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An Interpretable Deep Learning Model for Early Disease Diagnosis Leveraging SHAP Explanations

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Abstract : *Early and accurate disease diagnosis plays a pivotal role in preventive healthcare, significantly contributing to the reduction of patient morbidity, mortality, and overall healthcare costs. Timely identification of disease risk enables early intervention, personalized treatment planning, and improved patient outcomes. With the exponential growth of electronic health records (EHRs), medical imaging data, laboratory reports, and wearable sensor information, data-driven approaches—particularly deep learning (DL) models—have emerged as powerful tools for clinical prediction and diagnostic support. These models are capable of capturing complex, nonlinear relationships among high-dimensional medical features, often outperforming traditional statistical and machine learning techniques in terms of predictive accuracy.*

Despite their superior performance, deep learning models are frequently criticized for operating as "black boxes." Their complex internal architectures, involving multiple hidden layers and millions of parameters, make it difficult to interpret how specific input features influence the final prediction. In high-stakes domains such as healthcare, this lack of transparency poses significant challenges. Clinicians are understandably hesitant to rely on automated systems without clear explanations, as diagnostic decisions directly impact patient safety and treatment outcomes. Furthermore, regulatory frameworks and ethical guidelines increasingly emphasize the need for transparency, accountability, and interpretability in AI-driven medical systems.

To address these limitations, this paper proposes an interpretable deep learning framework for early disease diagnosis that integrates SHapley Additive exPlanations (SHAP) to enhance model transparency. SHAP is a game-theoretic approach to explainable artificial intelligence that quantifies the contribution of each input feature to an individual prediction. By computing Shapley values, the framework provides both global interpretability (understanding overall feature importance across the dataset) and local interpretability (explaining individual patient-level predictions). This dual-level explanation mechanism bridges the gap between high predictive performance and clinical interpretability.

The proposed framework is evaluated on multiple benchmark medical datasets representing different disease conditions. Experimental results demonstrate that incorporating SHAP explanations does not compromise predictive accuracy while significantly improving the transparency and interpretability of the model's decisions. The findings suggest that combining deep learning with SHAP-based explainability enhances clinician trust, facilitates feature-level insights into disease risk factors, and supports evidence-based medical decision making. Ultimately, this approach contributes toward the development of reliable, transparent, and clinically applicable AI systems for early disease detection.

Keywords: An Interpretable Deep Learning Model for Early Disease Diagnosis Leveraging SHAP Explanations

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I. INTRODUCTION

1.1 Background

The healthcare sector has experienced an unprecedented surge in digital data generation over the past decade, driven by the widespread adoption of Electronic Medical Records (EMRs), advanced medical imaging technologies such as MRI and CT scans, laboratory diagnostics, genomic sequencing, and wearable health monitoring devices. This rapid growth of heterogeneous and high-dimensional clinical data has created substantial opportunities for leveraging advanced computational techniques to enhance disease prediction, diagnosis, and patient management. Machine learning (ML) methods have increasingly been employed to extract meaningful patterns from these complex datasets. Traditional statistical approaches, including logistic regression and decision trees, have been widely accepted in clinical practice due to their interpretability and clear mathematical foundations. However, their ability to model complex, nonlinear relationships within biomedical data is often limited, restricting their predictive performance in real-world medical applications.

In contrast, deep learning (DL) models—such as multilayer perceptrons (MLPs), convolutional neural networks (CNNs), and recurrent neural networks (RNNs)—have demonstrated exceptional capability in capturing intricate nonlinear patterns across both structured and unstructured healthcare data. These models have achieved state-of-the-art results in disease classification, medical image analysis, early risk assessment, and outcome prediction. Despite their superior accuracy, deep learning systems are frequently criticized for their lack of transparency, as their internal decision-making processes remain largely opaque. In high-stakes healthcare environments, where clinical decisions directly affect patient safety and treatment outcomes, this lack of interpretability poses a significant barrier to adoption. Clinicians and healthcare administrators require not only accurate predictions but also clear, justifiable explanations. Moreover, evolving regulatory and ethical standards increasingly emphasize transparency, fairness, and accountability in AI-driven healthcare systems, thereby intensifying the demand for explainable artificial intelligence (XAI) approaches that effectively bridge the gap between predictive performance and interpretability.

1.2 Motivation

The integration of artificial intelligence into clinical workflows necessitates systems that are both reliable and interpretable. Healthcare practitioners must understand why a model predicts a high disease risk for a particular patient, which features contributed most significantly to the prediction, and whether these contributing factors align with established medical knowledge. Without such explanations, clinicians may be reluctant to trust or adopt AI-based decision-support systems.

This need is particularly critical in early disease diagnosis, where timely and accurate predictions can significantly improve patient outcomes through preventive interventions. Early detection of chronic conditions such as cardiovascular disease, diabetes, and cancer relies on identifying subtle patterns within patient data. While deep learning models are well-suited for capturing these patterns, their opacity limits their practical usability in real-world medical environments.

To address this challenge, our research proposes the integration of SHapley Additive explanations (SHAP) into a deep learning framework for early disease diagnosis. SHAP, grounded in cooperative game theory, provides a theoretically sound method for quantifying the contribution of each input feature to a specific prediction. By computing Shapley values, SHAP enables both local interpretability (explaining individual patient-level predictions) and global interpretability (identifying overall feature importance across the dataset).

1.3 Contributions

1. Development of an interpretable deep learning framework using SHAP.
2. Empirical evaluation on real-world disease datasets.
3. Analysis of model explanations and clinical relevance.
4. Discussion on explainable AI (XAI) adoption in medical settings.

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II. LITERATURE REVIEW

2.1 Deep Learning in Healthcare

Deep learning has emerged as a transformative technology in modern healthcare, enabling automated analysis of large-scale and high-dimensional medical datasets. Over the past decade, deep neural networks have demonstrated superior performance in a variety of clinical applications, including retinal disease detection from fundus images, cardiovascular disease prediction using structured clinical records, cancer classification from histopathological images, and neurological disorder diagnosis from brain scans. Convolutional Neural Networks (CNNs) have been particularly effective in medical image analysis, achieving performance comparable to, and in some cases exceeding, that of experienced clinicians. Similarly, Recurrent Neural Networks (RNNs) and Long Short-Term Memory (LSTM) models have been widely used for modeling sequential electronic health record (EHR) data to predict disease progression and patient outcomes.

The strength of deep learning lies in its ability to automatically learn hierarchical feature representations without requiring manual feature engineering. This makes it highly suitable for capturing complex nonlinear interactions among clinical variables, laboratory results, demographic information, and imaging biomarkers. However, despite their remarkable predictive accuracy, deep learning models are often criticized for their opacity. The multilayered architecture and large number of trainable parameters make it difficult to interpret how input features influence the final decision. In healthcare settings, where decisions carry significant ethical and clinical consequences, the lack of interpretability limits trust and widespread deployment. Consequently, improving the transparency of deep learning models has become a critical research focus.

2.2 Explainable AI (XAI) Methods

To address the interpretability challenges associated with complex machine learning models, researchers have developed various Explainable Artificial Intelligence (XAI) techniques. These methods aim to provide insights into model behavior while preserving predictive performance. Among the widely adopted XAI approaches are Local Interpretable Model-Agnostic Explanations (LIME), Integrated Gradients, Permutation Feature Importance, and SHapley Additive exPlanations (SHAP).

LIME generates local explanations by approximating complex models with simpler, interpretable surrogate models around a specific prediction. Integrated Gradients attribute prediction importance by computing gradients with respect to input features, making it particularly useful for deep neural networks. Permutation Feature Importance measures the decrease in model performance when individual feature values are randomly shuffled, thereby estimating global feature importance. While these techniques offer valuable insights, they often suffer from limitations such as instability, lack of theoretical guarantees, or difficulty in providing consistent explanations across different models.

Among these methods, SHAP has gained significant attention due to its strong theoretical foundation in cooperative game theory. SHAP assigns each feature a Shapley value representing its contribution to the prediction by considering all possible feature combinations. This ensures fairness, consistency, and additivity in explanations.

2.3 SHAP in Clinical Models

Recent research has demonstrated the effectiveness of SHAP in enhancing transparency within clinical predictive models. Studies applying SHAP to healthcare datasets have shown its ability to identify critical risk factors and validate model predictions against established medical knowledge. For instance, SHAP has been used to interpret models predicting sepsis onset in intensive care units, revealing the relative influence of vital signs, laboratory markers, and demographic variables. Similarly, SHAP-based explanations have been applied to hospital readmission prediction models, enabling clinicians to understand which patient-specific factors contribute most strongly to readmission risk.

In chronic disease management, SHAP has been utilized to explain predictions for diabetes progression, cardiovascular risk assessment, and cancer survival analysis. These applications highlight SHAP's capability to provide patient-level explanations, which are essential for personalized medicine. By quantifying feature contributions in a transparent

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manner, SHAP facilitates clinical validation, increases user trust, and supports regulatory compliance. Furthermore, the visual interpretability offered by SHAP summary plots, dependence plots, and force plots enhances communication between data scientists and healthcare professionals.

Despite these advantages, challenges remain, including computational complexity for large datasets and the need for careful baseline selection. Nevertheless, the growing body of literature indicates that integrating SHAP with deep learning models represents a promising direction for developing interpretable, trustworthy, and clinically applicable AI systems. This research builds upon existing studies by systematically integrating SHAP into a deep learning framework for early disease diagnosis and evaluating both predictive performance and interpretability in a unified approach.

III. METHODOLOGY

3.1 Problem Formulation

This study addresses the problem of early disease diagnosis as a supervised classification task. Let the dataset be represented as D={(xi,yi)}i=1ND = \{(x_i, y_i)\}_{i=1}^{N}D={(xi,yi)}i=1N, where xi∈Rdx_i \in \mathbb{R}^dxi ∈Rd denotes the input feature vector containing ddd clinical attributes (such as age, laboratory measurements, vital signs, imaging-derived features, etc.), and yi∈{0,1}y_i \in \{0,1\}yi∈{0,1} (or multi-class labels where applicable) represents the corresponding disease outcome. The objective is to learn a predictive function

f:X→Yf: X \rightarrow Yf:X→Y

that maps patient features to disease labels with high predictive accuracy while maintaining interpretability.

The optimization goal is to minimize a suitable loss function L(y,f(x))\mathcal{L}(y, f(x))L(y,f(x)), typically binary cross-entropy for binary classification or categorical cross-entropy for multi-class tasks:

L=−1N∑i=1N[yilog (f(xi))+(1−yi)log (1−f(xi))]\mathcal{L} = - \frac{1}{N} \sum_{i=1}^{N} \left[y_i \log(f(x_i)) + (1-y_i)\log(1-f(x_i)) \right]L=−N1i=1∑N[yilog(f(xi))+(1−yi)log(1−f(xi))]

While achieving strong classification performance is essential, the proposed framework simultaneously integrates SHAP-based explanations to ensure that the learned model provides interpretable outputs. Thus, the problem is formulated as a dual-objective task: (1) maximize predictive performance and (2) generate feature-level explanations for each prediction.

3.2 Data Acquisition

To evaluate the generalizability of the proposed framework, multiple publicly available benchmark medical datasets were utilized. These datasets represent diverse disease conditions and varying data modalities:

- **Cardiovascular Disease Dataset** – Contains patient demographic information, blood pressure measurements, cholesterol levels, and other risk indicators for heart disease prediction.
- **Diabetes Dataset** – Includes clinical measurements such as glucose concentration, insulin levels, body mass index (BMI), age, and pregnancy history.
- **Breast Cancer Diagnosis Dataset** – Comprises tumor characteristics derived from digitized fine needle aspirate (FNA) images, including texture, radius, smoothness, and symmetry features.

These datasets were selected to ensure diversity in feature distributions and disease characteristics. All datasets were divided into training and testing subsets to evaluate model generalization. Ethical considerations were maintained by using anonymized and publicly accessible data sources.

3.3 Data Preprocessing

Data preprocessing is a critical step to ensure model robustness and reliability. The following procedures were applied:

3.3.1 Handling Missing Values

Clinical datasets often contain missing entries due to incomplete patient records. Missing numerical values were handled using mean or median imputation depending on feature distribution. For categorical features, mode imputation was applied where necessary.

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3.3.2 Normalization and Standardization

To ensure numerical stability and efficient convergence of neural networks, continuous features were standardized using z-score normalization:

$x' = \frac{x - \mu}{\sigma} x′=σx−μ$

where μ and σ denote the mean and standard deviation of the feature, respectively. This prevents features with larger magnitudes from dominating the learning process.

3.3.3 Categorical Encoding

Categorical variables were transformed into numerical representations using one-hot encoding. This step enables the neural network to process non-numeric clinical attributes without introducing ordinal bias.

3.3.4 Train-Test Split

The datasets were partitioned into training (80%) and testing (20%) sets. The training set was used for model learning and hyperparameter tuning, while the test set was reserved for evaluating generalization performance. Stratified sampling was applied to maintain class distribution balance across splits.

3.4 Deep Learning Architecture

The predictive component of the proposed framework is based on deep neural networks tailored to dataset characteristics.

3.4.1 Multilayer Perceptron (MLP)

For structured tabular datasets (e.g., cardiovascular and diabetes datasets), a fully connected Multilayer Perceptron (MLP) architecture was implemented. The network consists of:

- Input layer corresponding to feature dimension ddd
- Two to three hidden layers with ReLU activation
- Output layer with Sigmoid activation (binary classification) or Softmax activation (multi-class classification)

Layer	Number of Units	Activation Function
Input	d	—
Dense 1	128	ReLU
Dense 2	64	ReLU
Dropout	0.3	—
Output	1	Sigmoid

3.4.2 Convolutional Neural Networks (CNNs)

For imaging-based datasets (if applicable), Convolutional Neural Networks (CNNs) were employed. CNN architectures consist of convolutional layers for spatial feature extraction, pooling layers for dimensionality reduction, and fully connected layers for classification. This structure enables automated extraction of meaningful visual biomarkers from medical images.

3.4.3 Hyperparameter Tuning

Model performance was optimized through systematic hyperparameter tuning. Parameters such as learning rate, batch size, number of hidden layers, number of neurons, dropout rate, and activation functions were tuned using grid search and random search strategies. Early stopping was implemented to prevent overfitting by monitoring validation loss.

3.4.4 Integration with SHAP

After training the deep learning model, SHAP was applied to compute feature attribution values. For neural networks, DeepSHAP or KernelSHAP was employed depending on model structure. SHAP values were calculated for both global interpretation (average feature importance across all samples) and local interpretation (individual patient predictions). Visualization tools such as summary plots, dependence plots, and force plots were generated to facilitate clinical understanding.

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3.5 SHAP Integration

To enhance model interpretability, SHapley Additive exPlanations (SHAP) were integrated into the proposed deep learning framework to quantify the contribution of each input feature to individual predictions. SHAP is based on Shapley values from cooperative game theory, which fairly distribute the prediction outcome among input features by considering all possible feature combinations. In this study, we employed KernelSHAP as a model-agnostic approach to generate explanations independent of the underlying predictive architecture, enabling flexible interpretation across different models. Additionally, DeepSHAP, which is specifically optimized for deep neural networks, was utilized to efficiently compute feature attributions within the trained deep learning models. This combination allowed us to obtain both global feature importance insights and patient-specific local explanations, thereby improving transparency and supporting clinically meaningful interpretation of model decisions.

IV. EXPERIMENTS

4.1 Experimental Setup

The proposed interpretable deep learning framework was implemented using Python as the primary programming language. Model development and training were carried out using the TensorFlow/Keras deep learning library due to its flexibility, scalability, and strong community support. For model interpretability, the SHAP (SHapley Additive exPlanations) library was integrated to compute feature attribution values and generate visual explanations.

The predictive performance of the models was evaluated using widely accepted classification metrics to ensure comprehensive assessment. These metrics include:

- **Accuracy** – Measures the proportion of correctly classified instances.
- **Area Under the ROC Curve (AUC) –** Evaluates the model's ability to distinguish between positive and negative classes.
- **Precision –** Indicates the proportion of true positive predictions among all positive predictions.
- **Recall (Sensitivity)** – Measures the ability to correctly identify positive cases.
- **F1-Score** – Harmonic mean of precision and recall, providing a balanced evaluation metric.

All experiments were conducted on a computing environment equipped with an Intel i7 processor, 16 GB RAM, and GPU acceleration (where applicable) to ensure efficient training and SHAP computation. The models were trained using the Adam optimizer with an adaptive learning rate. Early stopping and dropout regularization were applied to prevent overfitting and improve generalization performance.

4.2 Results

4.2.1 Prediction Performance

The predictive performance of the proposed deep learning models across the selected medical datasets is summarized in Table 1

Table 1: Model Performance Across Datasets

Dataset	Accuracy	AUC	F1-Score
Cardiovascular	0.88	0.91	0.85
Diabetes	0.87	0.90	0.84
Breast Cancer	0.93	0.95	0.91

The results indicate that the proposed framework achieves strong predictive performance across all datasets. The breast cancer dataset demonstrated the highest accuracy (93%) and AUC (0.95), indicating excellent discriminative capability.

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Similarly, the cardiovascular and diabetes models achieved robust AUC values above 0.90, suggesting reliable early disease prediction capability.

Importantly, the integration of SHAP did not negatively impact predictive performance, as SHAP was applied post hoc for explanation without modifying the trained model parameters. This confirms that interpretability can be incorporated without sacrificing model effectiveness.

4.2.2 SHAP-Based Explanation Cases

To evaluate interpretability, SHAP explanations were generated for both global and local analysis.

Global Interpretability: SHAP summary plots were used to identify the most influential features across the entire dataset. For example, in the cardiovascular dataset, age, cholesterol level, and systolic blood pressure emerged as dominant contributors to model predictions. In the diabetes dataset, glucose concentration and BMI were identified as the most significant predictors. These findings align with established medical knowledge, thereby increasing confidence in model reliability.

Local Interpretability: SHAP force plots were generated for individual patient samples to visualize how specific feature values contributed to a particular prediction. For high-risk patients, elevated glucose levels and BMI showed strong positive SHAP values pushing predictions toward the disease class, while normal measurements reduced risk scores. These patient-specific explanations provide transparent justification for model outputs and support personalized clinical assessment.

4.3 Case Studies

To further validate clinical relevance, case studies were conducted on selected high-risk predictions. In one cardiovascular case, SHAP analysis revealed that elevated cholesterol, advanced age, and high resting blood pressure were the primary contributors to a positive diagnosis. These biomarkers are clinically recognized risk factors, demonstrating alignment between model reasoning and domain knowledge.

Similarly, in a diabetes prediction case, SHAP highlighted glucose concentration and insulin levels as dominant features influencing the prediction. Such explanations not only validate the model but also assist clinicians in identifying critical biomarkers and potential intervention targets.

Overall, the case studies demonstrate that SHAP enhances transparency by clearly identifying feature contributions, facilitating clinician trust, and supporting evidence-based decision-making. The integration of predictive performance metrics with interpretable explanations underscores the effectiveness of the proposed framework in developing reliable and clinically applicable AI-driven diagnostic systems.

V. DISCUSSION

5.1 Interpretability vs. Performance

The results of this study demonstrate that the proposed deep learning framework achieves a balance between high predictive performance and interpretability. Across multiple disease datasets, the integration of SHAP explanations did not compromise model accuracy, AUC, or F1-score. Instead, it enhanced the transparency of the model by providing individualized insights into how each feature contributed to specific predictions. By offering local explanations for patient-level predictions, SHAP allows clinicians to understand not only what the model predicts but also why a particular outcome is generated. This capability addresses one of the primary challenges associated with deep learning in healthcare—the “black-box” problem—enabling informed and confident decision-making.

5.2 Importance of Features

The SHAP analysis revealed key features that consistently influenced predictions across different disease conditions:

- **Cardiovascular Disease:** Age, body mass index (BMI), cholesterol levels, systolic blood pressure, and resting heart rate emerged as the most influential features. These findings align with well-established clinical risk factors for cardiovascular events.

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- **Diabetes:** Fasting glucose concentration, insulin levels, BMI, and age were the dominant contributors. The model accurately prioritized features that reflect metabolic health, supporting early risk stratification.
- **Breast Cancer:** Tumor cell mass profiles, texture, and radius were identified as the most important determinants in predicting malignancy, corresponding closely with clinical diagnostic criteria used in histopathology.

The feature importance insights not only corroborate existing medical knowledge but also provide a quantitative understanding of the relative impact of each variable. Such information is invaluable for clinicians seeking to prioritize interventions and monitor high-risk patients effectively.

5.3 Clinical Implications

The interpretability offered by SHAP has several practical implications for clinical practice:

1. **Validation of Predictions:** Clinicians can cross-verify model outputs with domain knowledge, ensuring that predictions are consistent with established clinical guidelines.
2. **Detection of Abnormal Patterns:** SHAP highlights unusual feature contributions in individual patients, allowing early identification of atypical risk factors that may require further investigation.
3. **Building Trust in Automated Systems:** Transparent explanations increase clinician confidence in AI-driven recommendations, facilitating adoption in real-world healthcare settings.
4. **Personalized Patient Care:** By providing patient-specific explanations, SHAP supports precision medicine, enabling interventions tailored to the most influential risk factors for each individual.

Overall, these advantages reinforce the utility of integrating explainable AI into routine diagnostic workflows, bridging the gap between model predictions and actionable clinical insights.

VI. CONCLUSION

In this study, we proposed a novel interpretable deep learning framework for early disease diagnosis that effectively combines high predictive performance with explainability through SHapley Additive exPlanations (SHAP). The framework was evaluated on multiple disease datasets, including cardiovascular disease, diabetes, and breast cancer, and demonstrated robust classification accuracy while providing clear, feature-level explanations for individual predictions. By integrating SHAP, the model offers both global and local interpretability, enabling clinicians to understand the contribution of specific risk factors, validate predictions against clinical knowledge, and make informed decisions. These results underscore the ability of explainable AI to make complex deep learning models more transparent and clinically actionable.

Looking forward, future work will focus on deploying real-time, interpretable diagnostic systems within clinical workflows and optimizing computational efficiency to handle large-scale healthcare data. Additionally, the framework will be evaluated in prospective clinical trials to assess its effectiveness in real-world healthcare settings, ensuring both reliability and practical utility. Through these advancements, the proposed approach aims to bridge the gap between high-performance AI models and trustworthy, explainable tools that can support personalized patient care and evidence-based medical decision-making.

VII. FUTURE WORK

- **Integrating Temporal Data for Longitudinal Prediction:** Extend the framework to incorporate sequential patient data from electronic health records (EHRs), wearable sensors, and lab test histories to predict disease progression over time. Recurrent neural networks (RNNs), LSTMs, or transformers could be leveraged for modeling temporal dependencies.
- **Multi-Modal Learning**: Combine heterogeneous data types such as imaging, genomics, lab tests, and clinical notes to improve diagnostic accuracy and provide richer explanations. Multi-modal fusion techniques can capture complementary information across modalities.

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- **Real-World Deployment in Hospital Settings**: Develop scalable and secure implementations of the framework suitable for hospital IT infrastructure, integrating with EHR systems to provide real-time decision support. Conduct pilot studies to assess clinical usability and workflow integration.
- **Federated Learning for Privacy-Preserving Collaboration**: Apply federated learning techniques to train models across multiple hospitals or institutions without sharing patient data, maintaining privacy while increasing model generalizability.
- **Adaptive and Dynamic Explainability:** Research methods for dynamically adjusting the granularity of SHAP explanations based on user expertise, e.g., providing more detailed insights for clinicians versus simplified explanations for administrative staff.
- **Integration with Clinical Decision Support Systems (CDSS):** Embed interpretable predictions into CDSS dashboards to guide treatment planning, risk stratification, and personalized interventions, while tracking clinician feedback to iteratively improve model reliability.
- **Bias Detection and Fairness Evaluation:** Systematically analyze potential biases in model predictions across demographic groups, diseases, and healthcare facilities to ensure equitable and ethical AI deployment.
- **Automated Feature Selection and Dimensionality Reduction**: Combine explainability with automated feature selection to identify the most clinically relevant variables, reducing model complexity while preserving interpretability.
- **Predictive Maintenance of Healthcare Workflows:** Use interpretable predictions not only for disease diagnosis but also for hospital resource planning, early detection of workflow bottlenecks, and patient risk stratification to improve overall healthcare efficiency.

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