

Generative AI for Medical Diagnosis

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Abstract - Generative Artificial Intelligence (GenAI) has emerged as a transformative paradigm in medical diagnosis, offering novel approaches to data synthesis, disease detection, and clinical decision support. Unlike traditional machine learning methods, generative models such as Generative Adversarial Networks (GANs), Variational Autoencoders (VAEs), diffusion models, and large language models (LLMs) can learn complex data distributions and generate realistic medical data, including images, clinical reports, and synthetic patient records. Recent advancements between 2020 and 2026 demonstrate that GenAI enhances diagnostic accuracy, improves data augmentation, and supports multimodal analysis integrating imaging, genomic, and clinical data. Studies indicate that generative AI contributes to improved diagnostic workflows, particularly in radiology, cardiology, and oncology, where data scarcity and imbalance are significant challenges. Furthermore, generative models facilitate early disease detection and predictive diagnostics by modeling disease progression patterns. Despite its advantages, challenges such as ethical concerns, data privacy, model interpretability, and clinical validation remain critical barriers to widespread adoption. Meta-analyses reveal that while generative AI demonstrates promising diagnostic capabilities, it has not yet consistently surpassed expert clinicians, emphasizing the need for hybrid human-AI systems. This paper provides a comprehensive overview of generative AI in medical diagnosis, covering its introduction, literature survey, problem definition, methodology, and future implications. The study highlights the potential of generative AI to revolutionize healthcare by enabling personalized medicine, reducing diagnostic errors, and improving healthcare accessibility, while also addressing the limitations that must be overcome for safe and effective deployment.

Key Words: Generative Artificial Intelligence, Medical Diagnosis, GANs, Deep Learning, Healthcare Analytics, Synthetic Data, Disease Detection

1. INTRODUCTION

The integration of Artificial Intelligence (AI) into healthcare has significantly transformed diagnostic processes, enabling faster, more accurate, and data-driven decision-making. Among the various AI paradigms, Generative Artificial Intelligence has gained substantial attention due to its ability to generate new data samples that closely resemble real-world medical data. Generative AI encompasses advanced models such as GANs, VAEs, diffusion models, and transformer-based architectures that learn underlying data

distributions and produce realistic outputs. These capabilities are particularly beneficial in medical diagnosis, where high-quality labeled data is often limited, expensive, and sensitive.

Recent studies highlight that generative AI is revolutionizing healthcare by enhancing diagnosis, treatment planning, and patient care through advanced data modeling and synthesis techniques. The increasing adoption of multimodal AI systems, which combine imaging, textual, and genomic data, has further improved diagnostic performance by providing a comprehensive understanding of patient conditions. For instance, generative AI models can reconstruct missing medical images, simulate disease progression, and assist clinicians in interpreting complex diagnostic data.

Moreover, generative AI has demonstrated its effectiveness in addressing key challenges in medical diagnosis, such as data scarcity, class imbalance, and privacy concerns. By generating synthetic datasets, these models enable robust training of diagnostic systems without compromising patient confidentiality. According to recent findings, generative AI is widely used in radiology, cardiology, and other domains for disease detection, screening, and diagnostic support. Additionally, generative AI contributes to predictive healthcare by analyzing historical patient data to forecast disease risks and outcomes.

However, despite these advancements, the integration of generative AI into clinical practice is still in its early stages. Issues such as model interpretability, ethical considerations, and regulatory compliance pose significant challenges. Studies emphasize that while generative AI can enhance diagnostic accuracy, it should be used as a supportive tool rather than a replacement for human expertise. Therefore, there is a need for comprehensive research to explore the potential and limitations of generative AI in medical diagnosis.

2. LITERATURE SURVEY

The literature on generative AI in medical diagnosis has expanded rapidly between 2020 and 2026, reflecting the growing interest in this domain. Early studies focused on the application of GANs for medical image generation and enhancement. These models demonstrated the ability to produce high-quality synthetic images, improving the performance of diagnostic systems trained on limited datasets. Recent research highlights that GAN-based

approaches dominate the field, accounting for a significant portion of generative AI applications in medical imaging. Subsequent advancements introduced VAEs and diffusion models, which improved image quality and stability compared to GANs. These models have been widely used for anomaly detection, image reconstruction, and disease classification. For example, diffusion models have shown superior performance in generating high-resolution medical images, enabling more accurate diagnosis of conditions such as cancer and neurological disorders.

Recent systematic reviews emphasize the role of generative AI in improving diagnostic accuracy and clinical decision-making. A comprehensive analysis of 161 studies revealed that generative AI is increasingly used for disease detection, diagnosis, and screening, particularly in radiology and cardiology. Furthermore, generative AI has been applied in genomic analysis, where transformer-based models assist in identifying genetic variations and predicting disease risks. A 2025 meta-analysis evaluating generative AI models for diagnostic tasks reported an average diagnostic accuracy of 52.1%, highlighting both the potential and limitations of these systems. The study also emphasized the need for comparative analysis with human clinicians to assess real-world applicability. Similarly, recent reviews indicate that while generative AI enhances diagnostic capabilities, it has not yet achieved expert-level reliability.

In addition to diagnostic applications, generative AI has been used for clinical documentation, patient communication, and medical education. Large language models enable automated report generation and assist clinicians in decision-making processes. However, studies also highlight concerns related to data privacy, bias, and ethical implications, which must be addressed to ensure safe deployment.

3. PROBLEM DEFINITION

Medical diagnosis faces several challenges that limit the effectiveness of traditional AI and machine learning approaches. One of the primary issues is the scarcity of high-quality labeled medical data, which is essential for training accurate diagnostic models. Medical datasets are often limited due to privacy concerns, high annotation costs, and the complexity of data collection processes. Additionally, class imbalance in medical datasets, where certain diseases are underrepresented, leads to biased models and reduced diagnostic accuracy.

Another significant challenge is the heterogeneity of medical data, which includes imaging, clinical records, genomic data, and sensor data. Traditional models struggle to integrate these diverse data types, resulting in incomplete analysis and suboptimal diagnostic outcomes. Furthermore, existing diagnostic systems often lack interpretability, making it difficult for clinicians to trust and adopt AI-based solutions. Generative AI addresses some of these challenges by enabling synthetic data generation, multimodal data integration, and improved feature representation. However, several issues remain unresolved. These include the risk of

generating inaccurate or misleading data, lack of standardized evaluation metrics, and limited clinical validation. Moreover, ethical concerns related to data privacy, bias, and accountability pose significant barriers to the adoption of generative AI in healthcare.

Therefore, the key problem addressed in this study is to explore how generative AI can be effectively utilized to enhance medical diagnosis while addressing the challenges of data scarcity, heterogeneity, and reliability. The objective is to develop a robust framework that leverages generative models to improve diagnostic accuracy and support clinical decision-making.

4. METHODOLOGY

The proposed methodology for implementing generative AI in medical diagnosis consists of several stages, integrating data processing, model development, and evaluation.

The first stage involves data collection from multiple sources, including medical imaging datasets such as MRI, CT scans, and X-rays, as well as electronic health records and genomic data. These datasets are preprocessed through normalization, noise reduction, and anonymization to ensure data quality and privacy.

In the second stage, feature extraction techniques are applied to identify relevant patterns in the data. Deep learning models such as convolutional neural networks (CNNs) are used for image-based features, while transformer-based models are employed for textual and sequential data.

The third stage involves the selection and training of generative models. GANs are used for image synthesis and augmentation, VAEs for anomaly detection, and diffusion models for high-quality image generation. These models learn the underlying data distribution and generate synthetic samples that enhance the training dataset.

The fourth stage focuses on model integration and multimodal analysis. Data from different sources are combined to create a unified representation, enabling comprehensive diagnostic analysis. This stage leverages multimodal AI techniques to improve the accuracy and robustness of the diagnostic system.

The fifth stage involves model evaluation using performance metrics such as accuracy, sensitivity, specificity, precision, recall, and F1-score. Cross-validation techniques are used to ensure the reliability of the model.

Finally, the system is deployed in a clinical environment, where it assists healthcare professionals in diagnosis and decision-making. Continuous monitoring and feedback mechanisms are implemented to improve model performance over time. The methodology is given in figure 1

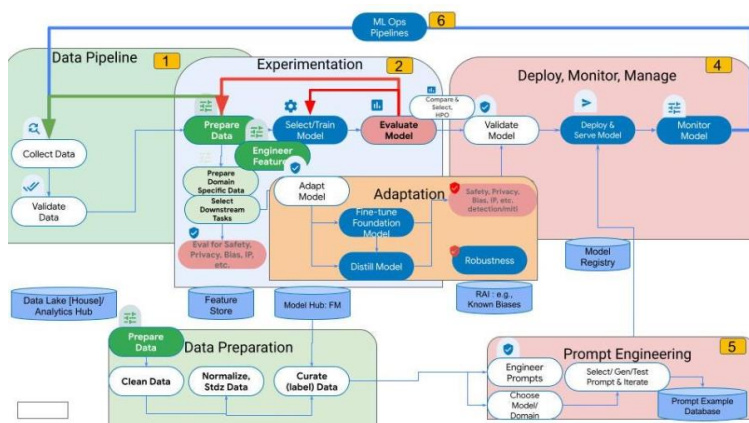


Figure 1: Methodology of proposed work

5. CONCLUSION

Generative AI represents a significant advancement in medical diagnosis, offering innovative solutions to longstanding challenges in healthcare. By enabling data synthesis, improving diagnostic accuracy, and supporting multimodal analysis, generative AI has the potential to transform clinical practice. Recent studies demonstrate its effectiveness in disease detection, predictive analytics, and personalized medicine, highlighting its role in improving patient outcomes.

However, the adoption of generative AI in healthcare requires careful consideration of ethical, technical, and regulatory challenges. Issues such as data privacy, model interpretability, and clinical validation must be addressed to ensure safe and effective implementation. Furthermore, collaboration between AI researchers, healthcare professionals, and policymakers is essential to develop robust and trustworthy AI systems.

In conclusion, while generative AI is not yet a replacement for human expertise, it serves as a powerful tool that can augment clinical decision-making and enhance diagnostic processes. Future research should focus on improving model reliability, integrating explainable AI techniques, and establishing standardized evaluation frameworks to facilitate the widespread adoption of generative AI in medical diagnosis.

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