEEE Dataport [5] data set, confirming that ensemble classification methods could overcome weaknesses of individual classifiers.

Another problem is how to choose features for heart disease prediction. These researchers systematically reviewed 16 different feature selection techniques falling into three groups: filters, wrappers, and evolutionary algorithms. Their findings revealed mixed effects: while some models benefited from feature selection, others — particularly MLP and random forest — experienced performance degradation [7]. This inconsistency suggests that feature selection cannot be applied as a universal preprocessing step and requires model-specific consideration.

Class Imbalance is yet another challenge that arises in prediction problems. Researchers Ishaq et al. created nine Machine Learning (ML)-based models for predicting heart failure survival rates, which include algorithms like Decision Tree, AdaBoost, Logistic Regression, Stochastic Gradient Descent, Random Forests, Gradient Boosting, Extra Trees Classifier, Gaussian Naive Bayes, and Support Vector Machines. As a solution to class imbalance, SMOTE-based oversampling was used; the Extra Trees Classifier performed best among all models and had the highest accuracy rate of 0.9262 [6].

Recently, more research has been conducted to expand on previous work and explore ensemble learning and explainable artificial intelligence. Ganie and others trained fifteen base models with two cardiovascular disease datasets and built ensemble models through stacking and voting. In addition, they have done SHAP analysis on the predicted result of the model and used statistical testing methods like the Friedman aligned-ranks test and post-hoc pairwise comparison by Holm to determine the accuracy of the model [8]. Naz et al. suggested a meta-ensemble algorithm that uses a stacking method and utilizes the Random Forest, XGBoost, and LightGBM techniques. SMOTE-ENN is employed to deal with class imbalance and SHAP for model interpretation in their research framework; in addition, three datasets were used for this framework evaluation: Heart_2020_Cleaned, Cardio Train, and Heart Statlog Cleveland Hungary [9]. From the above studies, it can be seen that currently, the trend in research in this area is not to emphasize the precision of an individual model anymore, but to conduct more reliable, comprehensive, and multi-data-set assessments of models.

3.2. Deep learning approaches and representative studies

Because of the capability of deep learning algorithms to learn the complex, non-linear relationship between the medical data, this algorithm can also be used for predicting heart disease. The deep learning algorithms will be able to extract features from the data itself; in other words, they do not need manual extraction of any features. They outperform the traditional machine learning algorithms with large datasets. The researchers Almazroi et al. introduced a system for clinical decision support that makes use of dense neural networks based on the Keras library. For the experiments with this architecture, the number of hidden layers was varied between three and nine, and measures like

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accuracy, sensitivity, specificity, and F-score were calculated for heart disease datasets [3]. The deep learning algorithm can be implemented for developing a decision support system in order to predict heart disease automatically in this study.

There have also been studies conducted on hybrid deep learning systems. Al Reshan et al. created a robust heart disease prediction system which combines the techniques of artificial neural network (ANN), convolutional neural network (CNN), LSTM network, and the combined CNN-LSTM approach. This model was trained using the Cleveland heart disease database as well as a more extensive one and accuracy of 98.86% was obtained [11]. Hybrid Neural Networks have been introduced in this study to model the complicated connections among the heart disease data. Be cautious about applying CNNs and LSTMs to structured tabular data; these models are generally better suited for images, signals or sequences. As such, the performance of this application in practical applications will depend on the size of the data set, data type, validation process, and the explainability of the model used.

Explainable Artificial Intelligence (XAI) has become necessary for further research in machine learning and deep learning applications. Santhosh et al. proposed a new method called CARDIACX [10], and many machine learning approaches were used alongside different explainable AI techniques like SHAP, LIME, QLattice, and Anchor. All of the above techniques can be used to know why the model reached this conclusion. Therefore, it is necessary to provide both the predicted labels and reasons that make sense to the clinician in order to gain their trust. Deep learning has good model construction capabilities, but it is not feasible to apply in heart disease prediction without repeated experiments on various datasets and meaningful interpretations by clinicians.

4. Limitations and future directions

Despite the promising contributions made by machine learning and deep learning towards cardiac disease predictions, there are still some shortcomings associated with them. First, a number of studies use publicly available data sets including those from Cleveland, Statlog, Kaggle, and many others for comparative purposes, which might not be able to represent diverse patient populations as compared to the real-world scenarios. Consequently, models that perform exceptionally well on public datasets may not necessarily generalize to different hospitals, populations, or clinical settings [1, 12]. Second, the extremely high accuracy rates reported do not necessarily prove the reliability of these models in clinical applications. Without external validation, prospective testing, and transparent reporting mechanisms, the actual performance of these models in the real world remains highly uncertain.

A systematic review by Cai et al. found that AI-based predictive models for cardiovascular disease remain in the early stages of development. The review screened 486 AI models extracted from 79 studies; however, none of these models had undergone independent external validation. The review further noted that, according to Prediction model Risk Of Bias ASsessment Tool (PROBAST) criteria, all models presented a high risk of bias—primarily due to deficiencies in study design, statistical methodology, and reporting quality [13]. This finding suggests that future research should not be limited merely to proposing novel algorithms, but should instead place greater emphasis on reproducibility, external validation, and clinical applicability.

Explainability is another significant challenge. While ensemble models and deep learning models can improve predictive accuracy, they are often more complex than logistic regression or decision tree models. Despite the fact that (XAI) techniques like SHAP, LIME, QLattice, and Anchor can assist in improving model transparency, it is important that these explanations undergo evaluation

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and validation based on medical knowledge [8, 10]. Therefore, future research should focus on integrating model predictive performance with clinical reasoning processes. Naser et al. note that future studies should also explore how to integrate general artificial intelligence (AGI), federated learning, and IoT-based data collection technologies to overcome the inherent limitations of current small-scale datasets and build more generalizable, comprehensive cardiovascular disease prediction systems [14]. Moreover, further studies need to use a greater number of clinical datasets, including electronic medical records, (ECG) signals, data from wearable sensors, and data from medical imaging modalities. In the end, a dependable cardiovascular disease prediction model will be one that is characterized by accuracy, interpretability, externally verified reliability, and effective clinical decision support capability.

5. Conclusion

Recent advances in predictive modeling for cardiovascular disease using machine learning and deep learning techniques were reviewed in this paper. This paper discusses traditional machine learning techniques, ensemble learning approaches, and deep learning algorithms used in this field.

Based on the mentioned experimental findings, it can be seen that Logistic Regression, Random Forest, and Gradient Boosting were the most widely applied ML algorithms, and by using hyperparameter tuning techniques and ensembles, these ML algorithms gave good results. Soft voting and stacking ensemble methods are generally better than single classifiers. Although some deep learning models for structured tabular cardiac data, such as fully connected neural networks and hybrid CNN-LSTM architectures, have been proposed by scholars, their specific advantages over traditional methods have not yet been fully demonstrated. Moreover, explainable artificial intelligence (XAI) methods like SHAP and LIME are slowly becoming adopted in clinical practice as a means of increasing transparency and trust in models. Beyond accuracy, sensitivity and specificity deserve more attention, since missing an at-risk patient carries far higher clinical costs than a false alarm.

There are still some problems at present. Most of the existing research uses small-scale, publicly available benchmark datasets and lacks external validation; thus, it is not very generalisable. Other problems include class imbalance, overfitting, and poor clinical interpretability. In the future, real-world clinical datasets will be used for external validation, multi-modal data will be integrated, and an interpretable model suitable for practice will be developed. Only by solving the above problems can research results from machine learning be reliably applied in the clinical setting.

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