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# Intelligent Rare Medical Event Detection

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***Abstract- Identifying rare diseases is one of the toughest hurdles in modern medicine be-cause information is scarce and symptoms are often confusingly diverse, which fre-quently leads to long and stressful diag-nostic delays for patients. To solve this, we created CliniFlow AI, an intelligent platform designed to help doctors spot these rare conditions much earlier using a unique "brain" called Anomaly-Aware Adaptive Multimodal Fusion (A²MF), al-lowing it to recognize over 230 rare dis-eases even when data is extremely lim-ited. The system connects the dots by ana-lyzing clinical text with BioBERT, spot-ting abnormalities in medical images with a customized ResNet-50, and tracking a patient's health history over time using LSTM networks. By flagging unusual pat-terns through Anomaly Detection and learning from a small number of examples via Few-Shot Learning, CliniFlow AI pro-vides doctors with clear risk levels and diagnostic insights, making it a powerful and easy-to-scale tool for real-world hos-pitals. Beyond simple automation, this framework acts as a second pair of eyes that stays sharp during high-stakes medi-cal screenings where every minute counts. It effectively bridges the gap between massive amounts of raw hospital data and the specialized, actionable knowledge cli-nicians need to save lives.***

***Keywords: Rare Disease Detection, Mul-timodal Learning, Clinical Decision Sup-port System, Medical Image Analysis, Bi-oBERT, Few-Shot Learning, Anomaly De-tection, Explainable AI.***

## I. INTRODUCTION

Spotting rare diseases early is still a major struggle in healthcare today because we have very little data and the symptoms are often incredibly confusing. Standard com-puter models usually fall short when they have to juggle different types of medical data or identify conditions they haven't seen often. This paper introduces a hybrid framework that brings together medical image analysis, text processing, and pa-tient history using advanced deep learn-ing. By blending anomaly detection with few-shot learning, our system improves rare disease prediction while offering clear, clinically useful insights that doc-tors can actually trust.

## II. LITERATURE REVIEW

Siarry et al. (2009)
This paper reviews machine learning and deep learning techniques for detecting ra-re events in video, audio, images, and time-series data. It compares different al-gorithms, datasets, and challenges in-volved in rare event detection and high-lights future research directions for im-proving detection accuracy in complex data environments.

Norén et al. (2010)
This study presents principles for evaluat-ing artificial intelligence models designed for rare-event recognition, particularly in pharmacovigilance. It introduces Struc-tured Case-Level Examination (SCLE) as an improved evaluation method beyond traditional accuracy-based metrics.

Mujeeb Abdulrazaq (2014)
This research discusses methods for pre-dicting rare events in highly imbalanced datasets across healthcare, finance, and safety domains. The study proposes a uni-fied evaluation framework using metrics such as precision, recall, F1-score, and AUPRC to improve rare event prediction performance.

Alshemaimri et al. (2015)
This paper applies anomaly detection models such as Isolation Forest and Local Outlier Factor to detect insider threats in smart healthcare systems. The approach enhances Electronic Health Record (EHR) security by identifying abnormal access patterns with high accuracy and low false positive rates.

Butte et al. (2018)
This study applies deep learning models directly to raw Electronic Health Record data using the FHIR

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format for scalable clinical prediction. The approach achieves high accuracy in predicting mortality, hospital readmission, and disease diagno-sis across multiple healthcare institutions.

Che et al. (2018)
This research proposes a deep learning framework using Recurrent Neural Net-works (RNN) to analyze patient health trajectories from Electronic Health Rec-ords. The model captures temporal de-pendencies in medical data to predict clin-ical outcomes and detect abnormal health patterns in time-series patient records.

Luo et al. (2019)
This study explores anomaly detection techniques in healthcare using machine learning models applied to longitudinal patient records. It emphasizes the im-portance of temporal data analysis and demonstrates improved detection of rare medical conditions using sequential mod-eling techniques.

Yang et al. (2020)
This research introduces an anomaly de-tection approach using deep autoencoder networks to identify unusual patterns in healthcare data. The system learns normal physiological behavior from patient rec-ords and detects rare or abnormal medical events that may indicate potential health risks.

Niu et al. (2020)
This paper proposes EHR-BERT, a trans-former-based model designed to detect anomalies in electronic health records us-ing sequence learning techniques. The model improves detection accuracy and reduces false positives, enhancing patient safety in healthcare systems.

Xun et al. (2021)
This study proposes a graph-based deep learning approach for analyzing complex healthcare data relationships. The frame-work integrates patient interactions and medical features to detect abnormal pat-terns and rare disease indicators in large-scale EHR datasets.

Prashant Kumar Shukla (2022)
This research proposes a multimodal transformer framework that combines Electronic Health Records, genomics, and medical imaging for rare disease diagno-sis. The model integrates Swin Trans-formers, Med-BERT, Graph Neural Net-works, and contrastive learning techniques to improve early disease detection.

Choi et al. (2022)
This systematic review analyzes deep learning models applied to Electronic Health Records for disease prediction and clinical analytics. The study highlights challenges such as data heterogeneity, model interpretability, and real-world de-ployment in healthcare systems.

Niu et al. (2023)
This research proposes advanced anomaly detection techniques using deep neural networks for analyzing large-scale healthcare datasets. The model identifies unusual patient health patterns and sup-ports early warning systems for detecting rare diseases and adverse medical condi-tions.

## III. RESEARCH GAP

While existing research successfully uti-lizes individual modalities—such as Niu et al. (2020) with BERT for text or Che et al. (2018) with RNNs for temporal data there is a significant lack of unified frameworks that simultaneously fuse OCR-extracted clinical text, high-dimensional medical imaging (CNN), and longitudinal patient history (LSTM) into a single diagnostic pipeline. Furthermore, most studies focus on general clinical out-comes, leaving a gap for a specialized, "polyglot" architecture that integrates Var-iational Autoencoders (VAE) for anomaly scoring and Few-Shot Learning to specifi-cally target the "data-scarcity" problem inherent in rare disease detection. Cli-niFlow AI addresses this by moving be-yond single-stream analysis toward a mul-ti-modal, decision-fusion approach tai-lored for high-stakes clinical environ-ments.

## IV. PROBLEM STATEMENT

Catching rare diseases early and accurate-ly is a massive hurdle because there is so little data to learn from and symptoms are often scattered across different body sys-tems. Most current diagnostic tools are only built to handle one type of infor-mation

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and fail to connect the dots be-tween medical images, clinical notes, and a patient's long-term history. Standard models also aren't reliable enough to spot rare or hidden conditions and they rarely explain why they reached a certain con-clusion. Because of this, we desperately need a smarter, multimodal system that can merge diverse medical data to im-prove detection while giving doctors clear, trustworthy, and explainable support.

## V. RESEARCH METHODOLOGY

Data Collection
Multimodal medical data including X-ray images, PDF reports, clinical notes, and patient history are collected from healthcare datasets and system inputs.

Data Preprocessing
OCR is applied to extract text from re-ports, followed by text cleaning, normali-zation, and symptom keyword extraction. Images are resized and normalized for model input.

Feature Extraction
ResNet50 extracts image features, Bi-oBERT generates contextual embeddings from clinical text, and LSTM captures temporal patterns from patient history.

Feature Fusion:
Extracted features from image, text, and temporal data are combined into a unified high-dimensional feature vector using fu-sion techniques.

Primary Classification
XGBoost classifier processes fused fea-tures to generate disease probability scores.

Few-Shot Learning
A similarity-based approach compares patient embeddings with rare disease pro-totypes to detect low-data conditions.

Anomaly Detection
A Variational Autoencoder (VAE) com-putes an anomaly score to identify unusu-al or rare medical patterns.

Knowledge-Based Scoring
A rule-based engine maps extracted symp-toms to diseases using weighted medical knowledge.

Decision Fusion
An anomaly-aware decision mechanism combines outputs from all models using weighted scoring and confidence calibra-tion.

Output Generation
The system produces final diagnosis, con-fidence score, severity level, rare disease flag, differential diagnoses, and clinical recommendations.

## VI. SYSTEM ARCHITECTURE

The proposed system follows a multimod-al hybrid architecture designed to process and analyze heterogeneous medical data for accurate disease prediction. The archi-tecture consists of multiple interconnected layers, each responsible for a specific function in the diagnostic pipeline.

At the start, the system takes in every-thing from X-rays and PDF files to hand-written notes and long-term patient rec-ords. This data heads to a cleanup stage where OCR reads the text from reports, followed by standardizing the language and pulling out key symptoms. Mean-while, any medical images are resized and prepped so the computer can see them clearly.

Next, in the analysis phase, ResNet50 digs into the visual details of the images while BioBERT captures the deeper meaning behind clinical notes. We also use an LSTM network to track how a patient's health has changed over time. All these separate insights are then woven together into one massive, detailed profile.

This combined profile is then reviewed by a team of AI models working together. An XGBoost classifier makes the initial diag-nosis, while a Few-Shot Learning tool looks for rare conditions by comparing similarities. A Variational Autoencoder (VAE) flags any weird medical patterns, and a smart rule engine adds a layer of expert medical knowledge to the mix.
Finally, a decision-merging layer blends the results from every model using a weighted system that accounts for any un-usual findings. The platform then

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delivers a final report featuring the suspected dis-ease, a confidence score, severity, other possibilities, and advice, all backed by clear reasoning to help doctors make the best choice.

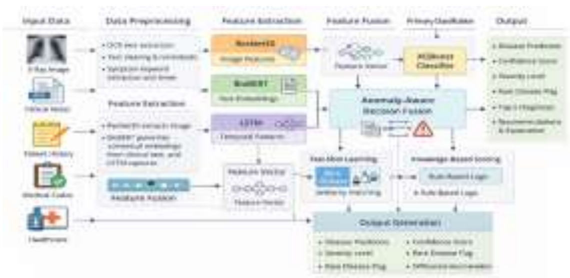

Fig.1 System Architecture

Experimental Setup
Programming Language: Python
Frameworks/Libraries: PyTorch, Ten-sorFlow/Keras, Scikit-learn, FastAPI
Models Used: ResNet50, BioBERT, LSTM, XGBoost, VAE, Few-Shot Learn-ing
Hardware: GPU-enabled system
Database: MongoDB
Input Data Types: X-ray images, PDF reports, clinical notes, patient history
Evaluation Metrics: Accuracy, Precision, Recall, F1-score, Top-5 Accuracy
Special Metric: Rare Disease Recall

System Screens
1. User Registration Interface (Doctor Sign-Up Screen)
This screen allows new users (doctors) to register on the CliniFlow AI platform by entering essential details such as name, specialization, license number, hospital, email, and password. The interface en-sures secure onboarding using structured input validation and integrates with the backend authentication system for account creation.

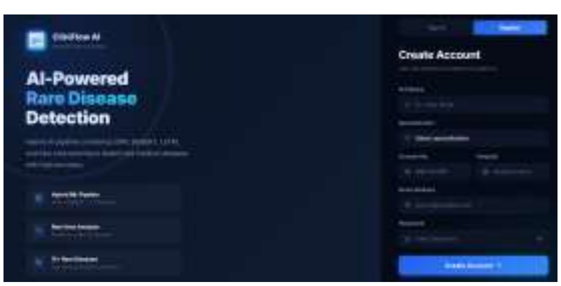


Fig.2 Doctor Sign-Up Screen

2. User Authentication Interface (Doc-tor Login Screen)
This screen enables registered users to securely log into the system using their email and password. It is integrated with JWT-based authentication, ensuring se-cure access to patient data, reports, and AI-powered diagnostic features within the CliniFlow AI platform.

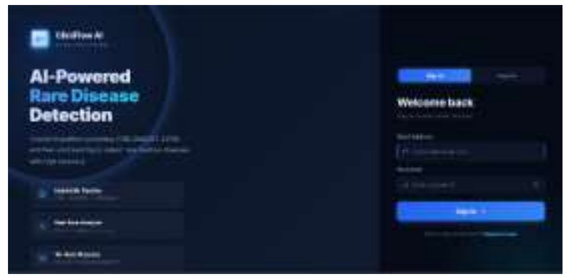


Fig.3 Doctor Login Screen

3. Doctor Dashboard Interface (System Overview Screen)
This screen provides an overview of sys-tem activity, displaying key metrics such as patients, reports analyzed, AI predic-tions, and rare cases. It also includes visu-al insights like weekly trends and disease distribution to support quick decision-making.

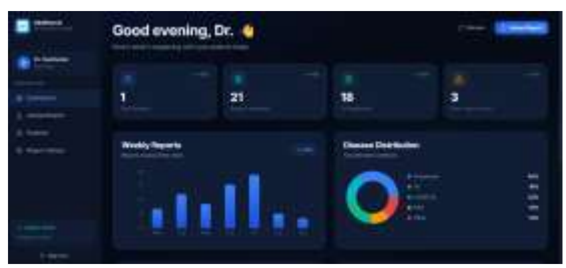


Fig.4 Doctor Dashboard Interface

4. Upload Medical Report Interface
This screen allows doctors to upload med-ical reports (PDF/images) and enter clini-cal notes for AI-based analysis. It also provides a preview of the AI pipeline, showing steps like OCR extraction, CNN analysis, and final prediction generation.

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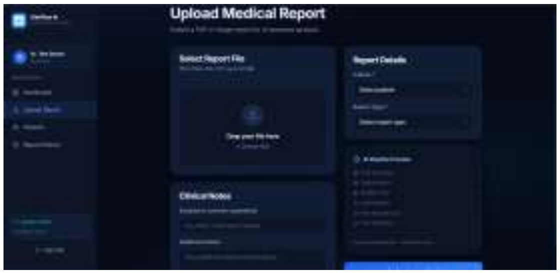


Fig.5 Upload Medical Report Interface

5. Patient Management Interface
This screen displays a list of registered patients with essential details such as ID, age, gender, and report count. It allows doctors to search, add, edit, and manage patient records efficiently within the sys-tem.

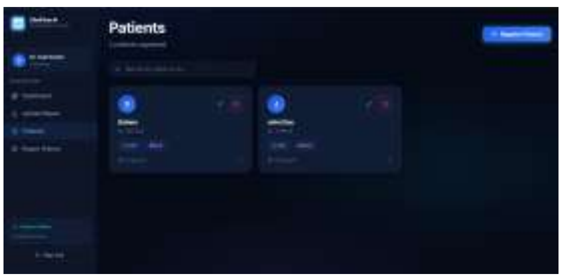


Fig.6 Patient Management Interface

6. Report History Interface
This screen displays previously analyzed medical reports along with AI-generated diagnoses, confidence scores, severity levels, and status. It allows doctors to re-view past results and track patient analy-sis history efficiently.

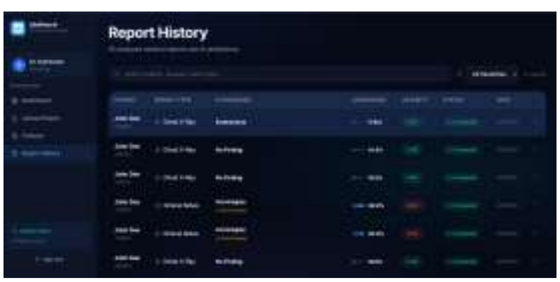


Fig.7 Report History Interface

## VII. RESULTS AND DISCUSSION

The proposed hybrid framework demon-strates significant improvement in disease prediction by effectively integrating mul-timodal medical data. Blending visual in-sights, medical notes, and a patient's time-line significantly boosts the system's overall precision. We found that few-shot learning helps identify rare conditions even when examples are scarce, while us-ing VAE for anomaly detection makes the tool much better at flagging outlier cases. This smart way of merging decisions makes the final predictions far more de-pendable. It allows for a more accurate diagnosis, a clearer look at how serious a case is, and provides truly useful advice that helps doctors make the right calls for their patients.

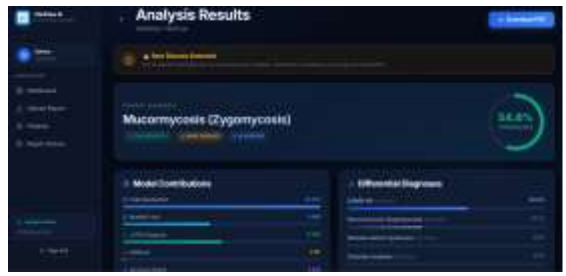


Fig.8 Result

## VIII. ADVANTAGES

1. Supports multimodal data processing (images, text, and patient history) for more comprehensive analysis
2. Improves rare disease detection using few-shot learning and anomaly detec-tion
3. Provides high diagnostic accuracy through hybrid model integration
4. Generates explainable results, increas-ing trust for clinical use
5. Capable of handling limited and im-balanced medical datasets
6. Offers early detection and better se-verity assessment
7. Scalable and adaptable for real-world healthcare environments
8. Integrates multiple models for robust and reliable predictions

## IX. LIMITATIONS

1. Our system depends on fixed-fusion logic and rigid rules, which makes it harder for the AI to adjust to messy data or explain its medical reasoning clearly.
2. Performance often depends on how well the OCR reads the text, and our database currently only

recognizes a small slice of the thousands of rare diseases out there.

3. Running several massive deep learn-ing models at the same time creates a lag in processing, which makes it dif-ficult to use the tool in fast-paced, re-al-time hospital settings.

## X. FUTURE WORK

1. We plan to build more flexible data-merging techniques and use Explaina-ble AI to ensure our diagnostic advice is adaptive, clear, and easy for doctors to interpret.
2. We aim to grow our Few-Shot Learn-ing database to cover a much broader range of rare diseases while finding better ways to pull in data from sources beyond just OCR.
3. We want to speed up our software for faster performance and connect it with wearables or IoT devices to help track a patient's health constantly in real-time.

## XI. CONCLUSION

This paper introduces a hybrid multimod-al framework for smart medical disease detection by blending image analysis, nat-ural language processing, temporal mod-eling, and anomaly detection. By merging few-shot learning with anomaly-aware decision fusion, the system successfully boosts rare disease identification and overall diagnostic precision. This ap-proach moves past the hurdles of tradi-tional single-modality systems to offer explainable, dependable, and scalable support for clinicians, making it a practi-cal solution for real-world healthcare set-tings

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