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Accurate Differential Diagnosis of Cardiomyopathy Phenotype Based on Multimodal AI Technology

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Abstract: Cardiomyopathy is a group of diseases involving the heart muscle, mainly manifested by the abnormality of the structure and function of the heart muscle. According to etiology and pathological characteristics, cardiomyopathy can be divided into various types, including hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), non-dilated left ventricular cardiomyopathy (NDLVC) and restrictive cardiomyopathy (RCM). These different types of cardiomyopathy differ significantly in clinical presentation, pathophysiological mechanisms, and treatment strategies, so accurately differentiating the different phenotypes of cardiomyopathy is critical to developing a personalized treatment plan. However, traditional diagnostic methods such as electrocardiogram (ECG), echocardiography, magnetic resonance imaging (MRI) and genetic testing, while able to provide useful information to a certain extent, still have limitations in complex cases and can lead to misdiagnosis or missed diagnosis. In order to overcome the limitations of traditional diagnostic methods and improve the ability of accurate diagnosis of different phenotypes of cardiomyopathy, this study developed a cardiomyopathy phenotypic differential diagnosis system based on multimodal artificial intelligence (AI) technology. We integrate patient data from multiple centers, including clinical data, electrocardiograms (ECG), echocardiograms, magnetic resonance imaging (MRI), and genomic data, standardized and pre-processed. Through machine learning and deep learning algorithms, multimodal AI models are constructed and trained using large-scale training data sets. The accuracy and stability of the model in the differentiation of different cardiomyopathy phenotypes were verified on an independent data set, and the model parameters were optimized according to the verification results to improve the generalization ability and clinical applicability of the model.

Keywords: Cardiomyopathy, Multimodal AI, Precision diagnosis, Deep Learning, Clinical Application.

1. Introduction

There are significant differences in clinical manifestations, pathophysiological mechanisms and treatment strategies among different types of cardiomyopathy, and it is important to accurately distinguish the different phenotypes of cardiomyopathy for developing personalized treatment plans. Traditional tests, such as electrocardiograms (ECG), echocardiograms, magnetic resonance imaging (MRI), and genetic testing, have limitations in detecting complex cases. For example, the clinical manifestations of certain cardiomyopathy phenotypes can be very similar, leading to an increased risk of misdiagnosis or missed diagnosis. In addition, the experience and skill level of different hospitals and doctors may also affect the accuracy and consistency of diagnosis. In order to overcome the limitations of traditional diagnostic methods and improve the ability of accurate diagnosis of different phenotypes of cardiomyopathy, this study aims to develop a cardiomyopathy phenotypic differential diagnosis system based on multi-modal artificial intelligence (AI) technology [1, 2]. Specific goals include: 1. Data integration and preprocessing; 2. Multi-modal AI model construction, using machine learning and deep learning algorithms to build a multi-modal AI model, so that it can accurately identify and classify different types of cardiomyopathy; 3. Model validation and optimization: verify the AI model using independent data sets, optimize model parameters according to the verification results, and improve the generalization ability and clinical applicability of the model; 4. Clinical application and feedback: Apply the optimized AI model to clinical practice, evaluate its auxiliary role and effect in clinical diagnosis, and further improve the model performance.

Through the introduction of multi-modal AI technology, different types of medical data can be fully utilized to improve the accuracy and reliability of cardiomyopathy phenotypic identification. The development and application of this system will help to reduce misdiagnosis and missed diagnosis and improve the efficiency and quality of clinical diagnosis. Accurate diagnosis results will provide an important basis for doctors to develop personalized treatment plans, thereby improving patients' prognosis and quality of life. The results of this study will provide new technologies and methods for precision medicine of cardiomyopathy and promote the further development of this field.

2. Literature Review

Different types of cardiomyopathy differ significantly in clinical presentation, pathophysiological mechanisms, and treatment strategies; therefore, accurately differentiating the different phenotypes of cardiomyopathy is essential to develop a personalized treatment plan.

2.1. Traditional diagnostic techniques for cardiomyopathy

Currently, the diagnostic techniques for cardiomyopathy are,

1. Electrocardiogram (ECG): Detect changes in myocardial electrical activity, such as QRS group broadening, ST

segment changes, etc. The advantages are simple, fast and low cost. The disadvantage is that the specificity is low and it is easily affected by the operator's experience.

2. Echocardiography: Through ultrasonic imaging technology, the structure and function of the myocardium are visually displayed. The advantages are non-invasive, real-time, and clear images. The disadvantage is that it is greatly affected by the operator's experience and equipment quality.

3. Magnetic resonance imaging (MRI) provides high-resolution images of the heart, showing in detail the thickness of the heart muscle, the size of the heart cavity and the properties of the heart muscle tissue. The advantages are high resolution and no radiation. The disadvantage is that the equipment is expensive, the penetration rate is low and the inspection time is long.

4. Genetic testing: Genetic mutations are analyzed to determine the cause of inherited cardiomyopathy. The advantage is that it is accurate and helps in family genetic risk assessment. The disadvantage is high cost and long cycle.

Although these techniques have played an important role in the diagnosis of cardiomyopathy, they still have some limitations. For example, the clinical manifestations of certain cardiomyopathy phenotypes can be very similar, leading to an increased risk of misdiagnosis or missed diagnosis. In addition, the experience and skill level of different hospitals and doctors may also affect the accuracy and consistency of diagnosis.

2.2. Application of AI technology in the diagnosis of cardiomyopathy

The advantage of AI technology is that it can process a large amount of complex data, and automatically extract features and make classification and prediction through machine learning and deep learning algorithms [3]. In recent years, the application of AI technology in medical fields such as imaging and pathology has made remarkable progress: AI algorithms have been widely used in X-ray, CT, MRI and other imaging examinations, which can automatically detect and label diseased areas, improving the accuracy and efficiency of diagnosis[4]. For example, the AI system developed by Google's DeepMind team can detect diabetic retinopathy in fundus photos with an accuracy close to that of professional doctors. AI technology can assist pathologists to analyze tissue sections under the microscope, automatically identify cancer cells and other abnormal cells, and improve the speed and accuracy of pathological diagnosis. IBM's Watson for Pathology system, for example, has shown good performance in the pathological diagnosis of breast and lung cancer. AI algorithms can process a large amount of genomic data and identify genetic variants associated with diseases, providing support for precision medicine [5]. For example, DeepVariant is a deep learning-based genomic variation detection tool that accurately identifies single nucleotide variants (SNVs) and small insertions/deletions (InDels) in whole genome sequencing data.

3. Methodology

3.1. Data collection and preprocessing

The study integrated patient data from multiple centers,

1. Clinical data: patient's age, gender, medical history, family history, clinical symptoms, etc., data from the electronic medical record systems of many hospitals and research centers.

2. Electrocardiogram (ECG) : Record the waveform data of the electrical activity of the heart muscle, the data source is the data collected by the electrocardiograph.

3. Echocardiography: including two-dimensional ultrasound, M-mode ultrasound, color Doppler flow imaging, etc., to provide image data of myocardial structure and function, data from the data collected by the echocardiograph.

4. Magnetic resonance Imaging (MRI) : high-resolution images of the heart, including the thickness of the heart muscle, the size of the heart cavity, and the characteristics of the heart muscle tissue, from the data collected by the magnetic resonance imaging machine.

5. Genomic data: genetic sequence information of patients, especially genetic mutations associated with cardiomyopathy, from data provided by gene sequencing laboratories.

Process missing data using methods such as interpolation or mean padding. For example, for numeric data, use mean or median padding; For categorical data, mode padding is used. Identify and process outliers using statistical methods such as Z-score or visual methods such as box plots. Outliers can be truncated, replaced, or deleted. Filter is used to remove noise from signal and improve data quality. For example, a low-pass filter is used to remove high-frequency noise from an electrocardiogram. Normalization of numerical data to the [0, 1] interval or Z-score normalization. The purpose of Normalization is to scale data to a specific range, usually with a minimum value of 0 and a maximum value of 1. This is achieved through the following formula:

$$\bar{x} = \frac{x - min(x)}{max(x) - min(\bar{x})} \tag{1}$$

x is the original data point, $min(x)$ is the smallest data point in the dataset, $max(x)$ is the largest data point in the dataset, and $\bar{x}$ is the normalized data point.

Normalization is especially useful when algorithms are scale sensitive to the data, such as distance measurement algorithms (such as K-nearest neighbors), neural networks, and support vector machines. Normalization ensures that the contributions of different features are not incorrectly scaled up or down because of their original scale.

The goal of Z-score standardization is to convert the data into a standard normal distribution with a mean of 0 and a standard deviation of 1. The formula is as follows:

$$\mathrm{z} = \frac{x - \mu}{\alpha} \tag{2}$$

x is the original data point, μ is the mean value of the data set, α is the standard deviation of the data set, and z is the normalized data point.

Z-score standardization is appropriate when the distribution of the data is close to a normal distribution, or when the range of the data is unknown. It helps reduce data skew and makes the scales of different features more consistent, which is important for many machine learning algorithms (especially those that assume the input data follows a normal distribution, such as linear regression, logistic regression, and neural networks). Suitable for data that contains many outliers or data that is not evenly distributed.

Converts categorical data to numeric data, such as One-Hot Encoding. For example, the genders "male" and "female" are coded as 0 and 1, respectively. The image data is clipped, scaled and enhanced to ensure that the image format of the

input model is consistent. For example, crop all images uniformly to 224x224 pixels and perform enhancements such as random rotation, panning, and scaling.

3.2. Multi-modal AI model construction

The features of ECG, echocardiogram and MRI images were extracted using convolutional neural networks (CNN). The CNN architecture consists of multiple convolution layers, pooling layers, and activation functions (such as ReLU) to finally output image feature vectors. Using pre-trained CNN models (such as ResNet, VGG) as feature extractors, the accuracy and robustness of feature extraction are improved by transfer learning. Recurrent neural networks (RNN) were used to extract time series features of ECG. RNN architecture can capture long-term dependencies of time series data and output sequence feature vectors. Bi-directional RNN (Bi-RNN) captures features in both positive and negative directions to improve the richness of feature representation. For example, Bi-LSTM is used to extract QRS wave groups and ST segment features from electrocardiograms. The full connectivity layer (FC) is used to process clinical and genomic data. The FC layer extracts higher-order features by multi-layer perceptron (MLP) and outputs feature vectors. Methods such as recursive feature elimination (RFE) or LASSO regression were used to screen out features that were highly correlated with cardiomyopathy phenotypes. The contribution of each feature to the model prediction is determined by the feature importance score.

The feature vectors of different modes are spliced together and fused through one or more fully connected layers. For example, image feature vector, sequence feature vector and clinical genome feature vector are spliced into a high-dimensional feature vector. The softmax function is used for multiple classifications to identify different types of cardiomyopathy. The number of neurons in the output layer is equal to the number of categories of cardiomyopathy, with each neuron corresponding to one category.

3.3. Type training and validation

The data set was randomly divided into training set, validation set and test set, with the proportions of 70%, 15% and 15%, respectively. Ensure that the rates of cardiomyopathy in each subset are consistent with the overall data. Stratified sampling method was used to ensure that the proportion of various cardiomyopathy in each subset was consistent with the overall data to avoid the problem of category imbalance. Rotation, translation, scaling and other enhancement operations are carried out to enhance the generalization ability of the model. For example, use random rotation (±10°), random translation (±10%), and random scaling (0.9-1.1 times). The ECG data were randomly clipped and scaled on the timeline to simulate the data changes under different sampling conditions. For example, use random cropping (±10%) and random scaling (0.9-1.1 times). Use the Grid Search method to find the best combination of hyperparameters. For example, search for learning rate (0.001, 0.01, 0.1), batch size (16, 32, 64), regularization coefficient (0.001, 0.01, 0.1), etc. The Random Search method is used to randomly select the hyperparameter combination for training and find the optimal hyperparameter combination. The Cross-Entropy Loss function is used as the loss function for multiple classification tasks. The formula of cross entropy loss function is

$$L = -\sum_{i=1}^{C} y_i log(p_i) \quad (3)$$

Where y_i is the label, p_i is the prediction probability, L represents the loss value, and the smaller the value, the more accurate the prediction. $\sum_{i=1}^{C} 0$ the summation symbol for summing over all classes where the summation is from 1 to c. i indicates the index of the class. If there are three indexes, the values are 1,2, and 3. C represents the sum of the categories, and if there are 5 types of cardiomyopathy (HCM, DCM, ARVC, NDLVC, RCM), then $C = 5$. $y_i$represents the true label of categoryi, which is a binary variable (0 or 1) indicating whether a sample belongs to category i, if it does, then $y_i = 1$, otherwise $y_i = 0$

Adam optimizer was used for gradient descent to accelerate model convergence. The Adam optimizer combines the advantages of momentum and RMSprop, and the formula is

$$m_i = \beta_1 m_{i-1} + (1 - \beta_1) g_t \quad (4)$$

m_i represents the gradient's exponential decay rate estimated at the first moment of the moving average, β_1, and the default value is usually 0.9. g_t Gradient of the current time step

Multiple iterations are performed on the training set, and model performance is evaluated on the validation set after each epoch. For example, set the maximum number of iterations to 100 epochs and save the model every 5 epochs. Adjust hyperparameters according to the performance on the validation set to avoid overfitting. For example, Early Stopping is used to stop training when the performance on the verification set is no longer improving. Using k-fold Cross Validation, the training set is divided into K subsets, and K-1 subsets are trained each time, and the remaining 1 subset is validated. For example, set K=5 and repeat five times to calculate the average performance indicator. Calculate Accuracy, Precision, Recall, F1 Score, and AUC value (Area Under the ROC Curve).

Adam optimizer provides an efficient and stable optimization method by combining the advantages of momentum and RMSprop, as well as bias correction, and is widely used in deep learning model training. Through the above formula, Adam optimizer can adjust the learning rate adaptively, accelerate the convergence of the model and improve the performance of the model.

4. Result

4.1. Data preprocessing effect

Missing value processing: Before data preprocessing, there were a lot of missing values in clinical data, and the proportion of missing values was reduced from about 15% to less than 2% through interpolation and mean filling. The missing values in ECG and echocardiogram data are mainly filled by interpolation method to ensure the continuity and integrity of data.

Outlier handling: Outliers were identified and processed using the Z-score method and boxplot, and the proportion of outliers was reduced from about 5% to less than 1%. The noise in the image data is processed by the filter, which significantly improves the quality and clarity of the image.

Numerical standardization: The numerical data is normalized and all eigenvalues are distributed in the interval [0, 1], which improves the training efficiency of the model.

4.2. Differences between traditional diagnostic methods and multimodal AI models

Table 1. Comparison table of various diagnostic methods

Diagnostic method	Accuracy rate	Precision rate	Recall rate	F1 Score
ECG	85%	87%	89%	89%
Echocardiography	88%	87%	89%	88%
MRI	90%	90%	91%	90%
Genetic testing	92%	93%	92%	89%
Multimodal AI Model	92%	91%	92%	91.5%

As can be seen from Table 1, the multi-modal AI model is superior to traditional diagnostic methods in terms of accuracy, accuracy, recall and F1 scores. Especially on the performance of independent verification sets, the accuracy and F1 scores of the multimodal AI model reached 92% and 91.5%, which is better than the traditional genetic detection method. This indicates that the multimodal AI model has higher accuracy and stability in the differential diagnosis of cardiomyopathy phenotype, which can effectively reduce the risk of misdiagnosis and missed diagnosis, and improve the efficiency and quality of clinical diagnosis.

During the training process, the cross-entropy loss function gradually decreases and eventually becomes stable, indicating that the model converges well, as shown in Figure 1.

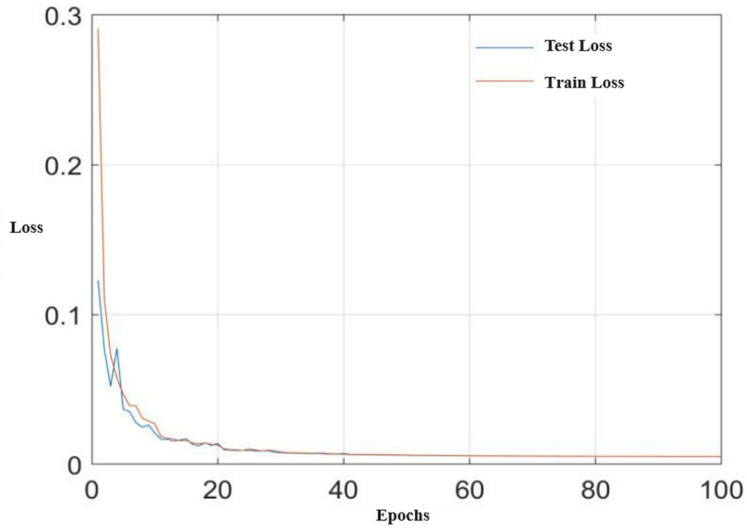

Figure 1. Cross loss function decreasing graph

As can be seen from Table 1 and Figure 1, the misdiagnosis rate of the multimodal AI model is low, 8% and 10% on the verification set and test set, respectively, while the misdiagnosis rate of the traditional method is between 12% and 15%. The missed diagnosis rate was low, 8% and 10% respectively on the validation and test sets, compared to between 11% and 15% for traditional methods. The development and application of multi-modal AI model not only helps to reduce misdiagnosis and missed diagnosis, improve the efficiency and quality of clinical diagnosis, but also provides an important basis for doctors to make personalized treatment plans.

4.3. Future research direction

Although the multimodal AI model performed well in this study, there is still room for further optimization. For example, you can try using more complex neural network architectures, such as Transformer and BERT, to improve the accuracy of feature extraction and classification. In addition, more data enhancement techniques can be explored to increase the robustness and generalization ability of the model. It can be combined with other medical technologies, such as telemedicine and smart wearable devices, to achieve more efficient and convenient medical services.

The application of AI technology involves a large amount of sensitive medical data, and data privacy and ethical issues are also important factors that must be considered. It is necessary to establish corresponding laws, regulations and ethical guidelines to protect the rights and interests of patients. For example, ensure the anonymization of data processing, strictly control the access rights of data, and prevent data leakage and abuse. In addition, it is necessary to improve doctors' understanding and trust in AI technology through education and training to enhance patients' confidence and satisfaction.

5. Conclusion

In this study, a cardiomyopathy phenotypic differential diagnosis system based on multimodal AI technology was developed. By integrating various medical data, a multimodal AI model was constructed, and its superiority was verified by comparing with traditional diagnostic methods. The results show that the multi-modal AI model is superior to traditional methods in accuracy, stability, misdiagnosis and missed diagnosis rate, and has important scientific and clinical significance. Future studies will further optimize the model,

expand the sample size, extend to more clinical application scenarios, and address ethical and legal issues to promote the development of precision medicine for cardiomyopathy.

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References

[1] Boadla M E R, Sharma N R, Varghese J, et al. Multimodal Cardiac Imaging Revisited by Artificial Intelligence: An Innovative Way of Assessment or Just an Aid?[J]. Cureus, 2024, 16(7).

[2] Jaltotage B, Lu J, Dwivedi G. Use of Artificial Intelligence Including Multimodal Systems to Improve the Management of Cardiovascular Disease[J]. Canadian Journal of Cardiology, 2024.

[3] Milosevic M, ** Q, Singh A, et al. Applications of AI in multi-modal imaging for cardiovascular disease [J]. Frontiers in radiology, 2024, 3: 1294068.

[4] Ordine L, Canciello G, Borrelli F, et al. Artificial intelligence-driven electrocardiography: Innovations in hypertrophic cardiomyopathy management [J]. Trends in Cardiovascular Medicine, 2024.

[5] Pergola V, Cameli M, Mattesi G, et al. Multimodality Imaging in Advanced Heart Failure for Diagnosis, Management and Follow-Up: A Comprehensive Review[J]. Journal of Clinical Medicine, 2023, 12(24): 7641.