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Comprehensive Deep Learning Framework for Disease Prediction

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Abstract: This study introduces an innovative hybrid deep learning framework designed to improve the precision of disease prediction by utilizing temporal data from Electronic Health Records (EHRs). The framework combines Convolutional Neural Networks (CNNs) with Long Short-Term Memory (LSTM) networks, harnessing CNNs' ability to extract hierarchical feature representations from intricate data and LSTMs' strength in capturing long-term dependencies in temporal information. Empirical analysis using real-world EHR datasets demonstrated that this hybrid network significantly outperforms Support Vector Machine (SVM) models, as well as standalone CNNs and LSTMs, in disease prediction tasks. This research not only enhances predictive model performance in health data analytics but also highlights the critical role of advanced deep learning technologies in managing the complexity of contemporary medical data. Our results suggest a paradigm shift towards integrating sophisticated neural network architectures in predictive model development, potentially paving the way for more personalized and proactive disease management and care, thus setting new benchmarks for future health management practices.

Keywords: Deep Learning, Convolutional Neural Network, Long Short Term Memory Neural Network, Hybrid Deep Learning.

1. Introduction

Electronic Health Records (EHRs) data, with its detailed record of patient health over time, plays a pivotal role in understanding disease progression and developing models to forecast future health trajectories. However, the nature of EHR data collection—sporadic, based on patient visits or specific healthcare encounters—poses unique challenges for model development. These challenges include handling diverse data formats and navigating the intricacies of longitudinal patient data[1]. Efforts to overcome these obstacles have taken two main paths: leveraging domain-specific knowledge to create unified patient groups from disparate data sources, and investigating the integration of varied EHR[2] data types, either pre- or post-model development.

Traditional approaches to disease forecasting often categorize patients with similar health patterns together, simplifying the prediction process. However, predicting outcomes based on a single variable remains a daunting task in machine learning[3], particularly when key variables are unknown. Such univariate predictions, though challenging, are adaptable for forecasting various diseases using historical EHR data, without needing additional contextual information.

The application of deep learning neural networks (DLNNs), including in areas like natural language processing[4][5] and image recognition[6][7], has seen a significant rise. Among these, Long Short-Term Memory (LSTM) networks[8] stand out for their superior predictive accuracy, attributed to their ability to retain information over extended periods through specialized memory gates. This has placed LSTMs ahead of many traditional and machine learning methods in prediction accuracy. Moreover, LSTM networks, a variant of Recurrent Neural Networks (RNNs)[9], alongside other DLNN forms such as Convolutional Neural Networks (CNNs) and Deep Belief Networks (DBN), have shown promise in various prediction tasks. Temporal CNNs[10][11], with their specialized convolution operations, are particularly noted for their efficacy in time series forecasting.

2. Related work

Reviewing related literature, disease forecasting emerges as a field of critical importance in medical diagnostics[12][13]. Traditional prediction models like Support Vector Regression and various time series methodologies[14] have been explored alongside deep learning techniques, which offer enhanced data processing capabilities and accuracy. Deep learning models, with their extensive internal layers, tackle more complex problems than their Artificial Neural Network counterparts[15]. Recent studies have demonstrated the effectiveness of deep learning models in medical diagnostics, disease trend forecasting, and even in identifying specific health conditions, outperforming traditional methods[16][17]. This research proposes a novel deep learning framework that combines LSTM networks with CNNs to tackle the challenges of disease prediction. By integrating a CNN preprocessing step, this framework aims to refine raw data into multidimensional inputs, thereby boosting LSTM's predictive performance. The effectiveness of this hybrid model was validated using real-world EHR datasets, showing its superiority over conventional prediction methods, including SVM, standalone CNN, and LSTM models. The key contributions of this study are two-fold: the introduction of a CNN preprocessing step for enhancing data dimensionality and the development of a comprehensive deep learning model for disease forecasting, demonstrating significant advancements over existing approaches.

3. Methodology

Long Short-Term Memory (LSTM) networks and Convolutional Neural Networks (CNNs) stand out as two influential subsets of deep learning techniques that have garnered significant interest globally in recent times[18]. In our study, we introduce a combined approach of LSTM and CNN to address the challenges posed by irregular patterns and long-term dependencies in temporal data forecasting tasks. This hybrid model surpasses traditional methods in delivering predictions that are both more precise and dependable[19][20].

In our methodology, we utilize real-world data, initiating the process by applying CNN for data preprocessing. The CNN-processed data is then fed into the LSTM network for training. This approach leverages the strengths of both networks: CNNs effectively capture spatial hierarchies in data, while LSTMs excel in managing temporal dependencies, resulting in a robust model that significantly enhances forecasting accuracy.

3.1. Data Description

The dataset used in our experiments was processed using Adadelta [21]. After preprocessing, it comprised 578 instances, with 361 classified as positive and 217 as negative. We partitioned the data into training, validation, and testing segments, following an 80:10:10 distribution ratio. The training segment was utilized to train our Deep Learning Neural Network (DLNN) framework, while the validation segment, an independent collection of samples, served to tune hyperparameters and provide an initial evaluation of the framework's performance. The testing segment was designated for assessing the trained model's generalization ability.

3.2. Long Short-Term Memory-based Recurrent Neural Network

LSTM models, a specialized category within Recurrent Neural Networks (RNNs), incorporate feedback connections in each neuron. Unlike traditional neural networks, an RNN's output is influenced not only by the inputs and weights of the current neuron but also by inputs from preceding neurons, rendering it particularly well-suited for analyzing sequential data. However, RNNs encounter significant challenges, such as gradient explosion and vanishing gradients, when processing long-term sequential information[22]. These challenges led to the development of LSTMs[23], which address these issues by incorporating memory cells and specialized gating mechanisms to maintain information over extended sequences, thereby enhancing the model's ability to learn long-term dependencies.

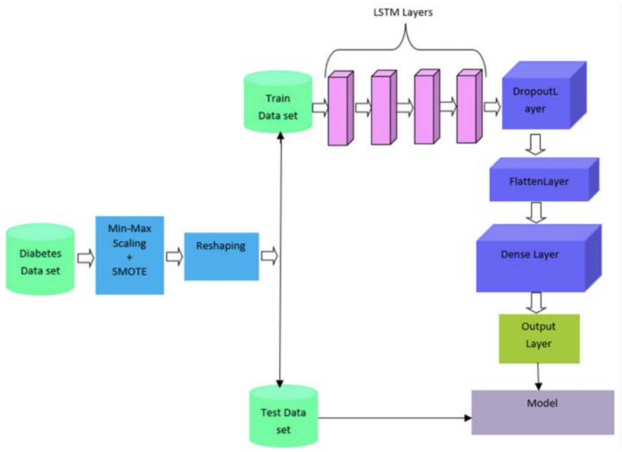

Figure 1. The training process of LSTM model

LSTMs are designed to mitigate the vanishing gradient problem encountered by RNNs, incorporating mechanisms to selectively retain or discard information. The architecture of an LSTM includes a cell state and three gates: input, forget, and output (Figure 1), which regulate the cell state's update, retention, and elimination of information. The process of forward computation in an LSTM is characterized by these components.

$$f_t = \sigma(W_{fh} \cdot h_{t-1} + W_{fx} \cdot x_t + b_f)(($$

$$it = \sigma(Wih \cdot ht-1 + Wix \cdot xt + bi)$$

$$\tilde{C} = \tanh(Wch \cdot ht-1 + Wcx \cdot xt + bc)$$

$$C_t = f_t \cdot C_{t-1} + i_t \cdot \tilde{C}$$

$$ot = \sigma(Wot \cdot ht-1 + Wox \cdot xt + bo)$$

$$ht = ot \cdot \tanh(Ct)$$

Where C_t ,C_{t-1}respectively represent the current cell state value, the cell state value at the previous moment, and the update to the current cell state value. The symbols f_t , i_t, and o_t respectively denote the forget gate, input gate, and output gate. With appropriate parameter settings, based on the values of C_t , the output value h_t is calculated according to equations (4) to (6). Based on the difference between the output values and the actual values, all weight matrices are updated through the Back-Propagation Through Time (BPTT) algorithm [24].

3.3. Temporal Convolutional Neural Networks

Convolutional Neural Networks (CNNs) are among the most commonly utilized deep learning neural networks, primarily applied in computer vision for image recognition and classification tasks. For large volumes of raw data samples, CNNs can efficiently extract a useful subset of the input data. Generally, CNNs are feedforward neural networks, derived from multi-layer neural networks (MLNNs). The main distinction between CNNs and traditional MLNNs lies in CNNs' characteristics of sparse interactions and parameter sharing[25].

Traditional MLNNs employ a fully connected strategy to establish neural networks between input and output layers, meaning each output neuron interacts with every input neuron. Assuming there are m input neurons and n output neurons, the weight matrix would have m × n parameters. CNNs significantly reduce the number of parameters in the weight matrix by utilizing convolutional kernels of size k × k. Two attributes of CNNs enhance the training efficiency of parameter optimization: with the same computational complexity, CNNs can train neural networks with more hidden layers, i.e., deeper neural networks.

Temporal Convolutional Neural Networks (Temporal CNNs) introduce specialized one-dimensional convolutions suited for processing univariate time series data. Unlike traditional CNNs that use k × k convolutional kernels, temporal CNNs employ kernels of size k × 1 . After temporal convolution operations, the original univariate dataset can be expanded into a dataset with m -dimensional features. Thus, temporal CNNs apply one-dimensional convolution to time series data, expanding the univariate dataset into a multi-dimensional feature-extracted dataset (the first stage in Figure 2). The expanded multi-dimensional feature data are more suitable for prediction using LSTM.

3.4. CNN-LSTM Prediction Framework

To address the challenges of sequence dependency and univariate data, this paper proposes a hybrid deep neural network (DNN) that combines CNN and LSTM models. The structure of the hybrid DNN framework is shown in Figure 2. In the preprocessing stage, the CNN extracts crucial information from the input data, using convolution to reorganize univariate input data into multidimensional feature data (Figure 2). In the second stage, the reorganized feature data are input into LSTM units for prediction. This approach leverages the strengths of CNNs in feature extraction and LSTMs in handling sequence dependencies, resulting in a robust model capable of accurate and efficient prediction.

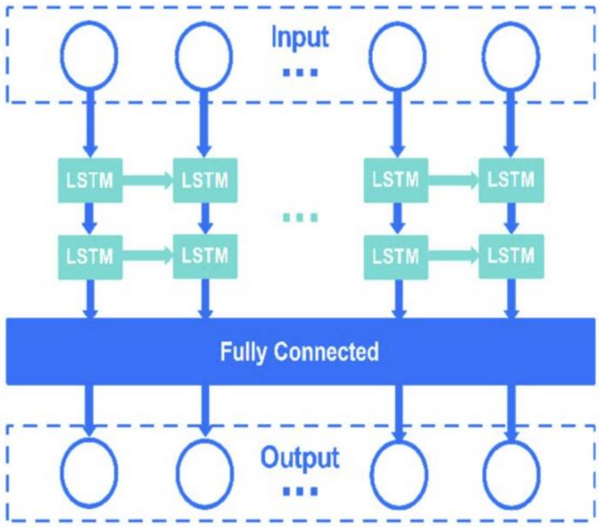

Figure 2. The training process of LSTM model

As illustrated in Figure 2, the input dataset undergoes preprocessing via a Convolutional Neural Network (CNN) composed of two hidden layers. It is worth mentioning that traditional temporal CNNs typically incorporate pooling operations when the hidden layers exceed five, aiming to mitigate overfitting. However, this study intentionally omits pooling operations to enhance the retention of extracted feature information.Subsequent to the preprocessing phase, a Long Short-Term Memory (LSTM) neural network is constructed for the purposes of training and disease prediction. The training workflow of the LSTM architecture is depicted in Figure 1, where features derived from the initial stage serve as the input for the LSTM model's training. To mitigate overfitting, a dropout layer is incorporated into the LSTM network. The discrepancy between predicted and actual output values, referred to as the loss, is employed to adjust the weights of all LSTM units. The optimization leverages the RMSprop algorithm, a gradient descent optimization method frequently utilized for weight tuning in deep neural networks [26].

4. Experimental results

The proposed hybrid Deep Neural Network (DNN) framework was developed using Python 3.7.3 (64-bit). It leverages Google's open-source deep learning library TensorFlow, with Keras version 2.3.1 serving as the frontend interface.

The prediction outcomes of the proposed CNN-LSTM model were benchmarked against existing methodologies, including Support Vector Machine (SVM) models, standalone CNNs, and standalone LSTMs. Performance evaluation metrics used in this study encompass accuracy, the F1 score, and the Area Under the ROC Curve (AUC). Accuracy measures the ratio of correctly predicted samples to the total number of samples. The F1 score offers a harmonic mean between precision and recall, where precision assesses the proportion of true positive predictions among all positive predictions, and recall evaluates the proportion of true positive predictions among all actual positive samples. The AUC is another critical metric, representing the trade-off between the false positive rate (the proportion of negative samples incorrectly classified as positive) and the true positive rate (the proportion of positive samples correctly identified). A higher AUC value signifies better model performance.

In this study, four prediction models were constructed and their performance metrics are summarized in Table 1. The results indicate that the choice of model significantly influences prediction performance. The CNN-LSTM algorithm demonstrated superior performance, with an increase in AUC by 6.5%, F1 score by 12.2%, and accuracy by 14.6% compared to the standalone LSTM model. These experimental outcomes underscore the effectiveness of the hybrid deep learning algorithm in predicting temporal data related to disease.

Table 1. Forecast results of different models

METHOD	CNN	SVM	LSTM	CNN&LSTM
F1	0.51998	0.49182	0.65216	0.74284
AUC	0.63234	0.5786	0.76616	0.819843
Accuracy	0.57893	0.4561	0.71928	0.842194

5. Conclusion

This research successfully engineered an innovative deep learning architecture by merging Convolutional Neural Networks (CNN) with Long Short-Term Memory (LSTM) networks, aimed at leveraging both singular and sequential data for disease prediction. The core of this novel architecture lies in its premise: combining the robust feature extraction capabilities of CNNs with the temporal analysis strength of LSTMs significantly enhances prediction accuracy compared to conventional LSTM models alone. In this framework, CNNs preprocess the input by identifying critical features[27], which the LSTM component then uses to track temporal variations in the data, offering a refined approach for disease forecasting.

The investigation not only validates the efficacy of the newly developed CNN-LSTM hybrid in predicting diseases from univariate data but also highlights its superiority over traditional LSTM approaches. This advancement holds significant implications for future medical research, particularly in the realms of precision medicine and personalized treatment strategies.

Looking ahead, expanding the application of this CNN-LSTM model to more complex and real-world medical datasets is a logical next step. This endeavor will not only corroborate the model's effectiveness and robustness across diverse data types and volumes but also demonstrate its capability in handling multivariate and unstructured data[28], such as medical imagery and narrative medical records, and in predicting a range of diseases. Furthermore, given the sensitive nature and complexity of medical data, future studies should explore strategies to leverage this model in a way that ensures patient confidentiality and incorporates advanced deep learning techniques (such as self-attention mechanisms and graph neural networks) to enhance its predictive capabilities.

Another crucial area for future research is enhancing the model's interpretability[29]. In healthcare, transparent interpretation of model outputs is essential for gaining the trust of healthcare professionals and patients alike[30]. Therefore, efforts to improve the interpretability of the CNN-LSTM hybrid, making its predictions more accessible and comprehensible, will be vital for its utility in clinical settings.

Additionally, considering the substantial data requirements of deep learning models, future research should investigate strategies for effective training with limited data resources, employing methods such as transfer learning, semi-supervised learning, or weakly supervised learning. Implementing these techniques would increase the model's feasibility in medical contexts where data availability is often restricted.

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