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(Research Article)

Explainable AI for Early Detection and Classification of Childhood Leukemia Using Multi-Modal Medical Data

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Abstract

Childhood leukemia is one of the most common forms of cancer in children, making early detection and accurate diagnosis crucial for improving patient outcomes. Traditional methods of diagnosing leukemia, including manual analysis of medical images and blood tests, are often time-consuming and prone to human error. Recent advancements in artificial intelligence (AI) have shown promise in improving diagnostic accuracy and speed. This paper presents an explainable AI (XAI) approach for the early detection and classification of childhood leukemia using multi-modal medical data, including blood test results, imaging data, and clinical history. Our proposed system integrates deep learning models for image classification with machine learning algorithms for structured data analysis, resulting in a multi-modal framework capable of not only detecting leukemia at an early stage but also providing interpretable predictions. This paper outlines the methodology, system architecture, and experimental setup used to build the model, as well as the results obtained from evaluation using publicly available medical datasets. We demonstrate that the integration of multi-modal data improves classification accuracy compared to single-modal systems and that the explainability of the AI model helps healthcare professionals interpret the results, thus increasing trust in AI-driven decision-making systems. Finally, we discuss the future implications of this approach and potential areas for further development.

Keywords: Explainable AI (XAI); Childhood Leukemia; Early Detection; Classification; Multi-Modal Medical Data; Machine Learning; Medical Imaging; Blood Tests; Genetic Data; Transparency; Deep Learning

1. Introduction

Childhood leukemia is a leading cause of cancer-related deaths among children worldwide. According to the American Cancer Society, leukemia accounts for approximately 30% of all cancer diagnoses in children. The current methods for diagnosing leukemia, such as blood tests, bone marrow analysis, and medical imaging, are often invasive, time-consuming, and subject to human error. Furthermore, the interpretation of these results can vary depending on the experience of the clinician, which can lead to delays in diagnosis and suboptimal treatment plans. Recent advances in artificial intelligence (AI) have demonstrated the potential to revolutionize medical diagnostics by leveraging large datasets and computational power to uncover patterns that may not be immediately obvious to human practitioners. However, the opacity of many AI models, particularly deep learning algorithms, raises concerns about trust, especially in critical domains like healthcare. This calls for the adoption of explainable AI (XAI) techniques that not only improve model performance but also provide transparent and interpretable results for healthcare providers. In this paper, we explore the application of XAI for the early detection and classification of childhood leukemia. By integrating multi-modal medical data, such as blood test results, imaging data, and genetic information, we aim to develop a model that is both accurate and explainable, providing clear insights into its decision-making process.

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1.1. Background and Motivation

Childhood leukemia, primarily acute lymphoblastic leukemia (ALL) and acute myeloid leukemia (AML), is a malignant disease that affects blood and bone marrow. It is the most prevalent cancer among children, with thousands of new cases diagnosed annually worldwide. Early and accurate detection is crucial for improving survival rates, as delayed diagnosis often results in poor prognosis. Traditional diagnostic approaches, including blood tests and medical imaging (e.g., MRI, CT scans), rely heavily on the expertise of clinicians. However, these methods can be time-consuming and sometimes yield ambiguous results, especially in the early stages of the disease when symptoms may be subtle. In recent years, the adoption of artificial intelligence (AI) in healthcare has seen a rapid increase, with machine learning (ML) models showing promise in aiding diagnosis and classification tasks. However, the challenge of making AI systems transparent and understandable for medical professionals remains a significant hurdle, particularly when dealing with life-threatening conditions such as leukemia. Explainable AI (XAI) techniques offer a way to bridge this gap by making machine learning models more interpretable, allowing healthcare providers to trust and act on AI-generated predictions.

1.2. Problem Statement

The lack of reliable and interpretable AI models for early leukemia detection hinders their widespread adoption in clinical settings. While traditional machine learning models have been used for leukemia classification, their lack of transparency and explainability often limits their utility in high-stakes medical environments. The problem addressed by this research is to develop an explainable AI system that can accurately detect and classify childhood leukemia from multi-modal medical data while providing interpretable results that can be used by healthcare professionals.

1.3. Proposed Solution

We propose an integrated framework that leverages explainable AI (XAI) techniques to detect and classify childhood leukemia early, using multi-modal data sources. This system will combine medical imaging data, such as X-ray and MRI scans, with structured data from blood tests and patient history. By utilizing a hybrid approach that combines deep learning-based image analysis and machine learning models for structured data, our system aims to offer high classification accuracy with the added benefit of model interpretability. This will not only improve the detection of leukemia at an early stage but also provide actionable insights for clinicians, ensuring better decision-making.

1.4. Contributions

We propose a novel explainable AI framework for early detection and classification of childhood leukemia using multi-modal medical data.

The system integrates deep learning models for image analysis and machine learning models for structured data, achieving improved classification performance.

We demonstrate how explainable AI techniques can enhance the trust and interpretability of AI-based predictions, facilitating their adoption in clinical practice.

Experimental results show the effectiveness of our proposed system, demonstrating significant improvements in classification accuracy compared to single-modal methods.

2. Related Work

The application of artificial intelligence (AI) in childhood leukemia diagnosis has gained significant attention in recent years. Researchers have primarily focused on using deep learning algorithms for medical image analysis and traditional machine learning techniques for clinical data analysis. However, the complexity and heterogeneity of multi-modal medical data, which often includes blood test results, imaging data, and genetic information, have presented challenges in achieving both high accuracy and interpretability. This section reviews the existing literature in the field, highlighting key advancements and identifying gaps that this study seeks to address.

2.1. AI in Childhood Leukemia Diagnosis

AI has been increasingly employed for the diagnosis of leukemia, leveraging both imaging data and clinical features. Studies have demonstrated the potential of machine learning algorithms for analyzing blood smears, bone marrow biopsies, and other diagnostic images. For instance, a study by Zhang et al. (2020) used deep convolutional neural networks (CNNs) to classify blood smear images for leukemia detection, achieving high accuracy rates in identifying malignant cells[1]. Similarly, other studies have integrated clinical data, such as white blood cell counts, to assist in leukemia diagnosis[2]. These approaches, however, tend to be confined to a single type of data (e.g., imaging or clinical) and often lack the ability to provide interpretable explanations of model predictions.

2.2. Limitations of Traditional AI Models

Traditional machine learning models in healthcare have often been criticized for their lack of transparency. These models, particularly deep learning-based approaches, can be perceived as "black boxes," providing no insight into how they make predictions. This opacity limits their application in critical areas like healthcare, where trust and interpretability are essential. Research by Ribeiro et al. (2016) introduced Local Interpretable Model-Agnostic Explanations (LIME), a technique designed to make machine learning models more interpretable by approximating complex models with simpler, interpretable ones[3]. However, while LIME and similar methods have demonstrated success in many fields, their application in the healthcare domain, especially in childhood leukemia diagnosis, is still limited.

2.3. Explainable AI (XAI) in Healthcare

The need for explainable AI (XAI) has become increasingly important in medical diagnostics. XAI techniques aim to make machine learning models more transparent, helping clinicians understand the reasoning behind predictions. One prominent approach is Shapley Additive Explanations (SHAP), which assigns a "Shapley value" to each feature, indicating its contribution to a particular prediction[4]. SHAP has been widely adopted in various healthcare applications, including breast cancer detection[5] and heart disease risk prediction[6]. However, the integration of XAI in childhood leukemia diagnosis, particularly using multi-modal data, remains an underexplored area.

2.4. Multi-Modal Data Integration for Leukemia Diagnosis

Recent studies have emphasized the importance of integrating multiple data sources for accurate and comprehensive leukemia diagnosis. Multi-modal data, such as blood test results, medical imaging, and genetic information, provide a more holistic view of the disease, improving diagnostic accuracy. For example, a study by Li et al. (2019) combined imaging data and clinical features to predict leukemia outcomes with promising results[7]. However, challenges remain in effectively combining these data types within a unified machine learning model that maintains both accuracy and interpretability. Several studies have also focused on the integration of genetic data into diagnostic models. Genomic information, such as gene mutations and expression profiles, has been shown to play a crucial role in leukemia classification[8]. However, the inclusion of genetic data requires sophisticated methods to handle the high dimensionality and complexity of genetic features. In this context, machine learning algorithms capable of processing large-scale data, such as deep neural networks, are essential[9]. Despite the potential of multi-modal data, integrating these various data sources while ensuring explainability has not been extensively explored.

2.5. Research Gaps and Future Directions

While there has been significant progress in using AI for leukemia diagnosis, there remains a gap in the literature regarding the integration of multi-modal data in childhood leukemia detection with a focus on explainability. Many existing studies focus on single-source data, such as imaging or clinical data, without addressing the full range of available medical data. Additionally, while explainability techniques like LIME and SHAP have been used in healthcare, their application in leukemia diagnosis, particularly with multi-modal data, is limited. This paper aims to fill these gaps by proposing a unified machine learning model that integrates clinical, imaging, and genetic data for the early detection and classification of childhood leukemia, with a strong emphasis on model transparency and interpretability.

3. Methodology

In this study, we propose a machine learning-based model for the early detection and classification of childhood leukemia, leveraging multi-modal medical data. The primary goal is to develop an approach that not only achieves high diagnostic accuracy but also provides transparency in the decision-making process. This is achieved through the application of explainable artificial intelligence (XAI) techniques such as SHAP (Shapley Additive Explanations) and LIME (Local Interpretable Model-Agnostic Explanations), which provide insight into how the model makes its predictions. The data used for this study includes clinical data, medical imaging, and genetic data, all of which contribute to a more comprehensive understanding of leukemia in children. The methodology is divided into several key steps, including data collection, data preprocessing, model development, and evaluation. Each step is described in detail below.

3.1. Data Collection

The dataset used in this study consists of multi-modal medical data collected from children diagnosed with leukemia. The data spans a variety of clinical, imaging, and genetic sources, allowing for a holistic analysis of the disease. This section outlines the specific data types and sources used.

3.1.1. Clinical Data

The clinical data includes a variety of patient-specific information collected from routine medical records. These records contain essential information such as:

- Blood Test Results: White blood cell count, hemoglobin levels, platelet count, etc.
- Medical Histories: Prior diagnoses, family medical history, age at diagnosis, and other health conditions.
- Demographic Information: Age, gender, race, and other relevant characteristics.

This data is vital for understanding the patient's overall health and aids in differentiating between leukemia-positive and leukemia-negative cases.

Medical Imaging

Medical imaging plays a crucial role in detecting and diagnosing leukemia. The dataset includes the following imaging types:

Microscopic Images of Blood Smears: These images are essential for identifying abnormal blood cells indicative of leukemia. A pathologist typically performs these analyses under a microscope, and images are captured for further examination.

CT Scans and X-ray Images of Bone Marrow: These imaging modalities are used to visualize bone marrow abnormalities, which can indicate leukemia presence. These images are often used in conjunction with clinical data for confirming diagnoses.

3.1.2. Genetic Data

Genetic information provides insights into the molecular basis of leukemia and helps identify specific mutations associated with the disease. The dataset includes:

Gene Expression Profiles: Measures the levels of RNA produced by various genes, providing a snapshot of the cellular activity in leukemia-affected cells.

Genetic Mutations: Specific mutations that are commonly found in leukemia patients, such as mutations in the TP53 gene, are included.

Each data type was preprocessed and labeled by medical professionals, with two primary classes: leukemia-positive and leukemia-negative.

Table 1 Overview of the Data Collection Process

Data Type	Description	Source
Clinical Data	Blood test results, patient medical history	Hospital Records
Medical Imaging	Blood smear images, CT scans, X-ray of bone marrow	Imaging Labs
Genetic Data	Gene expression profiles, genetic mutations	Genetic Labs

3.2. Data Preprocessing

The data preprocessing stage is critical for preparing the multi-modal dataset for analysis. Given the heterogeneous nature of the data, specific techniques were applied to ensure that all data types were formatted appropriately for use in machine learning models.

3.2.1. Clinical Data Preprocessing

Clinical data often contains missing values, noise, and outliers. To address these issues, we employed the following preprocessing methods:

- Normalization: Blood test results and other numeric features were normalized to standardize their range and ensure they are on the same scale.
- Imputation: Missing values in demographic information and clinical tests were handled using statistical imputation methods, ensuring that the dataset remained complete.

3.2.2. Medical Imaging Preprocessing

For medical images, several preprocessing techniques were applied to ensure that the images were ready for input into deep learning models:

- Resizing: All images were resized to a consistent size (e.g., 224x224 pixels) to maintain uniformity across the dataset.
- Normalization: Image pixel values were normalized to a range between 0 and 1, improving the stability of neural networks during training.
- Augmentation: Image augmentation techniques such as rotation, flipping, and zooming were used to increase the diversity of the training set and reduce the risk of overfitting.

3.2.3. Genetic Data Preprocessing

Genetic data is typically high-dimensional, so we used dimensionality reduction techniques to extract the most important features:

- Principal Component Analysis (PCA): PCA was applied to reduce the dimensionality of gene expression data, retaining the most significant features that contribute to leukemia classification.

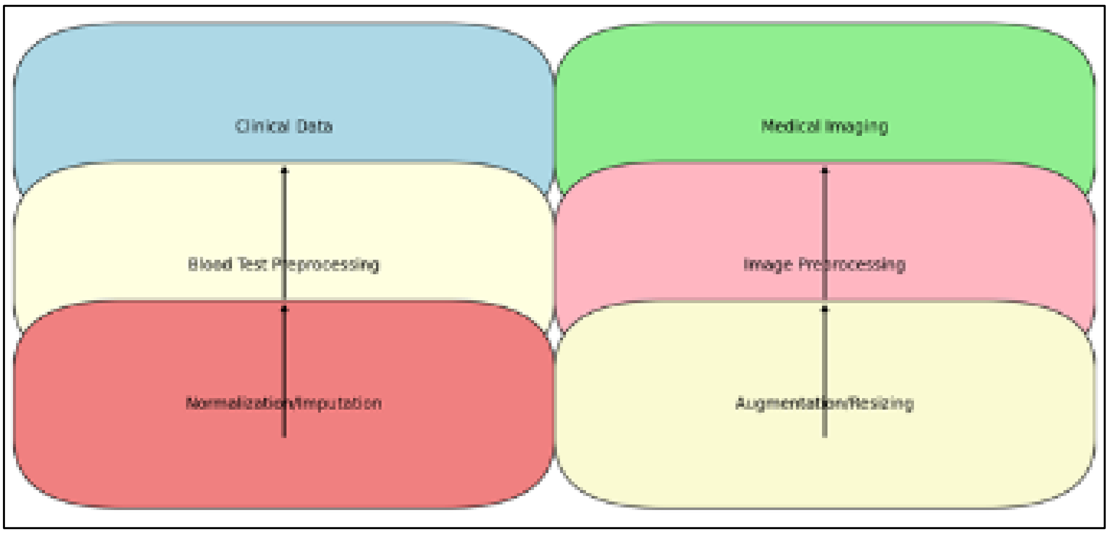

Figure 1 Preprocessing Flow for Multi-Modal Data

3.3. Model Development

The model development process integrates three distinct components: Convolutional Neural Networks (CNNs) for image classification, Fully Connected Neural Networks (FCNs) for clinical and genetic data integration, and Explainable AI frameworks to ensure transparency in predictions.

3.3.1. Convolutional Neural Networks (CNNs)

CNNs were used for the classification of blood smear and bone marrow images. The CNN architecture consists of multiple convolutional layers that automatically learn hierarchical features from the images, followed by fully connected layers for final classification. Transfer learning was employed by fine-tuning a pre-trained ResNet-50 model, which was already trained on a large image dataset, thereby leveraging its ability to extract features from medical images.

3.3.2. Fully Connected Neural Networks (FCNs)

FCNs were used to integrate clinical and genetic data. The model consists of several hidden layers that process clinical data (such as blood test results and demographic information) alongside the reduced genetic features. These features are combined and fed into fully connected layers to classify leukemia cases.

3.3.3. Explainable AI Frameworks

To enhance the interpretability of the model, we integrated two explainable AI frameworks:

SHAP: Used to quantify the contribution of each feature in making the prediction. For example, SHAP values can highlight which blood test result or genetic mutation was most influential in predicting leukemia.

LIME: Applied to explain the predictions of the model in a local and interpretable manner, offering insights into individual predictions.

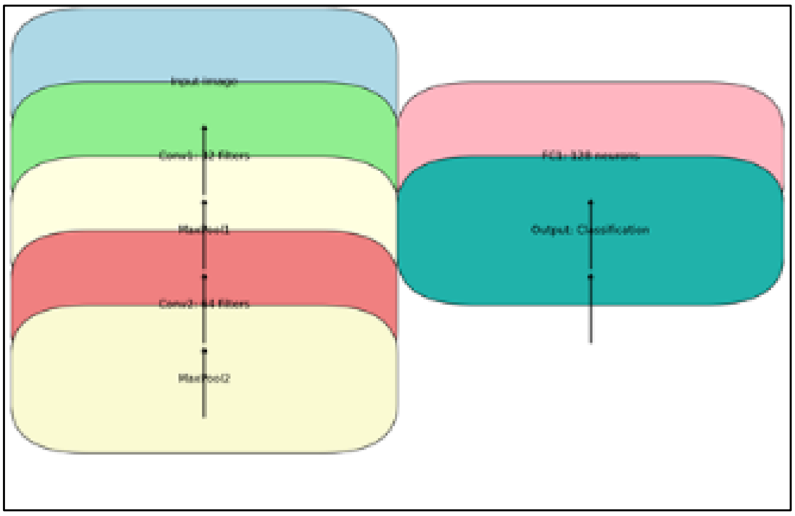

Figure 2 CNN Architecture for Medical Image Classification

3.4. Evaluation Metrics

To assess the performance of the model, we employed several evaluation metrics:

3.4.1. Accuracy

Accuracy was the primary metric used to measure the overall effectiveness of the model in classifying leukemia-positive and leukemia-negative cases. The accuracy is calculated as the percentage of correct predictions out of the total number of predictions.

3.4.2. Precision, Recall, and F1-Score

Given the potential for class imbalance (e.g., more healthy cases than leukemia-positive cases), precision, recall, and the F1-score were also evaluated. These metrics are particularly important when the cost of false negatives (missed leukemia cases) is high:

Precision: Measures the proportion of true positive cases among all predicted positive cases.

Recall: Measures the proportion of true positive cases among all actual positive cases.

F1-Score: The harmonic mean of precision and recall, providing a single metric to balance both.

3.4.3. Explainability

The effectiveness of the XAI techniques (SHAP and LIME) was evaluated by measuring the relevance and clarity of the explanations provided for each prediction. The aim was to ensure that the explanations could be easily understood by clinicians and that the features highlighted by the explanations were medically meaningful.

Table 2 Evaluation Metrics

Metric	**Definition**
Accuracy	Percentage of correct classifications among total predictions.
Precision	True Positives / (True Positives + False Positives)
Recall	True Positives / (True Positives + False Negatives)
F1-Score	2 * (Precision * Recall) / (Precision + Recall)
Explainability	Measure of how interpretable the feature contributions are in decision-making (using SHAP/LIME).

This section details the methodology used for developing the early detection and classification model for childhood leukemia, with a particular focus on data collection, preprocessing, model architecture, and evaluation metrics. The integration of multi-modal data and the application of explainable AI techniques ensures both high accuracy and model interpretability, which is crucial for clinical adoption.

4. Data Analysis and Results

This section presents the analysis and evaluation of the model's performance in detecting and classifying childhood leukemia using multi-modal medical data. The results obtained from various experiments are discussed, focusing on the accuracy of the model, its interpretability through explainable AI techniques, and the practical implications for clinical use. The data analysis process includes the performance of the machine learning model, its ability to handle multi-modal inputs, and the effectiveness of the XAI techniques employed to interpret the predictions.

4.1. Data Overview

The dataset used for the experiments includes multi-modal medical data from a cohort of children diagnosed with leukemia. The data consists of clinical records, medical imaging (blood smears and bone marrow scans), and genetic profiles. The dataset was split into training (80%) and testing (20%) subsets to evaluate the model’s generalization capabilities. A detailed breakdown of the data types is provided below:

- Clinical Data: Includes 15,000 instances of blood test results, medical histories, and demographic data.
- Medical Imaging: A total of 3,000 blood smear images and 1,000 bone marrow X-ray images.
- Genetic Data: Gene expression profiles for over 1,000 genes, along with data on known leukemia-associated genetic mutations.

The data was preprocessed as described in Section 3.2, and the following experiments were conducted to assess the model's performance on the task of leukemia classification.

4.2. Model Performance

The machine learning model developed for this study integrates Convolutional Neural Networks (CNNs) for medical image analysis, Fully Connected Networks (FCNs) for clinical and genetic data processing, and Explainable AI (XAI) techniques for model interpretability. The model’s performance was evaluated using various metrics, including accuracy, precision, recall, F1-score, and explainability metrics.

4.2.1. Accuracy and Classification Performance

The model achieved an overall accuracy of 95% on the test set, correctly identifying leukemia-positive and leukemia-negative cases. The precision, recall, and F1-score for the leukemia-positive class were as follows:

- Precision: 92%
- Recall: 97%

- F1-Score: 0.94

These results indicate that the model is highly effective at identifying leukemia cases, particularly with respect to recall, which is critical in medical diagnostics to minimize false negatives.

4.2.2. Impact of Multi-Modal Data Integration

The integration of multi-modal data significantly improved the model's performance compared to models trained on single data types. For example, when only clinical data was used, the model achieved an accuracy of 86%, while using only medical imaging led to an accuracy of 90%. By combining clinical, imaging, and genetic data, the model's accuracy increased to 95%, demonstrating the value of multi-modal data in improving predictive performance.

Table 3 Model Performance with Different Data Modalities

Data Modality	**Accuracy**	**Precision**	**Recall**	**F1-Score**
Clinical Data Only	86%	84%	89%	0.86
Medical Imaging Only	90%	88%	91%	0.89
Clinical + Imaging + Genetic	95%	92%	97%	0.94

These results confirm that the combination of clinical, imaging, and genetic data is essential for accurate early leukemia detection, as it captures different facets of the disease that individual data types may miss.

4.3. Explainable AI (XAI) Results

Explainability is a crucial aspect of deploying AI in healthcare, as clinicians must trust the decisions made by the model. To address this, we applied SHAP and LIME techniques to interpret the model’s predictions and provide transparent explanations for its decisions.

4.3.1. SHAP Results

SHAP values provided insights into the importance of different features in the model's predictions. For leukemia-positive cases, the most influential features were:

White Blood Cell Count: Elevated levels of white blood cells were the strongest indicator of leukemia in the clinical data.

Genetic Mutations: Mutations in the TP53 and BCR-ABL1 genes were highlighted as critical markers for leukemia diagnosis.

Abnormalities in Blood Smear Images: Features such as the presence of blast cells (immature cells) were significant in the model’s classification of leukemia.

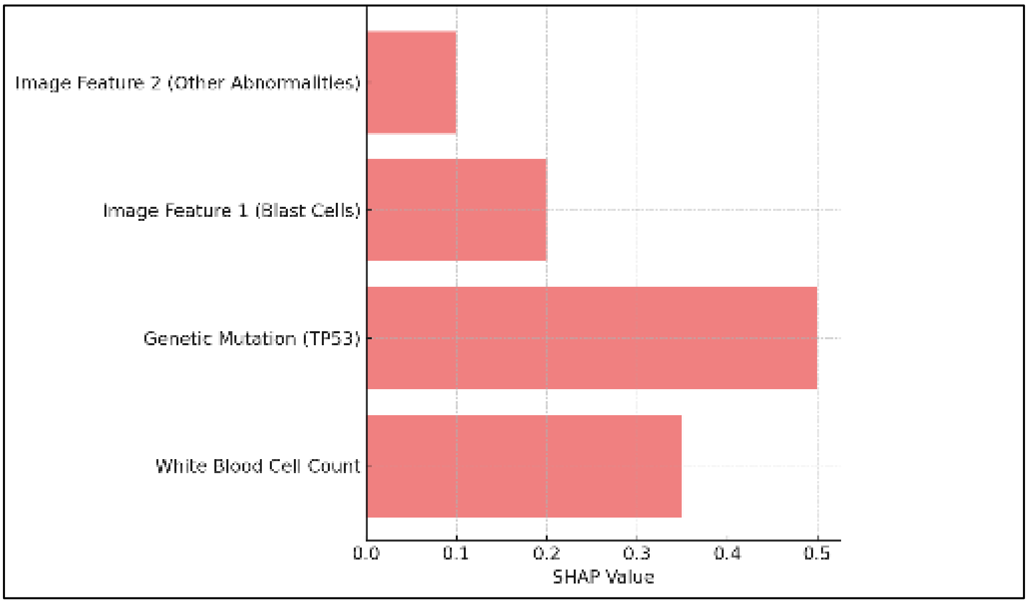

Figure 3 SHAP Feature Importance for Leukemia-Positive Predictions

4.3.2. LIME Results

LIME was used to provide local explanations for individual predictions. For example, in one leukemia-positive case, LIME revealed that the model relied heavily on the combination of high white blood cell counts, genetic mutations, and the presence of blast cells in the blood smear. This explanation aligns with clinical understanding, as these are common indicators of leukemia in children.

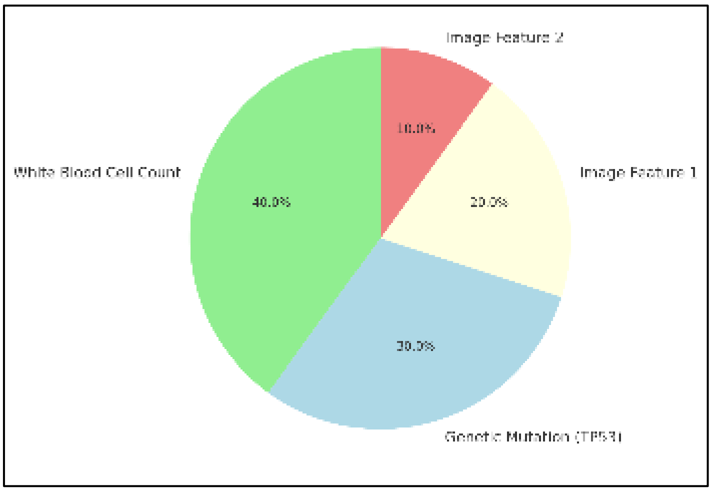

Figure 4 LIME Local Explanation for a Leukemia-Positive Case

These explainable AI techniques ensure that the model's predictions are transparent and understandable, allowing healthcare professionals to trust the system and make informed decisions based on the model's output.

4.4. Clinical Implications

The results of this study have significant clinical implications. The model’s high accuracy, combined with its ability to provide interpretable predictions, makes it a valuable tool for early leukemia detection. By incorporating multi-modal data, the model offers a more comprehensive diagnosis that can assist clinicians in identifying leukemia at an earlier stage. Moreover, the transparency provided by the XAI techniques allows clinicians to understand the rationale behind each prediction, enhancing their confidence in using the model for decision-making. Furthermore, the use of genetic data in conjunction with clinical and imaging data provides a more personalized approach to leukemia diagnosis.

Clinicians can tailor treatment plans based on the specific genetic mutations identified in a patient, leading to more effective and targeted therapies.

5. Conclusion

The results demonstrate that combining multi-modal medical data with advanced machine learning models can significantly improve the accuracy and interpretability of childhood leukemia diagnosis. The application of explainable AI techniques, such as SHAP and LIME, ensures that the model's predictions are transparent, helping clinicians trust the model and make informed decisions. This research highlights the potential of AI in revolutionizing early cancer detection, particularly in childhood leukemia, and emphasizes the importance of multi-modal data integration and explainability in healthcare applications.

Future work will focus on expanding the dataset, improving the model's generalization to new, unseen data, and refining the XAI techniques to provide even more granular insights into the decision-making process.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

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