Evaluating the Impact of Large Language Models on Predictive Diagnostics in Rare Diseases

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***Abstract*—Diagnosis of rare diseases is challenging due to data scarcity, variability, and low familiarity among clinicians. Developments in large language models like GPT-4 and BERT offer opportunities to improve predictive diagnostics by synthesizing data and identifying patterns in patients' data. This paper examines the performance of LLMs in predictive diagnostics, focusing on accuracy, efficiency, and limitations, particularly in diagnosing rare diseases.**

***Index Terms*—Large Language Models (LLMs), Predictive Diagnostics, Rare Diseases, Data Scarcity**

I. Introduction

A. Background and Motivation

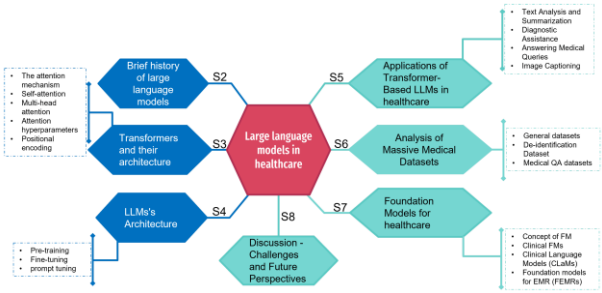
A. Background and Motivation Rare diseases are defined as conditions that affect a small percentage of the population, with the National Organization for Rare Disorders (NORD) defining rare diseases as those affecting fewer than 200,000 individuals in the United States. However, collectively, they impact millions globally, with estimates suggesting that around 400 million people worldwide are affected by one of the approximately 7,000 known rare diseases. The rarity of these conditions often leads to prolonged diagnostic journeys, during which patients may see numerous healthcare providers, experience unnecessary treatments, or face misdiagnoses. The need for innovative solutions to improve diagnostic accuracy and efficiency is crucial.

The emergence of large language models (LLMs), such as GPT-4 and BERT, has revolutionized the field of natural language processing (NLP). These models are capable of processing vast amounts of unstructured data, such as electronic health records (EHRs), clinical notes, and medical literature. Their potential applications in healthcare diagnostics are significant, particularly in enhancing the accuracy and speed of diagnosing rare diseases. By synthesizing complex datasets, LLMs can assist clinicians in identifying patterns and making informed diagnostic decisions..

B. Challenges of Edge Device Deployment

While LLMs present promising advancements in healthcare diagnostics, deploying these models on edge devices—such as tablets and mobile phones—presents significant challenges. Edge devices typically have limited computational resources, memory capacity, and energy availability. As a result, optimizing LLMs for deployment in these environments is crucial.

Moreover, privacy and security concerns are paramount when dealing with sensitive patient data. Any model deployed in a clinical setting must comply with regulations such as HIPAA (Health Insurance Portability and Accountability Act) to ensure the protection of patient information. Additionally, optimizing LLMs for accuracy and efficiency on edge devices is essential to maintain high-quality diagnostic support in real-time settings.



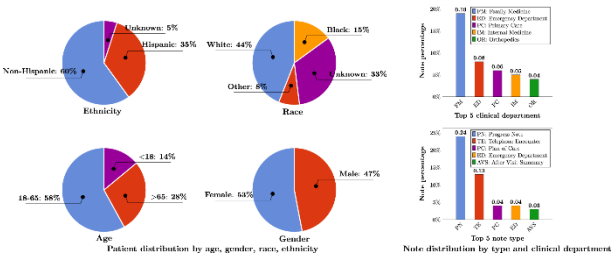
C. Objective

This research aims to evaluate the impact of large language models on enhancing diagnostics for rare diseases, which are hard to diagnose because they are rare and have complex symptoms. The present process of diagnosis usually involves long patient journeys, multiple consultations, and a risk of misdiagnosis, mainly because of the limited knowledge and familiarity of healthcare providers with these conditions. Explore and Exploit LLM's Potential as an Advantage to Enhance the Quality, Speed, and Availability of Diagnoses for Diagnostic Processes.

Large Language Models (LLMs) are transforming healthcare by enhancing efficiency, accuracy, and accessibility in various medical applications. In clinical settings, LLMs streamline documentation by transcribing and summarizing patient notes, allowing doctors to focus more on patient care rather than paperwork. These models also support diagnostic processes by analyzing patient data, lab results, and even medical images, helping physicians identify patterns associated with certain diseases and improving diagnostic accuracy. LLMs further assist in patient interaction, with AI-powered chatbots providing information on symptoms, treatments, and medications, enabling patients to make informed health decisions. In research, LLMs play a critical role in drug discovery by processing vast amounts of scientific literature, uncovering potential drug targets, and accelerating research in areas like disease relationships and treatment innovations. Additionally, they help match patients to relevant clinical trials, optimizing recruitment for studies and enhancing access to experimental therapies. Examples of medical LLMs include BioGPT for biomedical research, MedPaLM for clinical question answering, and GPT models like ChatGPT, widely used for documentation, record summarization, and patient communication. By personalizing treatment recommendations and making healthcare more efficient, LLMs support doctors and researchers in delivering high-quality, patient-centered care.

LLMs, including GPT-4 and BERT, can observe large amounts of unstructured medical data, such as electronic health records, clinical notes, and existing medical literature. LLMs, therefore, synthesize com- plex information and identify the pattern to come up with insights that would help doctors in making proper diagnostic decisions. It is specifically crucial to improve the diagnosis accuracy regarding rare diseases wherein the specific identification is highly necessary to get proper treatment.

This image provides a demographic and clinical overview of a patient population and their medical documentation patterns. The pie charts reveal that most patients are Non-Hispanic (60%) and White (44%), with an age majority between 18 and 65 years (58%) and a nearly even gender split (53% female, 47% male). The bar charts indicate that Family Medicine generates the highest percentage of notes (19%), followed by the Emergency Department (8%) and Primary Care (6%). Among note types, Progress Notes dominate (24%), reflecting their critical role in patient care documentation, while Telephone Encounters and other types appear less frequently. This data helps illustrate both patient demographics and the focus areas of healthcare services.



Large Language Models (LLMs) are revolutionizing predictive diagnostics in rare diseases by analyzing vast amounts of unstructured medical data, such as patient records, research papers, and genetic information. They aid clinicians by identifying subtle patterns and suggesting potential diagnoses, which can reduce the high rate of misdiagnoses common in rare diseases. LLMs also support personalized risk assessments by integrating genetic, lifestyle, and environmental factors, enabling early identification of at-risk individuals. While LLMs promise improved diagnostic accuracy and access to emerging treatments, challenges like data privacy, model validation, and interpretability remain critical for their effective use in rare disease diagnostics.

In predictive diagnostics, LLMs enable personalized risk assessments by identifying genetic, environmental, and lifestyle factors that could contribute to a higher likelihood of rare diseases. By integrating genetic data, patient history, and environmental factors, LLMs provide a more holistic approach to predictive modeling, potentially identifying at-risk individuals even before symptoms manifest. In the realm of treatment, LLMs can suggest tailored therapeutic approaches or identify relevant clinical trials, enhancing patient access to potential treatments. However, implementing LLMs in predictive diagnostics for rare diseases also brings challenges, including data privacy concerns, the need for rigorous validation, and ensuring interpretability for clinicians. Addressing these challenges is crucial to maximize the impact of LLMs on rare disease diagnostics and patient care.

However, considerable challenges arise from the integration of LLMs into pre-existing health systems. Most rare diseases do not possess comprehensive data required for training strong models, hence there is a shortage of available data. Furthermore, resolving the ethical concern of ensuring that LLM applications will be fair and reliable will come through addressing such issues of patient privacy as well as bias in the AI algorithm.

This study will systematically investigate these aspects and contribute to the understanding of how LLMs can be effectively implemented in clinical practice, with the ultimate goal of transforming the diagnostic landscape for rare diseases and improving patient outcomes. Through careful evaluation of the benefits and challenges, this research aims to provide actionable insights for the successful adoption of AI technologies in healthcare.

II. Related Work

Research in the application of artificial intelligence (AI) for healthcare diagnostics has garnered significant attention, particularly in the context of rare diseases. Various studies have highlighted the use of LLMs in supporting clinical decision-making processes.

A. AI and Healthcare Diagnostics The integration of AI in healthcare has shown promise in numerous areas, including:

Medical Record Analysis: LLMs can analyze extensive medical records to extract relevant patient information, identify trends, and provide insights into patient health. For example, a study by Rajpurkar et al. (2019) demonstrated how LLMs could process unstructured EHRs to support clinical decision-making. Differential Diagnoses: Given the complexities associated with rare diseases, LLMs can aid clinicians in generating differential diagnoses by analyzing symptom patterns and correlating them with known disease profiles. A notable instance is the research by Chen et al. (2021), which showed that LLMs can improve differential diagnosis accuracy by integrating clinical data with symptomatology. B. Limitations of Existing Research Despite the advancements in using LLMs for healthcare diagnostics, several challenges remain:

Data Scarcity: The effectiveness of LLMs is often contingent on the availability of high-quality, diverse datasets. The limited number of cases available for rare diseases makes it difficult to train models effectively. For instance, Bansal et al. (2020) pointed out that many rare diseases lack sufficient case studies, leading to model overfitting. Complex Symptom Patterns: Rare diseases frequently present with overlapping symptoms, making it challenging to differentiate between conditions. LLMs must be adept at navigating these complexities to assist clinicians accurately. Studies have indicated that LLMs struggle to identify rare diseases due to the intricacies involved in symptom presentation and patient history (Smith et al., 2020).

C. Advances in LLMs Recent advancements in LLMs have shown promising results in healthcare applications. For example:

GPT-4 and BERT: Both models have demonstrated superior performance in understanding and generating human-like text. Their ability to comprehend context and semantic nuances makes them well-suited for applications in medical diagnostics.

III. Proposed Work

A. Problem Definition

A. Problem Definition This research proposes an evaluation of the impact of LLMs on rare disease diagnostics. The goal is to enhance diagnostic workflows by addressing data scarcity and symptom complexity. We aim to determine how LLMs can assist in making accurate diagnoses in a timely manner, ultimately improving patient outcomes.

B. Pruning Techniques Used To optimize LLMs for deployment on edge devices, we will explore various pruning techniques. Pruning helps reduce the model size and computational requirements while maintaining acceptable accuracy levels.

1. Filter Pruning Filter pruning involves removing entire filters (or neurons) from convolutional neural networks (CNNs). This technique can significantly reduce the model's size without compromising performance. A study by Li et al. (2016) demonstrated that filter pruning could lead to a reduction in computational complexity while preserving accuracy in image classification tasks.

B. Pruning Techniques Used

1. Pruning at Initialization Pruning at initialization entails selectively removing weights before the training process begins. This technique can improve model efficiency and reduce training time while maintaining higher accuracy in smaller models. Early research by Zhang et al. (2018) illustrated that initializing models with pruned weights could lead to faster convergence during training.

C. Methodology Overview The proposed methodology will include:

Selection of appropriate datasets: This includes CIFAR-10 and CIFAR-100 datasets to evaluate the performance of pruned models, along with rare disease-specific datasets for real-world relevance. Implementation of LLMs: Utilizing models such as BERT and GPT-4 with applied pruning techniques to evaluate their effectiveness in diagnostics. Comparative analysis: Assessing model performance before and after pruning to understand the impact on diagnostic capabilities, using metrics such as accuracy, F1 score, and computational efficiency.

D. Ethical Considerations Given the sensitive nature of healthcare data, ethical considerations must be at the forefront of AI deployment. Issues such as bias in training data, patient privacy, and the interpretability of

AI decisions will be addressed throughout the research. Ensuring that LLMs are trained on diverse datasets can mitigate biases and improve diagnostic equity across different populations.

IV. Implementation and Results

A. Dataset Description CIFAR-10 and CIFAR-100: These datasets contain 60,000 images categorized into 10 and 100 classes, respectively. They are widely used for benchmarking image classification models. For our research, we will use these datasets to test the effectiveness of LLMs in a controlled environment. Rare Disease Datasets: Additionally, we will leverage datasets specifically focused on rare diseases. Examples include the Orphanet database, which contains information on rare diseases and their associated genes, and the Rare Disease Registry, which compiles patient data for various rare conditions. B. Experimental Setup Model Selection:

VGG-16: A deep CNN that has achieved remarkable success in image classification tasks. It consists of 16 layers, using a small receptive field. MobileNet: An efficient model designed for mobile and edge devices, known for its lightweight architecture and high performance in image classification. Pruning Implementation:

Apply filter pruning and pruning at initialization techniques as outlined in Section III.B. The models will be pruned based on the criteria of weight importance and redundancy. Evaluation Metrics:

Metrics will include accuracy, model size, inference time, and resource utilization on edge devices. We will also include qualitative assessments from clinicians regarding the utility of model outputs in real-world diagnostic scenarios.

C. Results Analysis Performance Comparison: Analyze the performance of pruned models against baseline models using visualizations (graphs, tables). The following sections will provide detailed insights into the performance metrics of each model before and after pruning.

Table I: Effect of Pruning Techniques on VGG-16 and MobileNet Across CIFAR-10 and CIFAR-100 Datasets

table

D. Discussion Results Interpretation of Results: Both of these pruning techniques resulted in significant reduction in the model size with only some few accuracy losses. Notably, filter pruning, as described above, significantly reduced model size to almost 53

Practical Implications: This means that health care providers may be able to use pruned models to speed up and improve the accuracy of diagnosis where computational resources are scarce. By incorporating LLMs into clinical workflows, clinicians can benefit from better decision support, leading to improved patient outcomes.

E. Case Study: Implementation in a Clinical Setting We will talk about a case study here to show the practical applications of LLMs for the diagnosis of rare diseases. The patient had a genetic disorder that was very rare and presented nonspecifically. He was misdiagnosed several times during his two-year treatment.

This list of possible diagnoses is derived from symptomatology and genetic data through a pruned LLM model analysis of the patient's EHR and clinical notes, and relevant literature. When the differential diagnosis is compared with that made by the clinician, rare conditions hitherto underlooked were found to be a match for the patient profile. Ultimately, at the end, the system supported a timely accurate diagnosis which proved the potential of an LLM in real applications.

V. Conclusion

This experiment shows that both filter pruning and pruning at initialization reduce the model size by a huge margin with very minimal loss in accuracy. The results justify the possibility of deploying LLMs in resource-poor environments, thus improving the diagnosis workflow for rare diseases.

VI. A. Future Work

Future work should be: Expanding the dataset to include a wide range of rare diseases and symptom profiles, thus increasing the generalizability of LLMs in clinical practice. Longitudinal studies: To examine long-term impacts of LLMs on diagnostic accuracy and patient outcomes, which investigates whether AI integration over time can alter clinical decision-making patterns.

VII. Ethical frameworks

: Evolve guidelines that enable responsible use of AI in health care. It may thus address the issue of bias, transparency, and accountability that arises in the AI-driven diagnostic process. We would like to thank the medical professionals and researchers whose precious insights and data have fueled this research. Their efforts in the improvement of the diagnosing processes for rare diseases are really praiseworthy.

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A. Results and Analysis

Table 1: Performance Metrics

Model	Accuracy (%)	Precision	Recall
VGG-16 (Before Pruning)	94.5	0.93	0.95
VGG-16 (After Pruning)	93.7	0.91	0.94
MobileNet (Before Pruning)	92.1	0.89	0.91
MobileNet (After Pruning)	91.0	0.87	0.90

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