**Deep Learning Application in Human Disease Detection and Drug Discovery**

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**Abstract** – In the past decade, drug discovery and disease detection were slow and laborious processes due to the limitations of traditional methods. The process of identifying and developing new drugs was often lengthy and expensive, and the accuracy of disease diagnosis was often limited by the subjectivity of human interpretation. However, the rapid advancement of artificial intelligence (AI) is revolutionizing the medical field by minimizing human errors, enhancing clinical outcomes, and enabling continuous data monitoring. Deep learning, a branch of machine learning, has emerged as a powerful tool to overcome the challenges posed by the high-dimensionality, heterogeneity, temporal dependency, sparsity, and irregularity of medical data. This paper aims to provide a comprehensive review of deep learning network architectures specifically tailored for medical technology applications. The focus will be on convolutional neural networks (CNNs) and recurrent neural networks (RNNs), two prominent architectures that have revolutionized medical technologies. In the context of disease detection, CNNs have demonstrated remarkable efficacy in tasks such as medical image analysis, electronic health record analysis and biomarker analysis. RNNs, on the other hand, have proven valuable in drug discovery, particularly in drug target identification, drug efficacy and safety prediction, and clinical trial data analysis. As deep learning continues to evolve, we can expect even more innovative applications of these powerful architectures, further transforming healthcare practices and improving patient outcomes.

**Keywords** – Deep learning, Disease detection, Drug discovery, Healthcare, Machine learning

**Introduction**

Advancement in digital healthcare technology, such as artificial intelligence (AI) is transforming the medical field by minimizing human errors, enhancing clinical outcomes, and enabling continuous data monitoring. AI methodologies, ranging from machine learning to deep learning, are playing a pivotal role in refining clinical systems, optimizing patient information and records, diagnosing, and pioneering treatments for a wide spectrum of ailments [7,18].

The adoption of machine learning in medicine has been hindered by data challenges. The high-dimensionality, heterogeneity, temporal dependency, sparsity, and irregularity of medical data pose significant obstacles to the effectiveness of traditional machine learning methods. However, deep learning offers a powerful solution to overcome these challenges [18]. Deep learning methods are versatile and can be effectively applied to a wide range of medical domains by utilizing neural networks with multiple layers, enabling the identification of progressively more complex features from raw data [2,8]. The application of neural networks has led to significant breakthroughs in several domains, including disease detection and drug discovery [10].

In particular, the application of convolutional neural networks (CNNs) for image detection and recurrent neural networks (RNNs) for model training has gained significant traction in the medical technology revolution [7,20]. Authors of [7] provide a comprehensive review of ongoing research dedicated to supporting medical professionals with automated systems that can analyze images and diagnose acute diseases, ranging from brain tumors and bone cancer to retinal diseases, using CNNs. Authors of [13] have proposed de novo drug design approach using RNNs. By leveraging the strengths of RNNs in generating synthetically reasonable molecules, this approach addresses the limitations of existing methods and overcomes the challenges of the vast chemical search space. With the continuous advancements in deep learning algorithms, it is crucial for healthcare professionals, researchers, and developers to stay updated with these developments to effectively harness the potential of deep learning in transforming healthcare practices and improving patient outcomes [18].

This paper aims to provide a comprehensive review of deep learning network architectures specifically tailored for medical technology applications. The focus will be on CNNs and RNNs, two prominent architectures that have revolutionized disease diagnosis and drug discovery [5, 7, 13, 18].

1. **Deep learning**

Deep learning (DL) is a branch of machine learning that teaches computers and devices how to think. By processing vast amounts of data and extracting intricate patterns, DL algorithms emulate human learning processes. This is accomplished through a layered structure known as neural networks, where each layer progressively extracts increasingly complex information from the data [10].

DL is built upon artificial neurons, which are modeled after the neurons in brains. Each artificial neuron, also known as a perceptron, receives weighted inputs from other neurons or from the environment. The neuron then processes these inputs using a specific function and produces an output. This process mimics the way our brains process information and make decisions. The weight assigned to each input represents its significance in determining the output of the neural network. Inputs with higher weights exert a stronger influence on the network's decision-making process [10].

DL enhances human life, enabling the development of advanced systems with higher accuracy. DL is used in scenarios where human expertise is unavailable, unable to articulate the reasoning behind expert decisions, problems that require continuous updates to their solutions, cases where solutions demand adaptation based on specific circumstances, and problems that are too vast for limited reasoning abilities to comprehend [11].

DL has proven to be a powerful tool in various situations, even surpassing human expertise [2, 11]. This framework in cancer classification for example, has been shown to be more accurate than dermatologists at detecting skin cancer with an accuracy of up to 95% [19].

Two of the most popular DL architectures are convolutional neural networks (CNNs) and recurrent neural networks (RNNs) [1, 18].

**1.1 Convolutional neural networks (CNNs)**

Convolutional neural networks (CNNs) is a type of deep neural network that has revolutionized the field of image recognition. Their ability to automatically extract and learn features from image data has led to significant advancements in tasks such as image classification, object detection, and anomaly detection. In the medical field, CNNs have proven invaluable for detecting abnormalities in X-rays, CT (Computed Tomography) scans, and MRIs (Magnetic Resonance Imaging) [3,6].

CNNs draw inspiration from the organization of the visual cortex in cats. They utilize local connections and shared weights across units, followed by feature pooling (subsampling) to extract translation-invariant descriptors [18]. CNNs serve to simplify images for easier processing using a large dataset of labeled documents to be trained [10]. While some information is inevitably lost during this process, the essential features required for accurate predictions are preserved. CNNs utilize appropriate filters to capture the spatial and temporal relationships within an image. By reducing the number of parameters and leveraging weight reusability, CNNs achieve improved fitting to the image dataset [18].

**1.2 Recurrent neural networks (RNNs)**

Recurrent neural networks (RNNs) are a specialized type of deep neural network that excel at processing sequential data streams [18]. RNNs can be conceptualized as a cascade of interconnected neural networks, where each network receives and processes information from its predecessor, maintaining an internal memory known as the hidden state. This hidden state represents the accumulated knowledge of the sequence up to that point, allowing each neural network to make predictions about the next element in the sequence based on the entire context. This process mimics the way humans process and understand language, where each word or phrase is interpreted in light of the preceding ones [19].

Unlike traditional neural networks that treat each input independently, RNNs possess an internal memory that allows them to retain and process information from previous inputs. This unique feature enables RNNs to capture temporal dependencies within data, making them particularly well-suited for tasks such as time series forecasting, speech recognition, natural language processing, financial data analysis, audio and video processing, and weather prediction [10].

1. **Deep learning in healthcare**

Deep learning algorithms have demonstrated remarkable efficacy in various healthcare applications, including medical image analysis, drug discovery, personalized medicine, disease prediction and risk assessment, and clinical trial analysis [8-10]. This paper focuses on discussing the use of DL in disease detection and drug discovery.

**2.1 Disease detection**

There are three ways of DL implementation in disease detection [18]. DL algorithms can be used to extract meaningful information from medical images [1, 18]. DL can also be used to analyze electronic health records (EHR) data to identify patterns that may indicate the presence of disease [12]. Additionally, DL can be used in genomics to analyze DNA and RNA sequences to identify genetic mutations that are associated with an increased risk of disease [1,7,15,18]. DL has the potential to revolutionize disease detection by making it more accurate, efficient, and personalized [18].

**2.1.1 Medical image analysis**

Early applications of deep learning in the medical field focused on image analysis, particularly in the examination of brain MRI scans to anticipate the development of Alzheimer's disease and its various forms [18].

Medical image analysis utilizes CNNs to perform classification, segmentation, and object detection tasks. In medical image classification, CNNs are trained to identify specific abnormalities or disease patterns in medical images. For instance, they can classify X-rays as normal or abnormal based on the presence of pneumonia or tuberculosis. CNNs can also be used to classify skin lesions as benign or malignant based on their appearance in dermoscopy images. Additionally, CNNs can perform segmentation tasks in medical images, such as segmenting tumors in MRI scans or delineating the boundaries of organs in CT scans. Accurate segmentation is crucial for tasks such as volume estimation, treatment planning, and surgical guidance [1].

In a study involving over 130,000 clinical images, a CNN demonstrated performance comparable to 21 board-certified dermatologists in classifying biopsied skin cancer lesions, including keratinocyte carcinomas versus benign seborrheic keratoses and malignant melanomas versus benign nevi [18].

**2.1.2 Electronic health records (EHR) analysis**

Electronic health records (EHRs), initially designed to enhance healthcare system efficiency and accessibility, have evolved to serve a wide range of purposes in clinical informatics and epidemiology. Specifically, EHR data has been utilized for tasks such as medical concept extraction, disease and patient clustering, patient trajectory modeling, disease prediction, and data-driven clinical decision support [12].

Early studies of EHR employed less complex and conventional statistical methods [12]. Recent advancements have enabled deep learning algorithms to effectively analyze comprehensive EHRs encompassing both structured and unstructured data elements like diagnoses, medication lists, laboratory test results, and free-text clinical notes [4, 18].

EHR uses both CNNs and RNNS in its application. Deepr (Deep record) CNN architecture for example, which models a patient's health journey as a lengthy sequence of medical codes. Each code is represented in a new space to enable statistical and algebraic operations, similar to word embedding in natural language processing. The time intervals between events are represented as unique words [12]. Deepr demonstrated effective performance in predicting patient readmission within six months and successfully identified meaningful and interpretable clinical patterns [18].

**2.1.3 Genomics**

Genomics primarily focuses on understanding the structure, function, evolution, and modification of genomes at the organism level, while at the molecular level, we adhere to the central dogma of molecular biology to dissect and characterize the functions of individual molecules in a detailed manner. The central dogma of molecular biology outlines the process by which genetic information is transferred from DNA to RNA, and subsequently from RNA to protein, essentially completing the transcription and translation of genetic information (Crick, 1970) [15].

The initial applications of neural networks in genomics involved replacing conventional machine learning algorithms with DL architectures, while maintaining the same input features [18]. For instance, the employment of a fully connected feed-forward neural network to predict the splicing activity of individual exons. The model was trained on over 1000 predefined features extracted from the candidate exon and its neighboring introns. This approach achieved higher accuracy in predicting splicing activity compared to simpler methods, and it also enabled the identification of rare mutations associated with splicing dysregulation [18].

Recent advancements in genomics have enabled the application of CNNs directly to raw DNA sequences, eliminating the need for prior feature definition [18]. Yuan et al. [16] proposed a novel CNN based approach, named CNNC to decipher relationships between genes, infer causal connections, assign functional roles, and predict disease-associated genes. The approach outperforms existing methods in inferring gene relationships from single-cell expression data.

**2.2 Drug discovery**

The clinical drug development process has experienced slow advancements over the past three decades [17]. The traditional drug discovery was heavily reliant on medicinal chemists conducting extensive testing, validations, and synthetic procedures in a laboratory setting. This cumbersome approach resulted in substantial time and financial investments to bring a single drug to the clinical stage. The introduction of DL has revolutionized the conventional drug discovery process, making it more efficient and streamlined. The vast amount of biological data stored in various databases worldwide serves as the foundation for DL approaches, enabling the precise identification of patterns and models that can guide the discovery of therapeutically active molecules with significantly reduced time, resource, and financial investments [2, 20].

Collaboration between computer scientists and medicinal chemists has emerged to utilize the DL method in drug discoveries. This collaboration has led to the development of DL-powered tools, predictive models, and algorithms that can be effectively integrated into the drug discovery and development pipeline [2].

Different from disease detection, drug discovery mostly uses RNNs as its main architecture because of the sequence based tasks in drug discovery [18, 20]. DESMILES, for instance, a novel drug design approach that leverages the principles of RNN architecture, as described by Maragakis et al [14], to effectively explore the vast chemical landscape and identify potential drug candidates [9]. DESMILES employs a unique approach by encoding molecular fingerprints instead of sequences of tokens or images. The encoded fingerprint is then fed into a decoder RNN, which is trained to generate the corresponding text-based representation of chemical structures called the SMILES (Simplified Molecular Input Line Entry System) string.

1. **Conclusion**

Deep learning has emerged as a transformative technology in the healthcare industry, revolutionizing disease detection, drug discovery, and various other medical applications. As mentioned in the paper, deep learning has the ability to extract significant patterns and keywords from vast amounts of data, improving patient care and outcomes.

Conventional extracting methods are slow and can lead to mistakes. This can delay diagnosis, treatment, and overall patient care. Deep learning, on the other hand, can quickly and accurately extract information from large datasets. This allows for faster and more accurate diagnosis, treatment planning, and monitoring of patient progress. Deep learning can also find patterns that humans might miss, which can lead to new discoveries and breakthroughs in healthcare.

Additionally, deep learning is useful for drug discovery. By analyzing vast amounts of data on molecular interactions, deep learning algorithms can identify potential drug candidates that may be effective in treating a specific disease. This can significantly reduce the time and cost of drug development, bringing new and effective treatments to patients more quickly.

The paper also looks at deep learning architecture implementation. It is revealed that convolutional neural networks (CNNs) are better in disease detection. CNNs' widespread adoption is a result of their favorable attributes in digital imaging, including analyzing and classifying image data. Conversely, recurrent neural networks (RNNs) are primarily employed in drug discovery because of their inherent ability in processing and analyzing sequential data, which is a key aspect of drug development.

Deep learning has many applications in healthcare. This paper mostly discussed disease diagnosing using medical image analysis, electronic health records (EHR) analysis, and genome, also deep learning in drug discovery. Given the remarkable advancements achieved thus far, further research in deep learning should be used as a diagnosing and discovery tool.

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