International Research Journal of Innovations in Engineering and Technology (IRJIET)

ISSN (online): 2581-3048

Volume 9, Issue 10, pp 54-60, October-2025

https://doi.org/10.47001/IRJIET/2025.910008

Deep Learning-Based Medical Diagnosis Systems Using Artificial Neural Networks: A Comprehensive Review

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***Abstract* - The rapid advancements in Artificial Intelligence (AI), particularly in Deep Learning (DL) and Artificial Neural Networks (ANNs), have revolutionized various sectors, with medical diagnosis emerging as a prominent beneficiary. This paper provides a comprehensive review of deep learning-based medical diagnosis systems utilizing ANNs, drawing insights from recent research. We explore the fundamental concepts, architectural designs, and diverse applications of these systems across a spectrum of medical conditions, including cardiovascular diseases, various cancers, neurological disorders, and respiratory illnesses. Furthermore, we delve into the challenges associated with implementing these advanced technologies, such as data requirements, labelling complexities, model interpretability, and ethical considerations. By synthesizing information from multiple studies, this paper aims to offer a structured understanding of the current landscape, future prospects, and critical areas for further research in deep learning-driven medical diagnosis.**

Keywords: Deep Learning, Artificial Neural Networks, Medical Diagnosis, Machine Learning, Image Processing, Healthcare AI, Convolutional Neural Networks, Residual Networks.

I. INTRODUCTION

The healthcare industry, characterized by its high-priority status and the demand for exceptional care, faces persistent challenges in accurate and timely disease diagnosis. Diagnostic errors, often stemming from subjectivity, disease complexity, and human factors, can lead to inappropriate treatments, increased medical expenses, and significant patient distress. The advent of Artificial Intelligence (AI) offers transformative solutions to these issues, with Machine Learning (ML) and Deep Learning (DL) techniques at the forefront these advanced computational paradigms are adept at identifying intricate patterns and trends within vast datasets, making them exceptionally well-suited for prediction and forecasting tasks in medicine.

Deep learning, a subfield of machine learning, leverages multi-layered Artificial Neural Networks (ANNs) to process complex data, including medical images, signals, and electronic health records. Unlike conventional ML algorithms that often rely on expert-engineered features, DL models can automatically learn hierarchical features, leading to highly accurate results and efficient analysis of massive datasets. This capability is particularly crucial in medical imaging, where devices now produce high-resolution data (e.g., X-Ray, CT, MRI scans) that can be challenging for human interpretation alone.

This paper aims to provide a structured and detailed overview of deep learning-based medical diagnosis systems using ANNs. We will explore the historical context, describe the core components and architectures, highlight their diverse applications, discuss the inherent challenges, and outline future directions for this rapidly evolving field.

II. HISTORY

The concept of neural networks, initially inspired by biological neurons, has evolved significantly over time. Early work on Artificial Neural Networks (ANNs) in the mid-1980s saw applications like the "instant physician," which trained auto-associative memory neural networks to store and retrieve medical records for diagnosis and treatment suggestions. These early systems demonstrated the potential of ANNs to process and learn from medical data.

The broader field of AI has seen significant advancements, particularly driven by ANNs, leading to what is now known as Deep Learning (DL). The success of DL has been fuelled by increased access to data ("big data"), user-friendly software frameworks, and an explosion in computing

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power. DL became prominent in image processing when neural networks began outperforming other methods in high-resolution image analysis. A notable milestone was in 2012, when a Convolutional Neural Network (CNN) model significantly reduced the error rate in the ImageNet Large-Scale Visual Recognition Challenge (ILSVRC), demonstrating the power of deep learning for visual recognition tasks.

In the medical domain, the application of DL has gained momentum with the release of large-scale annotated datasets. For instance, the "ChestX-ray8" database, comprising over 100,000 frontal-view X-ray images, was introduced to facilitate the development of deep learning paradigms for computer-aided diagnosis (CAD) systems. Subsequent datasets like ChestX-ray14 further expanded the scope to include more thoracic diseases, providing a rich resource for training advanced DL models. These developments have paved the way for algorithms like CheXNet, a 121-layer CNN, which demonstrated radiologist-level performance in pneumonia detection on chest X-rays.

III. DESCRIPTION

A Deep Learning-Based Medical Diagnosis System Using Artificial Neural Networks fundamentally involves leveraging multi-layered neural networks to analyse complex medical data for diagnostic and predictive purposes. The core idea is to enable machines to learn from vast amounts of medical information, identify subtle patterns, and make informed decisions, often surpassing human capabilities in speed and consistency.

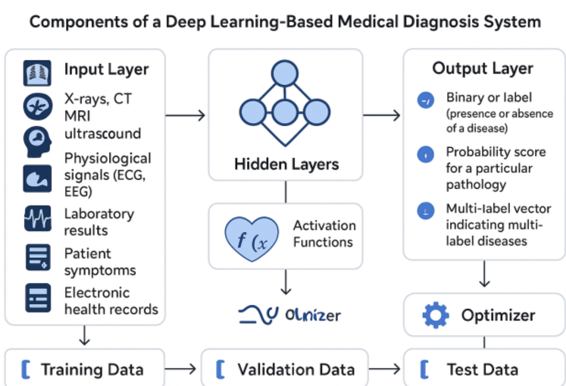
The process typically begins with data preprocessing, a crucial step to reduce false predictions, speed up processing, and improve data quality. This is followed by feature extraction, which can be automated by DL algorithms, unlike classical methods that require manual feature engineering. The system then undergoes model training and parameter adjustment based on the chosen DL or ML algorithm, aiming for accurate classifications or predictions.

The "deep" aspect of these systems refers to the numerous processing layers within the neural network architecture. These layers allow the Gradient Descent) used to adjust the network's network to learn hierarchical features, building simple concepts into more complex representations. This deep structure enables the system to handle the inherent complexity and variability of medical data, which often cannot be accurately simulated with simple equations.

IV. COMPONENTS

A typical deep learning-based medical diagnosis system comprises several key components:

- **Input Layer:** Receives raw medical data, which can include images (X-rays, CT, MRI, ultrasound), physiological signals (ECG, EEG), laboratory results, patient symptoms, and electronic health records (EHRs).
- **Hidden Layers:** These are the core of the deep neural network, consisting of multiple layers of interconnected artificial neurons. Each hidden unit processes information from the previous layer, applying weights and activation functions to learn increasingly abstract features. The number and configuration of these layers define the "depth" of the network.
- **Output Layer:** Produces the system's prediction or classification. This could be a binary label (e.g., presence or absence of a disease), a probability score for a particular pathology, or a multi-label vector indicating the presence of multiple diseases.
- **Weights and Biases:** Parameters within the network that are adjusted during the training process to optimize the network's performance.
- **Activation Functions:** Non-linear functions applied within neurons to introduce non-linearity into the model, enabling it to learn complex relationships.
- **Loss Function:** A mathematical function that quantifies the difference between the network's predictions and the actual ground truth labels. The goal of training is to minimize this loss.
- **Optimizer:** An algorithm (e.g., Adam, Stochastic weights and biases based on the calculated loss, guiding the learning process.
- **Training Data:** A large, annotated dataset of medical information used to teach the network to recognize patterns and make accurate predictions. The quality and quantity of this data are critical for model performance.
- **Validation Data:** A subset of the data used to tune hyperparameters and evaluate the model's performance during training, preventing overfitting.
- **Test Data:** An independent dataset used to evaluate the final, trained model's generalization ability to unseen data.



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V. TOOLS

The development and implementation of deep learning based medical diagnosis systems rely on a suite of specialized tools and frameworks:

Deep Learning Frameworks:

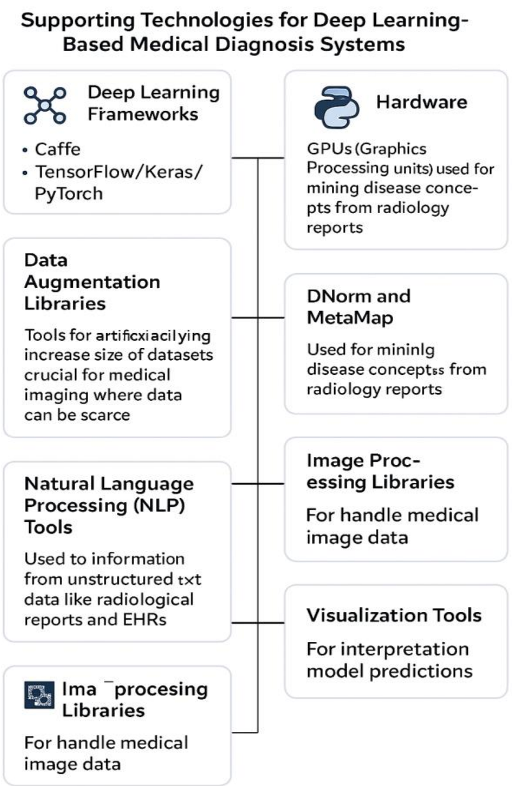
- **Caffe:** Used for implementing CNN architectures, particularly for image recognition tasks.
- **TensorFlow/Kera's/PyTorch:** Widely used open-source libraries that provide flexible platforms for building, training, and deploying various deep learning models.

Programming Languages:

- **Python:** Dominant language in AI and DL due to its extensive libraries and ease of use.

Hardware:

- **GPUs (Graphics Processing Units):** Essential for accelerating the computationally intensive training of deep neural networks.
- **Data Augmentation Libraries:** Tools for artificially increasing the size of datasets by applying transformations to existing data, crucial for medical imaging where data can be scarce.



- **Natural Language Processing (NLP) Tools:** Used to extract information from unstructured text data like radiological reports and electronic health records, enabling the mining of disease labels and other clinical insights.
- **DNorm and Meta Map:** Tools specifically mentioned for mining disease concepts from radiology reports.
- **NLTK (Natural Language Toolkit):** Used for splitting and tokenizing reports into sentences.
- **Blip parser and Stanford dependencies converter:** Used for parsing sentences and obtaining syntactic dependencies.
- **Image Processing Libraries:** For handling medical image data, including resizing, normalization, and other preprocessing steps.
- **Visualization Tools:** For interpreting model predictions, such as Class Activation Maps (CAMs) that highlight areas of an image indicative of pathology.

VI. ARCHITECTURE

The architecture of deep learning models for medical diagnosis is diverse, often tailored to the specific task and data type. However, several prominent architectures and concepts are frequently employed:

- **Feed-forward Networks:** Signals travel in one direction from input to output without loops. Extensively used in pattern recognition.
- **Feedback Networks:** Allow signals to travel in both directions, introducing loops. These are dynamic and can become very complicated, often referred to as recurrent networks.
- **Multi-Layer Perceptron's (MLP):** A type of feed-forward ANN with one or more hidden layers, capable of classifying non-linear separable patterns.

Convolutional Neural Networks (CNNs):

- **Structure:** Consist of convolutional layers, pooling layers, and fully connected layers. They are designed to extract spatial information from input images.
- **Weakly-Supervised Localization:** Frameworks like the one used in ChestX-ray8 leverage CNNs for multi-label image classification and disease localization, generating heatmaps to indicate approximate spatial locations of pathologies.
- **Pre-trained Models:** Often initialized with weights from models pre-trained on large datasets like ImageNet (e.g., Alex Net, Google Net, VGGNet-16, ResNet-50) and then fine-tuned for medical tasks.
- **Dense Net (Dense Convolutional Network):** CheXNet, a 121-layer CNN, is based on the DenseNet architecture, which improves information flow and gradient propagation, making very deep networks tractable.

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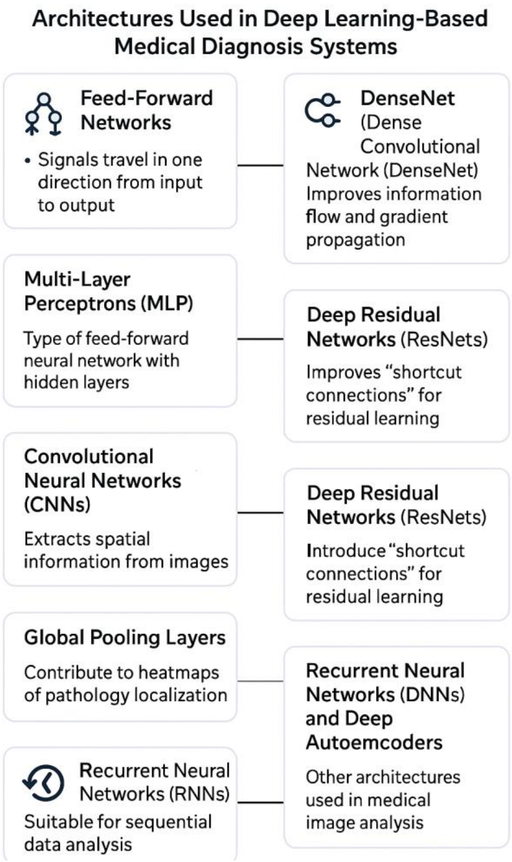
Deep Residual Networks (ResNets):

- **Concept:** Introduce "shortcut connections" that bypass one or more layers, allowing the network to learn residual functions. This addresses the "degradation problem" in very deep networks, where accuracy saturates and then degrades with increasing depth.
- **Building Blocks:** Typically involve a stack of 2 or 3 layers (e.g., 1x1, 3x3, 1x1 convolutions in "bottleneck" designs) with identity shortcuts for efficient training.
- **Depth:** Successfully trained models with over 100 layers, and even explored models with over 1000 layers.
- **Global Pooling Layers:** Used in CNNs to reduce spatial dimensions and aggregate features, contributing to the generation of likelihood maps (heatmaps) for pathology localization.
- **Log-Sum-Exp (LSE) Pooling:** An adjustable pooling method that can range from max pooling to average pooling, offering flexibility in feature aggregation.
- **Recurrent Neural Networks (RNNs) and LSTMs:** Suitable for sequential data analysis, such as time-series medical data or for processing natural language in reports.
- **Deep Belief Networks (DBNs) and Deep Autoencoders:** Other types of deep learning architectures used in medical image analysis.



VII. APPLICATIONS

Deep learning-based medical diagnosis systems using ANNs have found extensive applications across various medical specialties, significantly enhancing diagnostic capabilities:

Cardiovascular Diseases:

- **Diagnosis:** ANNs are used to model the human cardiovascular system, detect arrhythmias, and diagnose coronary artery diseases.
- **Prediction:** ML and DL methods show remarkable progress in predicting coronary atherosclerotic heart disease and major adverse cardiac events using CTFractional Flow Reserve (CT-FFR).
- **Accuracy:** SVM and ANN models have achieved high accuracies (e.g., 89.1% for Arrhythmia with SVM) in diagnosing various heart conditions.

Cancer Diagnosis:

- **Breast Cancer:** DL models have demonstrated improved accuracy in detecting breast cancer from mammograms, often outperforming radiologists, especially in early stages and with specific breast types. Hybridfuzzy artificial immune systems and optimized SVMs have achieved classification accuracies up to 99.14%.
- **Lung Cancer:** DL models, particularly CNNs, are highly effective in diagnosing lung cancer from CT scans, identifying dangerous lung nodules, and classifying different cancer types (adenocarcinoma, squamous cell carcinoma, small cell carcinoma).
- **Melanoma (Skin Cancer):** CNN models trained on vast datasets of skin pathologies can identify and classify skin cancer with expert-level precision, with potential for mobile-based non-professional skin exams.
- **Prostate Cancer:** AI-based technologies assist in diagnosing prostate lesions, predicting prostate cancer, patient survival rates, and treatment response using MRI data and DL approaches like XmasNet.

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- **Microarray Data Analysis:** AI-based methods, including ensemble ML, play a significant role in classifying cancerous microarray data to identify highly expressed genes in tumourcells.

Neurological Disorders:

- **Alzheimer's Disease (AD):** DL-based CNN models are widely used for predicting and detecting AD, with pretrained models like EfficientNetB0 achieving high accuracy (92.98%).
- **Parkinson's Disease (PD):** Hybrid ML systems combining genetic algorithms and random forests have achieved high accuracy (95.58%) in early diagnosis.
- **Brain Tumors:** DL approaches are crucial for detecting brain tumors, which are often difficult to identify in early stages.

Respiratory Diseases:

- **Pneumonia:** CNNs are highly effective in detecting pneumonia from chest X-rays, with models like CheXNet exceeding average radiologist performance.
- **Chronic Obstructive Pulmonary Disease (COPD):** DL based CNN models analyze respiratory audio data for COPD detection.
- **COVID-19:** CNN-based framework models diagnose COVID-19 using X-ray images, achieving high accuracy.

Other Medical Conditions:

- **Diabetes:** ANNs are used for diabetes recognition and prediction, with some models achieving 91% accuracy.
- **Dermatological Diseases:** ANNs and DL models are trained to diagnose various dermatological diseases with high accuracy.
- **Urinary Tract Infection (UTI):** ANNs improve symptomatic understanding and diagnosis of UTI.
- **Paediatrics Traumatic Brain Injury (TBI):** ANNs are leveraged for moderate to serious prediction of TBI.
- **Diarrhea:** ANN-based models can predict the occurrence of diarrhea for prevention.
- **Kidney Disease:** DL models coupled with predictive analytics are used for early prediction of persistent kidney infections.
- **Autism Spectrum Disorder (ASD):** MLP models have shown 100% accuracy in classifying ASD.

VIII. CHALLENGES

Despite the immense potential, the widespread adoption and optimal functioning of deep learning-based medical diagnosis systems face several significant challenges:

Data Requirements:

- **Massive Data Needs:** DL models are "data-hungry" and require vast amounts of high-quality, annotated data for effective training, which is not always available, especially for rare diseases.
- **Data Scarcity:** For many specific medical conditions or rare diseases, obtaining sufficiently large datasets is a major hurdle.
- **Data Consistency and Storage:** Non-consistency in data collection and storage across different institutions can lead to fluctuating precision of models.

Data Labelling and Annotation:

- **Expertise and Cost:** Labelling medical data (e.g., annotating pathologies in images) requires specialized medical expertise, is time-consuming, and highly costly.

Fig. Instant Physician

- **Subjectivity:** Human interpretation in labelling can introduce variability and subjectivity, impacting the ground truth for training.

Model Complexity and Interpretability:

- **"Black Box" Nature:** Deep neural networks, especially very deep ones, can be complex and profound, making their internal workings difficult for humans to understand ("black box" problem). This lack of interpretability can hinder trust and acceptance by medical professionals.
- **Computational Resources:** Training complex DL models requires significant computational power, often necessitating specialized hardware like GPUs.

Generalization and Robustness:

- **Overfitting:** Aggressively deep models can overfit to the training data, leading to poor generalization performance on unseen data.

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- **Variability in Patient Information:** The broad variety of patient information can make conventional learning strategies unreliable.
- **Noise in Data:** Medical data often contains noise, which can sometimes lead to misleading results, especially in complex systems like the human body.

Ethical and Regulatory Considerations:

- **Patient Privacy:** Handling sensitive patient data requires robust privacy protection systems and adherence to strict regulations.
- **Bias and Fairness:** AI algorithms trained on population representation information can inherit biases present in the data, potentially leading to inaccuracies or discrimination in diagnosis.
- **Accountability:** Establishing accountability for diagnostic errors made by AI systems is a complex ethical and legal challenge.

Integration into Clinical Workflow:

- **Acceptance by Specialists:** Convincing medical professionals to accept and integrate AI technologies into their daily practice requires demonstrating reliability, accuracy, and clinical utility.
- **Standardization:** A lack of standardized guidelines for study design and reporting can lead to overestimation of AI model performance.

IX. FEATURES

Deep learning-based medical diagnosis systems offer several compelling features that distinguish them from traditional diagnostic approaches:

- **High Accuracy and Precision:** DL models, particularly CNNs and ResNets, have demonstrated the ability to achieve and even surpass human expert-level performance in detecting and classifying various diseases, especially in image-based diagnostics.
- **Automated Feature Extraction:** Unlike traditional ML, DL algorithms can automatically learn and extract relevant features from raw data, eliminating the need for manual, expert-driven feature engineering, which is often subjective and time-consuming.
- **Efficiency and Speed:** These systems can process large volumes of data rapidly, significantly reducing diagnosis time and workload for physicians.
- **Weakly-Supervised Learning:** Advanced frameworks can perform disease localization (e.g., identifying the exact region of a pathology in an image) even with only image level labels, reducing the burden of detailed pixel-level annotations.
- **Handling Complex Data:** DL is adept at analysing complex, high-dimensional medical data, including high-resolution images and intricate physiological signals, which are challenging for human interpretation.
- **Pattern Recognition:** Exceptional ability to identify subtle patterns and correlations in data that might not be obvious to the human eye, leading to earlier and more accurate diagnoses.
- **Scalability:** Capable of analysing massive datasets, making them suitable for hospital-scale applications and large-scale screening programs.
- **Generalization:** Well-trained models can generalize effectively to new, unseen patient data, providing consistent diagnostic support.
- **Reduced Human Error:** By automating parts of the diagnostic process, these systems can minimize errors caused by human fatigue, inexperience, or cognitive biases.
- **Personalized Medicine:** Potential to develop personalized treatment plans based on individual patient characteristics and medical history.
- **Accessibility:** Can increase access to medical imaging expertise in underserved regions by providing automated diagnostic support.

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Citation of this Article:

Prof. S.B. Bele, Pratiksha Sahare, Bhushan Dobale, Nivedita Chaudhari, & Prathamesh Gorde. (2025). Deep Learning-Based Medical Diagnosis Systems Using Artificial Neural Networks: A Comprehensive Review. *International Research Journal of Innovations in Engineering and Technology - IRJIET*, 9(10), 54-60. Article DOI https://doi.org/10.47001/IRJIET/2025.910008

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