

Machine Learning for Healthcare: Emerging Challenges and Opportunities in Disease Diagnosis

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Received date: July 28, 2020, **Accepted date:** July 30, 2020

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Keywords: Machine learning, Artificial neural network, Healthcare, Disease diagnosis, Heart diseases, Cancer

Diagnosis is a process that identifies, explains, or establishes the individual's disease from its symptoms and signs. Early and precise diagnosis is crucial since it influences the efficacy of treatment and avoids long-term complications for the infected person. Further, in the case of infectious diseases, undiagnosed patients can transmit the disease to a healthy population unknowingly. Besides, most of the diseases evolve with the time that significantly affects the clinical outcomes. Also, diseases including anthrax and pulmonary embolism are important to establish immediately as the late diagnosis can lead to significant patient harm. Some diseases can be diagnosed in a short time, while others may take months due to the complexity of disease presentation. Importantly diagnostic errors can contribute to about 10% of patient deaths and also account for several adverse complications and/or events in hospitals [1-3]. The physician's performance is typically not directly attributed to the cause of diagnosis error. Several factors including lack of communication between clinicians, patients and their families, inadequate diagnostic processes, and inefficient health information systems can contribute to diagnostic errors. Machine learning (ML) offers a sophisticated, automatic approach for analysis of high-dimensional and multimodal biomedical data that can expedite and improve medical diagnostics significantly. The ML algorithms once designed can perform the given task over and over with high reproducibility or accuracy, which is vital for making clinical decisions.

Non-invasive diagnosis performed without breaking skin or any contact with the body cavity has a great significance in clinical practice for the diagnosis of heart diseases. ML image-based approaches are improving the diagnostic

accuracy and reducing the needless downstream testing. Support Vector Machine (SVM), a set of supervised ML algorithm which is helpful in classification, regression, and outliers detection demonstrated promising results for the diagnosis of coronary artery disease (CAD). N2Genetic-nuSVM optimization technique delivered the accuracy of 93.08% and F1-score of 91.51% in predicting CAD outcomes among the patients [4]. Kukar et al. employed different ML models and utilized scintigraphy, and ECG of patients to detect CAD. Interestingly, some ML models showed 0.92 accuracies compared to clinicians score that was 0.91 [5]. Similarly, Guner et al. develop and analyzed the efficacy of artificial neural networks (ANN) that are powerful ML-based techniques in detecting CAD from myocardial perfusion SPECT (MPS) [6]. A cohort of 243 patients with MPS and coronary angiography were selected to train the ANN. Interestingly, the area under the curve that often measures the quality of the classification models and accuracy was 0.74, similar to the expert analyses, suggesting that ML has the potential to assist in the nuclear cardiology environment.

ANN and fuzzy clustering methods were developed by Sun et al. that detect influenza-infected patients by classifying vital signs such as respiration rate, heart rate, and facial temperature [7]. The combinations of SVM, nested one-versus-one-SVM, Matlab, and leave one out cross-validation method showed 100% accuracy in separating bacterial gene sequences over other popular methods including high-resolution melt (HRM) [8]. The conventional method for malaria diagnosis is time-consuming and demands special skill sets and expertise. A simple ML approach coupled with digital in-line holographic microscopy (DIHM) was developed to identify the red blood cells (RBCs) characteristics [9]. Out of 13 segmented holograms of individual RBCs, 10 featured showed a statistical difference between healthy RBCs and

infected RBCs. Six ML algorithms applied to enhance the diagnostic capacity showed that SVM had best accuracy [training ($n=280$, 96.78%) and testing set ($n=120$, 97.50%)] in separating healthy from infected RBCs [9]. For the early diagnosis of tuberculosis and to improve the classification accuracy of the Artificial Immune Recognition System (AIRS), Saybani et al. developed an SVM model. A cohort of 175 samples including 114 positive samples for tuberculosis and 60 samples in the negative group were utilized. The AIRS method successfully classified tuberculosis patients, and the model performed with a 100% accuracy, sensitivity, and specificity [10].

Images obtained from histopathology biopsy, magnetic resonance, computed tomography, and mammograms are helpful in the diagnosis and staging of several malignancies. ML approaches are playing a significant role in cancer prediction, diagnostics, and forecasting therapeutic outcomes. Wang et al. developed predictive models using ML-based SVM, Least Squares-SVM, ANN, and Random Forest (RF) approaches to detect prostate cancer. Cohorts of 1625 patient's biopsies were evaluated [11]. Among these ML models, ANN demonstrated the highest accuracy of 0.95 with 0.97 AUC value. Moreover, RF showed the highest accuracy (0.97) in classifying benign and cancerous tumors compared to the other three approaches. Cheerla et al. developed an ML technique for pancreatic cancer diagnosis. The authors used tissue microRNA and clinical data from The Cancer Genome Atlas (TCGA) database and achieved 97.2% accuracy for classification [12]. Diagnosis of benign or malignant breast tumors by cancer cell images is an important computer-aided feature. An Extreme Learning Machine classification performed for image segmentation using the UC Irvine Machine Learning Repository database showed performance with 98.99% accuracy [13].

Machine learning algorithms trained for image analyses can identify abnormalities and pinpoint the area that requires immediate attention. It offers an objective opinion that can significantly improve efficiency. However, it is also crucial for ML approaches to deliver the results in a simple form so the healthcare professionals can understand and interpret the output with high confidence. ML could be an additional tool to help physicians to improve ongoing care [14]. However, it may not replace the physician as patients will always need the human touch and an empathetic relationship with a healthcare professional. ML models are precise when they trained with clean, accurate, and a large amount of data. The chance of achieving better output is dependent on the quality of input. The health care system is continuously evolving, and it may not surprise many if the machine learning tools become part of regular health care. As we are moving forward towards the future, we are creating tons of data every single day.

However, in the data-driven society, most of the generated data is unstructured and messy. To connect it with the real world and make it more meaningful, we surely need more sophisticated approaches. ML provides an opportunity to link the current data to useful future predictions. Although, the development of innovative algorithms for maximum information from data coupled with the most suitable ML model is key to better predictions. The continuous inflow of data can be a valuable resource and help in solving critical problems in healthcare, which could lead to better clinical outcomes.

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