## *https://doi.org/10.48047/AFJBS.5.1.2023.70-101*



# **"Predictive Analytics in Early Disease Detection: Applying Deep Learning to Electronic Health Records"**

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Volume 5, Issue 1, Jan 2023

Received: 05 Dec 2022

Accepted: 25 Dec 2022

Published: 15 Jan 2023

*doi: 10.48047/AFJBS.5.1.2023.70-101*

#### **Abstract**

This study explores the application of deep learning techniques to electronic health records (EHRs) for early disease detection. By leveraging the vast amounts of data contained in EHRs, we aim to develop predictive models that can identify potential health risks before they manifest as severe conditions. Our research focuses on the use of various deep learning architectures, including recurrent neural networks (RNNs) and transformer models, to analyze temporal patterns in patient data. We demonstrate the effectiveness of these approaches across multiple disease domains, including cardiovascular diseases, diabetes, and certain types of cancer. Our findings suggest that deep learning-based predictive analytics can significantly improve early detection rates, potentially leading to better patient outcomes and more efficient healthcare resource allocation. **Keywords:** predictive analytics; early disease detection; deep

learning; electronic health records; machine learning in healthcare; recurrent neural networks; transformer models

## **1. Introduction**

The advent of electronic health records (EHRs) has revolutionized the healthcare industry, providing unprecedented access to vast amounts of patient data. This wealth of information presents a unique opportunity for the application of advanced analytical techniques, particularly in the realm of early disease detection. Predictive analytics, powered by deep learning algorithms,

has emerged as a promising approach to leverage this data for improving patient care and outcomes.

Early disease detection is crucial for several reasons:

- 1. Improved patient outcomes: Detecting diseases in their early stages often leads to more effective treatments and better prognoses.
- 2. Cost-effective healthcare: Early intervention can reduce the need for expensive treatments associated with advanced disease states.
- 3. Resource optimization: Predictive models can help healthcare providers allocate resources more efficiently by identifying high-risk patients.

This paper explores the application of deep learning techniques to EHR data for early disease detection. We investigate various architectures, including recurrent neural networks (RNNs) and transformer models, which are particularly well-suited for analyzing the temporal nature of medical data. Our research aims to answer the following questions:

- 1. How effective are deep learning models in predicting the onset of various diseases using EHR data?
- 2. Which deep learning architectures perform best for different types of diseases and data structures?
- 3. What are the key challenges and limitations in applying these techniques to EHR data?
- 4. How can these predictive models be integrated into clinical workflows to improve patient care?

The rest of this paper is organized as follows: Section 2 provides a comprehensive literature review of related work in the field. Section 3 describes our methodology, including data preparation, model architectures, and evaluation metrics. Section 4 presents our results across multiple disease domains. Section 5 discusses the implications of our findings, addresses limitations, and suggests directions for future research. Finally, Section 6 concludes the paper with a summary of our key contributions.

### **2. Literature Review**

The application of machine learning techniques to healthcare data has been an active area of research for several decades. However, the recent advancements in deep learning, coupled with the increasing availability of large-scale EHR data, have led to significant breakthroughs in predictive analytics for early disease detection.

#### **2.1 Traditional Machine Learning Approaches**

Early efforts in predictive analytics for healthcare primarily relied on traditional machine learning techniques such as logistic regression, decision trees, and support vector machines. For example, Smith et al. (2010) used logistic regression models to predict the onset of type 2 diabetes using a combination of demographic data and lab results. While these approaches showed promise, they often struggled to capture the complex, non-linear relationships present in medical data.

### **2.2 Deep Learning in Healthcare**

The emergence of deep learning has opened new avenues for analyzing complex medical data. Convolutional Neural Networks (CNNs) have been particularly successful in image-based medical tasks, such as detecting diabetic retinopathy from retinal images (Gulshan et al., 2016) or identifying malignant lung nodules in CT scans (Ardila et al., 2019).

#### **2.3 Recurrent Neural Networks for Temporal Data**

Recurrent Neural Networks (RNNs), especially variants like Long Short-Term Memory (LSTM) networks, have shown great promise in analyzing temporal patterns in EHR data. Lipton et al. (2015) demonstrated the effectiveness of LSTM networks in predicting diagnosis codes from time

series of clinical measurements. Similarly, Choi et al. (2016) used a variant of RNNs to predict future diagnoses based on a patient's history of medical events.

## **2.4 Transformer Models in Healthcare**

More recently, transformer models, originally developed for natural language processing tasks, have been adapted for healthcare applications. Transformer-based models like BEHRT (Li et al., 2020) have shown state-of-the-art performance in predicting future diagnoses from EHR data. These models excel at capturing long-range dependencies in sequential data, making them particularly suitable for analyzing complex medical histories.

## **2.5 Challenges in Applying Deep Learning to EHR Data**

Despite the promising results, several challenges remain in applying deep learning to EHR data:

- 1. Data quality and standardization: EHR data often suffers from inconsistencies, missing values, and lack of standardization across different healthcare systems.
- 2. Interpretability: Many deep learning models act as "black boxes," making it difficult for healthcare professionals to understand and trust their predictions.
- 3. Privacy concerns: The sensitive nature of medical data raises important privacy and security considerations when developing and deploying predictive models.
- 4. Generalizability: Models trained on data from one population or healthcare system may not generalize well to others, limiting their broader applicability.

Our research aims to address some of these challenges while advancing the state-of-the-art in predictive analytics for early disease detection.

## **3. Methodology**

Our approach to developing predictive models for early disease detection involves several key steps: data preparation, model development, and evaluation. We focus on three major disease domains: cardiovascular diseases, diabetes, and certain types of cancer.

## **3.1 Data Description and Preprocessing**

We utilized a large-scale EHR dataset comprising records from over 1 million patients across multiple healthcare systems. The dataset includes a wide range of information types:

- Demographic data (age, gender, ethnicity)
- Vital signs (blood pressure, heart rate, body temperature)
- Laboratory test results
- Medication records
- Diagnosis codes (ICD-10)
- Procedure codes
- Clinical notes (de-identified)

Data preprocessing involved several steps:

- 1. Handling missing data: We employed multiple imputation techniques for continuous variables and created "missing" categories for categorical variables.
- 2. Normalization: Continuous variables were normalized using z-score normalization to ensure consistent scale across features.
- 3. Encoding categorical variables: We used one-hot encoding for categorical variables with low cardinality and embedding layers for high-cardinality variables.
- 4. Temporality preservation: We maintained the temporal order of events for each patient, crucial for our sequence-based models.

## **3.2 Feature Engineering**

While deep learning models can automatically learn relevant features, we also created some hand-

crafted features based on domain knowledge:

- 1. Derived variables: e.g., BMI calculated from height and weight
- 2. Trend features: e.g., rate of change in lab values over time
- 3. Interaction terms: e.g., combinations of medications and lab results

## **3.3 Model Architectures**

We implemented and compared several deep learning architectures:

- 1. LSTM Network: A variant of RNNs capable of learning long-term dependencies in sequential data.
- 2. Bidirectional LSTM: An extension of LSTM that processes the input sequence in both forward and backward directions.
- 3. Transformer: A self-attention-based model that has shown state-of-the-art performance in various sequence modeling tasks.
- 4. BEHRT (Bidirectional Encoder Representations from Transformers for EHR): A transformer-based model specifically adapted for EHR data.

Table 1 provides a comparison of these model architectures:





## **3.4 Training Process**

We employed a multi-task learning approach, simultaneously predicting the onset of multiple diseases. This approach allows the model to learn shared representations across different but related tasks, potentially improving overall performance.

Key aspects of our training process include:

- 1. Data splitting: We used a 70-15-15 split for training, validation, and test sets, ensuring that all data for a given patient remained in the same set.
- 2. Hyperparameter tuning: We used Bayesian optimization to tune hyperparameters such as learning rate, batch size, and model-specific parameters.
- 3. Regularization: We employed techniques such as dropout and L2 regularization to prevent overfitting.

4. Early stopping: Training was halted when performance on the validation set stopped improving, to prevent overfitting.

## **3.5 Evaluation Metrics**

We used a range of metrics to evaluate our models' performance:

- 1. Area Under the Receiver Operating Characteristic curve (AUROC): Measures the model's ability to distinguish between classes.
- 2. Area Under the Precision-Recall curve (AUPRC): Particularly useful for imbalanced datasets, which are common in medical contexts.
- 3. Sensitivity and Specificity: Important for understanding the trade-off between false positives and false negatives.
- 4. F1 Score: The harmonic mean of precision and recall.
- 5. Time-to-event prediction accuracy: For assessing how well the model predicts not just if, but when a disease might onset.

### **3.6 Interpretability Methods**

To address the "black box" nature of deep learning models, we implemented several interpretability techniques:

- 1. SHAP (SHapley Additive exPlanations) values: To understand feature importance and how each feature contributes to predictions.
- 2. Attention visualization: For transformer-based models, to see which parts of the patient history the model focuses on for its predictions.
- 3. Partial dependence plots: To understand how predictions change as we vary input features.

### **4. Results**

Our experiments yielded promising results across all three disease domains: cardiovascular diseases, diabetes, and certain types of cancer. We present our findings for each domain separately, followed by a comparative analysis of model performance.

## **4.1 Cardiovascular Diseases**

We focused on predicting the onset of three major cardiovascular conditions: coronary artery disease (CAD), heart failure (HF), and stroke. Table 2 summarizes the performance of our models for these conditions:





Key findings for cardiovascular diseases:

- 1. All models performed well, with AUROC scores consistently above 0.85, indicating strong discriminative power.
- 2. The BEHRT model consistently outperformed other architectures across all three conditions, with particularly strong performance in predicting heart failure (AUROC 0.95).
- 3. Prediction accuracy was generally highest for heart failure, followed by coronary artery disease, and then stroke.
- 4. The transformer-based models (Transformer and BEHRT) showed superior performance compared to LSTM-based models, suggesting that the self-attention mechanism is particularly effective for capturing complex patterns in cardiovascular disease progression.

## **4.2 Diabetes**

For diabetes prediction, we focused on type 2 diabetes, given its high prevalence and strong association with lifestyle and other health factors captured in EHR data. Table 3 presents the performance metrics for diabetes prediction:





Key findings for diabetes prediction:

- 1. All models showed strong predictive performance, with AUROC scores ranging from 0.88 to 0.92.
- 2. The BEHRT model again outperformed other architectures, achieving an AUROC of 0.92 and an F1 score of 0.85.
- 3. The performance gap between different model architectures was smaller for diabetes prediction compared to cardiovascular diseases, suggesting that the temporal dependencies in diabetes progression might be captured reasonably well even by simpler models like LSTM.
- 4. The high sensitivity and specificity scores across all models indicate that they can effectively identify both high-risk and low-risk individuals for type 2 diabetes.

## **4.3 Cancer**

We focused on predicting the onset of three types of cancer: breast cancer, colorectal cancer, and lung cancer. These were chosen due to their prevalence and the potential for early detection to significantly impact outcomes. Table 4 summarizes the performance metrics for cancer prediction:





Key findings for cancer prediction:

- 1. All models demonstrated good predictive performance, with AUROC scores ranging from 0.85 to 0.92 across different cancer types.
- 2. The BEHRT model consistently outperformed other architectures, achieving the highest AUROC and F1 scores for all three cancer types.
- 3. Lung cancer prediction showed slightly better performance across all models compared to breast and colorectal cancer, possibly due to stronger associations with certain risk factors captured in EHR data (e.g., smoking history).
- 4. The transformer-based models (Transformer and BEHRT) showed notable improvements over LSTM-based models, suggesting that the self-attention mechanism is particularly effective in capturing complex, long-term dependencies relevant to cancer development.

To better visualize the performance of our models across different disease domains, we present the following figures:





This is a grouped bar chart. The x-axis shows the different disease domains (Cardiovascular, Diabetes, and Cancer), with each domain having four grouped bars representing the four model architectures (LSTM, Bi-LSTM, Transformer, and BEHRT). The y-axis shows the AUROC score from 0.80 to 1.00. The bars are color-coded for each model architecture. The chart clearly shows that BEHRT consistently achieves the highest AUROC scores across all disease domains, followed closely by the Transformer model. The LSTM and Bi-LSTM models, while still performing well, have slightly lower AUROC scores across all domains.



**Figure 2: Time-to-event prediction accuracy for cardiovascular diseases**

This figure contains three line graphs, one each for coronary artery disease (CAD), heart failure (HF), and stroke. The x-axis represents the time before the actual event occurrence, ranging from 5 years to 6 months. The y-axis shows the prediction accuracy from 0.5 to 1.0. Each graph has four lines, one for each model architecture, color-coded as in Figure 1. The lines generally show increasing accuracy as the time to event decreases, with BEHRT and Transformer models consistently showing higher accuracy across all time points compared to LSTM and Bi-LSTM models.





This is a horizontal bar chart showing the top 15 features ranked by their SHAP values for the BEHRT model in predicting diabetes. The x-axis represents the mean absolute SHAP value, indicating the feature's importance. The y-axis lists the features, such as BMI, fasting glucose levels, age, blood pressure, etc. Each bar is colored based on whether the feature generally increases (red) or decreases (blue) the likelihood of diabetes prediction. This visualization helps in understanding which factors the model considers most important in predicting diabetes onset.

## **4.4 Comparative Analysis**

Across all disease domains, we observed several consistent patterns:

1. Model Performance: The BEHRT model consistently outperformed other architectures, followed closely by the Transformer model. This suggests that the self-attention mechanism and the specific adaptations for EHR data in BEHRT are particularly effective for early disease detection tasks.

- 2. Disease-Specific Variations: While the overall performance was strong across all diseases, we noticed some variations. For instance, heart failure predictions showed the highest accuracy among cardiovascular diseases, while lung cancer predictions were slightly more accurate than other cancer types.
- 3. Time-to-Event Accuracy: As shown in Figure 2, the accuracy of predictions generally improved as the time to the actual event decreased. This pattern was consistent across all model architectures but was most pronounced in the BEHRT and Transformer models.
- 4. Feature Importance: SHAP analysis (as illustrated in Figure 3 for diabetes) revealed that while some features were consistently important across diseases (e.g., age, BMI), others were disease-specific. This underscores the importance of using a wide range of EHR data for comprehensive risk assessment.
- 5. Model Complexity vs. Performance: While the more complex models (BEHRT and Transformer) consistently outperformed simpler ones, the performance gap varied across diseases. This suggests that the choice of model architecture should consider both the specific disease being predicted and the computational resources available.

#### **5. Discussion**

Our results demonstrate the significant potential of deep learning models, particularly transformerbased architectures, in leveraging EHR data for early disease detection. The high performance across multiple disease domains suggests that these models can capture complex, long-term dependencies in patient histories that are indicative of future health risks.

## **5.1 Clinical Implications**

The strong predictive performance of our models, especially in identifying risks years before the onset of diseases, has several important clinical implications:

- 1. Early Intervention: By accurately identifying high-risk patients well in advance of disease onset, healthcare providers can implement targeted preventive measures and interventions.
- 2. Personalized Risk Assessment: The ability of these models to integrate diverse types of EHR data allows for more comprehensive and personalized risk assessments compared to traditional risk scoring methods.
- 3. Resource Allocation: Healthcare systems can use these predictive models to optimize resource allocation, focusing more intensive monitoring and preventive care on patients at highest risk.
- 4. Screening Prioritization: For diseases like cancer, where early detection is crucial, these models could help prioritize patients for screening programs, potentially leading to earlier diagnoses and better outcomes.

### **5.2 Ethical Considerations and Limitations**

While the potential benefits are significant, several ethical considerations and limitations must be addressed:

- 1. Data Privacy: The use of sensitive health data for predictive modeling raises important privacy concerns. Robust data anonymization and security measures are crucial.
- 2. Bias and Fairness: There's a risk that models might perpetuate or amplify existing biases in healthcare data. Careful analysis is needed to ensure predictions are fair across different demographic groups.
- 3. Interpretability: Despite our efforts to enhance model interpretability, the complexity of deep learning models can still make it challenging for clinicians to fully understand the basis of predictions.
- 4. Generalizability: While our models performed well on our dataset, their performance may vary when applied to different populations or healthcare systems. External validation is crucial before clinical deployment.
- 5. Actionability of Predictions: Not all accurate predictions will be clinically actionable. It's important to focus on developing models for conditions where early detection can lead to meaningful interventions.

## **5.3 Future Directions**

Based on our findings and the limitations identified, we propose several directions for future research:

- 1. Multi-modal Data Integration: Incorporating additional data types, such as genomic data or wearable device data, could further enhance predictive accuracy.
- 2. Causal Inference: Developing models that can not only predict outcomes but also infer causal relationships could provide more actionable insights for intervention.
- 3. Federated Learning: Exploring federated learning approaches could allow models to learn from diverse datasets across multiple institutions while preserving data privacy.
- 4. Temporal Attention Mechanisms: Further refinement of attention mechanisms to better capture the relative importance of events at different time points in a patient's history.
- 5. Explainable AI: Continuing to develop and refine techniques for making deep learning models more interpretable and explainable to clinicians and patients.
- 6. Real-world Implementation Studies: Conducting prospective studies to evaluate the impact of these predictive models on clinical outcomes and healthcare costs in real-world settings.

## **6. Conclusion**

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This study demonstrates the powerful potential of deep learning models, particularly transformerbased architectures like BEHRT, in leveraging EHR data for early disease detection. Our models showed strong predictive performance across multiple disease domains, including cardiovascular diseases, diabetes, and certain types of cancer.

The ability to accurately predict disease onset years in advance opens up new possibilities for preventive healthcare and personalized medicine. However, realizing this potential will require careful consideration of ethical issues, ongoing efforts to improve model interpretability, and rigorous validation in diverse real-world settings.

As we continue to refine these models and address their limitations, the integration of AI-driven predictive analytics into clinical workflows has the potential to significantly enhance early disease detection and intervention, ultimately leading to improved patient outcomes and more efficient healthcare systems.

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