Significance of Machine Learning Algorithms to Improve Predictive Analytics in Chronic Disease Management through Pharmacogenomics

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KEYWORDS

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ABSTRACT

The treatment of chronic diseases is an important concern in global health. Machine learning (ML)-based disease prediction models are becoming more important for making informed medical decisions in light of the paradigm shift towards preventative care. Integrating genetic, pharmacogenomic, personal health, and psychosocial data can greatly assist healthcare practitioners in making treatment-related decisions for patients with chronic diseases. This study utilizes a Deep Convolutional Neural Network-assisted Chronic Disease Management (DCNN-CDM) through pharmacogenomics and an improved predictive analytics model to enable informed real-time decision-making at the point of care. Data augmentation in terms of feature space allows the DCNN model to avoid over-fitting while effectively capturing high-level features submerged in chronic disease datasets. Afterwards, this work suggests an attention-empowered DCNN model to enhance sick case diagnosis accuracy, which augments data regarding sample space, thereby alleviating the class imbalance issue. Electronic health information data mining is now using predictive analytics to determine individuals at risk of acquiring chronic disease problems. The suggested model may aid in the early and accurate diagnosis of chronic diseases. The numerical outcomes demonstrate that the recommended DCNN-CDM model increases the accuracy rate of 98.7%, patient monitoring rate of 97.5%, F1-score rate of 96.3% and predictive performance rate of 95.1% compared to other existing methodologies.

1. Introduction

Chronic diseases are long-lasting disorders that often cannot be prevented with vaccination or completely cured without medical intervention. The progression of several chronic illnesses often occurs gradually over a period and is frequently brought on by unhealthy behaviours such as smoking or a lack of physical activity [1]. Some examples of chronic illnesses are diabetes, bipolar disorder, arthritis, congestive heart failure, and asthma[4]. Programs that attempt to make individuals healthy and independent to the greatest extent feasible by recognizing and treating these problems early [2] are described as chronic illness prevention and management programs. Both the management and prevention of chronic illnesses are the goals of these initiatives [3] Recent research has shown that pharmacogenomics (PGx) can potentially be advantageous in treating chronic pain [21]. Pharmaceutical treatment responses in humans are investigated in the pharmacogenomics field, often referred to as PGx. This is accomplished by analyzing the whole genome to identify the genes involved and the combinations of those genes with other loci [5]. Pharmacology, which investigates the effects of medications, and genomics, which investigates the structure, function, evolution, and mapping of genomes, are the two fields combined to form PGx [20]. Because of this, it is possible to develop treatment plans that are both safe yet successful and tailored to each person's specific genetic makeup. There have been suggestions for future chronic pain pharmacotherapy that use both quantitative and qualitative techniques from precision medicine. This is in response to the high rates of pharmacological ineffectiveness, the subjective nature of pain, and the emotional experience associated with this actual or prospective tissue damage [26]. Due to its autonomous learning capabilities and low error rate, ML has recently emerged as the most promising technology for assisting in medical diagnostics [8]. One of the most critical difficulties has not been well investigated, even though representation learning approaches for EHR data have advanced [22]. Capturing the association between features, particularly, is difficult while attempting to maintain data completeness [10]. Vectors and sequences, two types of feature representations, exclude data on the elapsed time between diagnoses [11]. In addition, convolutional neural networks and other deep learning methods rely heavily on feature correlation for spatial layout [23]. However, since EHR data is so diverse, it isn't easy to reduce exactly what the associations are or how to quantify them[9]. By comparing the health trajectories of patients who have not yet received a diagnosis with the captured network, Convolutional Neural Networks (CNNs) were able to predict the probability of chronic disease [13]. The paper's main contribution is designing the Deep Convolutional Neural Network-assisted Chronic Disease Management (DCNN-CDM) through

pharmacogenomics and an improved predictive analytics model to enable informed real-time decision-making at the point of care[12]. Assessing the statistical model of the DCNN model to effectively seize high-level features hidden in chronic disease database and attain maximum classification accuracy. The numerical outcomes have been implemented, and the recommended DCNN-CDM model enhances the accuracy, predictive performance, and F1 scores compared to existing models [7]. The remainder of the paper has been prearranged: section 2 deliberates the literature survey, section 3 suggests the DCNN-CDM model, section 4 discourses the results, and section 5 concludes the research paper.

2. Literature Review

Christoph Gross et al. [25] suggested the Conversational Agent-Patient Interaction Styles (CA-PIS) for Chronic Disease Management. Findings from research on personalized CAs have focused on various topics, including anthropomorphic cues and personalized suggestions[14]. On the other hand, there is a lack of data on how CAs and patients engage in discussions when CAs provide medical treatment. Previous research on physician-patient interactions has shown that these interactions significantly impact treatment adherence, patient satisfaction, and overall results. Potential socioeconomic factors of health (SDOH), COVID-19, and chronic illness were studied in the geographical context by Jessica Embury et al. [15]. Using spatial approaches such as local bivariate association analysis, Global Moran spatial autocorrelation, and regionally weighted regression, the author identified susceptible populations via the lens of COVID-19 and chronic illness. Based on their importance ($P \leq .05$) concerning COVID-19 case ratios, the author of the Pearson correlation analysis selected 26 socioeconomic variable as possible SDOH.

Kaman Fan and Yi Zhao [16] discussed Mobile health technology (mHealth) for chronic disease management. The author provides an overview of using different mHealth techniques, assesses their strengths and weaknesses, and discusses possible obstacles to further growth. No current mHealth technology is less effective than conventional medicine. The most often reported therapies include telehealth and web-based technologies. Improved efficiency and individualization in patient care are probable outcomes of the next generation of mHealth devices built on various technologies. Hohman, Katherine H et al. [17] deliberated the Multi-State Electronic Health Record (EHR)-Based Network for Chronic Disease Surveillance (MENDS). MENDS monitors new developments on a regional level by use of query and visualization tools[6]. The program uses statistical and geographic methodologies to generate estimates of the national and local prevalence of risk factors for chronic diseases. MENDS introduces a distributed network architecture that may be scaled up or down to provide nationwide chronic illness monitoring using EHR data. Anilkumar Chunduru et al. [24] presented the Convolutional Neural Network and Random Forest (CNN-RF) for Multi Chronic Disease Prediction System. This study suggests a way to anticipate seven diseases: cardiovascular disease, renal disease, diabetes, breast cancer, liver disease, pneumonia and malaria. Using Flask, the authors of this paper build a web app that can forecast several diseases, including those on the aforementioned clinical spectrum[18]. A CNN model predicts malaria and pneumonia, whereas deep-learning models in this study predict breast cancer, diabetes, heart, liver and kidney disease. Based on the survey, existing models have numerous issues in achieving high accuracy, F1-score, patient monitoring and predictive performance ratios. Hence, this research proposes Deep Convolutional Neural Network-assisted Chronic Disease Management (DCNN-CDM) through pharmacogenomics and an improved predictive analytics model to enable informed real-time decision-making at the point of care.

3. Methodology

As part of chronic disease management, patients can better take charge of their health by having their doctors consider their medical history and current state. After that, the doctor will advise a course of therapy for the patient's condition. A system of care and payment that considers the progression of a condition over time to provide the best possible therapy in a short period. A directed, integrative, and

tech-supported strategy benefits the paradigm change toward precision medicine. The field of pharmacogenomics is pivotal to this shift because of the invaluable information it provides on the individual heterogeneity in drug responses. Personalized treatment uniquely suited to each patient's requirements may be achieved using genetic information. Hence, this study proposes Deep Convolutional Neural Network-assisted Chronic Disease Management (DCNN-CDM) through pharmacogenomics and an improved predictive analytics model to enable informed real-time decision-making at the point of care.

4. Results and discussion

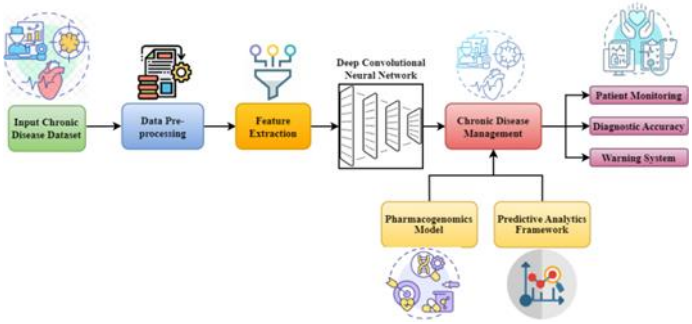

Figure 1. Proposed DCNN-CDM model.

Figure 1 shows the suggested DCNN-CDM model. The data are taken from the Chronic Disease Kaggle Dataset [19]. Data pre-processing includes re-sampling to handle the dissimilar rate of irregular events and the latest observation carried forwards (LOCF) interpolation for null points or inconsistent frequency. DCNN can train weights automatically without human feature extraction and is very good at extracting and expressing features. The objective of this model is to aid patients in detecting illnesses and doctors in quickly generating treatment plans by achieving accurate and efficient chronic disease diagnosis via feature extraction and classification using a DCNN and an ensemble learning algorithm. There are several ways in which pharmacogenomics could improve hypertension therapy in clinical practice. Treatment based on genetic architecture rather than doctors' empirical recommendations may prevent chronic kidney disease (CKD) and cardiovascular disease, among other benefits, as can the efficient improvement of ideal control of blood pressure rates and the prediction of medication responses. By harnessing past patient data, risk factors, and other relevant features, predictive analytics may identify people susceptible to acquiring a particular illness, making it an invaluable tool for early chronic disease identification. Medical professionals can enhance the treatment of chronic diseases, provide prompt medical advice, and provide early warning signs. The outcomes of the decision-making process may help clinicians manage chronic diseases, respond to warnings, provide medical guidance quickly, and comprehend patient information in real time. For the early diagnosis of chronic illnesses, this study denotes the labelled training dataset as $D = (Y, x)$, where $Y \in \mathbb{R}^{m \times d}$ is the set of m sample with d feature; $x = (x_1, x_2, \ldots, x_m)^T$ is m-dimensional column vectors, and $x_j \in \{-1, 1\}$, $\forall j = 1, 2, \ldots, m$. Here, $x_j = 1$ means that the jth sample is labelled as a sick case, and $x_j = -1$ otherwise. The N-order multivariate polynomials on the samples $y_j = (y_{j1}, \ldots, y_{jd}) \in Y$ is inscribed as

$$q(y_j) = \sum_{i=0}^{N} \sum_{\beta(i)} s_{\beta(i)} \prod_{w=1}^{d} y_{jw}^{\beta_w^{(j)}} \quad (1)$$

As shown in equation (1) where $\beta^{(i)}$ is a d-dimensional vector composed of a non-negative integer and $s_{\beta(i)}$ is a coefficient of the monomial $\prod_{w=1}^{d} y_{jw}^{\beta_w^{(j)}}$ is of degree i represents the values of every polynomial q on m samples by linear projections.

DCNN is an end-to-end model that trains multi-level features successfully using multi-layer convolutional processes. The local high-level ECG features were essentially obtained by assembling

low-level features and enriching their levels with numerous stacked layers. This research used a 1D convolutional neural network (CNN) that propagated across time to derive feature representations of the electrocardiogram (ECG) beat in a one-dimensional temporal space. The input signal y_{1d} of length K^t had been convolved with the overall number of filters $m^f = 32$, with everyone having kernel sizes $L = 32$. The kernel in the convolution layer had been prepared by Glorot Uniform, which initialize the convolution weights S_{1d} based on uniform distribution within the ranges $[-limit, limit]$ where $limit = \sqrt{\frac{6}{f_{in}+f_{out}}}$ where f_{in} is the number of input unit and f_{out} is the number of output unit.

To evade overfitting and overtraining due to cohort heterogeneity, this study executed a 20% dropout, nullifying the influence of 20 % of the convolutional weight. This can be expressed as an expression with y_{1d} as ECG sequences, S_{1d} as 1-D convolutional weight in equation (2),

$$x_{1d} = [Dropout(S)]_{1d} \otimes y_{1d} + a \quad (2)$$

As expressed in equation (2), where S denotes the convolution weights, and a indicates the bias.

A patient's EHR is a series of temporal healthcare event $PE = N_j, t_j$ where N_j denotes recorded healthcare events such as medication or lab tests, and diagnosis t_j denotes the timing when N_j occurred. This system is based on the attention graph depiction and adjusts the elements of the attention graph depiction to match the features of EHR information. Firstly, this study defines the attention graph for a patient's EHR as a connected directed graphs $H = (U, E, \delta, \omega)$ where functions $\delta : U \rightarrow \{B\}$ maps every nodes to a set of heterogeneous attribute. The key variance between this description and the prior one is that, instead of using attributes in Euclidean space, a directed network with heterogeneous characteristics is built. Therefore, this study avoids misleading the similarity between the medical event. Graphs H have positive edge weightings $\omega : E \rightarrow R + 0$, computed from the temporal interval.

$$\omega(j, i) = \sum_{n,m} \exp(-\alpha \cdot |t_n - t_m|) \quad (3)$$

This research must first generate the receptive fields and then design a convolutional operator to procedure the convolutional in every receptive field. Heterogeneous convolution, the first convolutional layer, is where this takes place. The operation objects of the receptive fields may be signified as a list of attributed edge, as each node represents some medical event. Diagnostic findings, such as the International Classification of Diseases (ICD-9) codes, often describe these events. The following formalization of the mathematical representation is possible:

$$RF(j) = \{Dy1, \dots Dy(l)\} \quad (4)$$

$$OI(j) = \{BE(1), \dots BE(l+1)^2\} \quad (5)$$

$$BE(n) = \begin{cases} \{DyB, DyA, \omega_{DyB-DyA}\} \\ DyB, DyA \in RF(j) \end{cases} \quad (6)$$

As inferred from equations (4) to (6), where the $RF(j)$ signifies the list of node corresponding to the therapeutic event in the jth receptive fields, $OI(j)$ denotes the start and end nodes, and the edge weights signify the list of attention edges and every attributed edge. Defining Diagnosis B as being closer to Diagnosis A than Diagnosis C is impractical, as previously mentioned. Thus, this study starts the distance amongst every medicinal occurrence at unit sizes to prevent any potential misinterpretation of the notion of similarity. The medical event is represented by converting it to a binary vector. This is the conversion function:

$$G = Dy(j) \rightarrow R^d \quad (7)$$

$$G = 1, G(j) \in \{0,1\} \quad (8)$$

The rectified linear units (ReLUs), which solved the issue of vanishing gradients, have been used as the activation functions in the hidden layer. To speed up model training and avoid overfitting, batch normalization was performed for every layer after the activation functions. To decrease overfitting, this research used dropouts after batch normalization. Since this research is content with a true/false outcome, sigmoid activations were used for the output layer. The optimizer and the loss function utilized Adam and binary cross entropy (BCE), respectively. Since the study's output was binary, BCE was chosen for the analysis. The equation (9) that defines BCE Loss is.

$$BCE\ Loss\ (P,T) = \frac{1}{m}\sum_t (T[j] * \log(P[j]) + (1 - T[j]) * \log(1 - P[j])) \quad (9)$$

As shown in equation (1), BCE Loss equation, where P indicates the predicted values, and T denotes the ground truths.

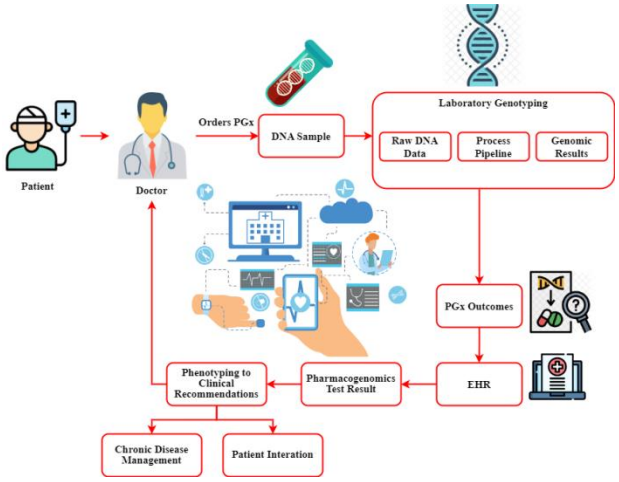

Figure 2. Pharmacogenomics (PGx) test for Chronic Disease Management

Figure 2 shows the Pharmacogenomics (PGx) test for Chronic Disease Management. Everyone desires to be in charge of their PGx findings and share them with family and friends. The advantage of PGx findings is that they last a patient's lifetime, unlike tests like bloodwork that need to be repeated since germline DNA does not alter. Much work has been done to help family members and doctors share PGx data despite research concentrating on EHR PGx integration within health systems. After evaluating the patient's need, the doctor must first order a PGx test. The laboratory will send a DNA collection kit to the patient once they have the request from the doctor. These days, it's common practice for patients to get their PGx test results in a static text format, such as a PDF. Results from PGx tests may be easier to discuss with present and future healthcare professionals if an auxiliary genomics system could store and exchange discrete, calculable data. Laboratory findings are sent to an auxiliary patient-managed genomic system (PMGS) in a machine-readable format when prescribers request a PGx test using an electronic health record (EHR) or a web-based platform for chronic illness point-of-care. This system would allow patients to access and handle their results. An interpretable form of the clinically relevant PGx findings will be kept in the PMGS. Everybody with an internet connection or a mobile device may see their PGx test results in the EHR.

Simulation Results

The data are taken from the Chronic Disease Kaggle Dataset [19]. Utilizing a consensus-based set of 124 indicators, the Centers for Disease Control and Prevention (CDC) Division of Population Health enables territories, states, and large metropolitan regions to consistently describe, gather, and report data on chronic illnesses. This data is vital for public health practice and is accessible to these entities.

The CDI website serves as a portal to supplementary data and information resources and offers access to indicator data that is particular to states. People have taken health-related surveys throughout the United States at different times and locations for the last 15 years. The data is presented with demographic stratification and confidence ranges.

Accuracy Ratio

This research aims to prove that the DCNN can reliably forecast chronic illnesses despite missing data. At the beginning, the network is given a set of input-output values. Afterwards, the outputs are computed by training the backpropagation algorithm to use suitable connection weights. A comparison with known values is then used to measure the accuracy of the expected outputs. By comparing these values, error signals are generated and sent back and forth between the layers. Next, the network modifies and refreshes the weights using the new information. The network learns to change the weights and arrives at predictions that show a small deviation from the actual values by repeating these training iterations.

$$Accuracy = \frac{TP+TN}{TP+TN+FP+FN} \quad (9)$$

As shown in equation (9), FP, TP, FN, and TN correspondingly signify false positives, true positives, false negatives and true negatives. Figure 3 shows the accuracy ratio.

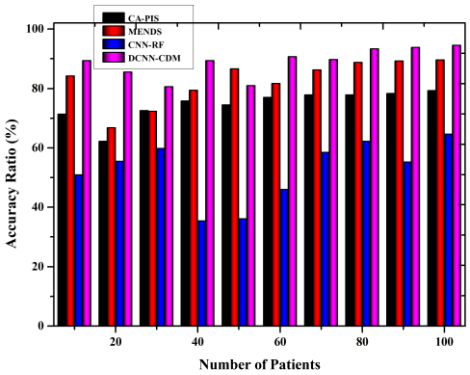

Figure 3. Accuracy ratio

Patient Monitoring Ratio

With machine learning and cloud computing, medical professionals may store patient data remotely and retrieve it whenever needed, enabling them to do remote health monitoring. Instruments that measure and analyze physiological and biometric data are necessary for remote patient monitoring. Integrating ML with the Internet of Medical Things (IoMT) allows healthcare practitioners to remotely monitor their patient's health status, eliminating the necessity for in-person assessments. This is very helpful for managing chronic diseases and recovering from surgeries. Figure 4 shows the patient monitoring ratio.

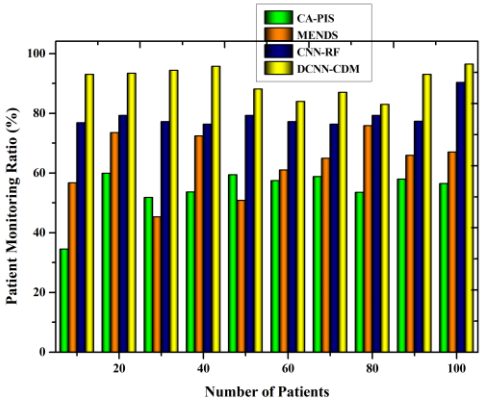

Figure 4. Patient Monitoring Ratio

Predictive Performance Ratio

The research trained the model with several feature sets and retrieved the best performance model for platform arrangement to guarantee the model applies to the real world. The best result was achieved using every feature in the prediction. The model that relied only on automatically uploaded features performed well in prediction. This paradigm may improve the everyday lives of patients. The findings provide evidence that DCNN-CDM prediction relies on lifestyle and environmental data rather than clinical questionnaire assessment. DCNN-based closely mirrors a human's decision-making and has better explainability, predictive performance, and robustness to outliers. Figure 5 shows the predictive performance ratio.

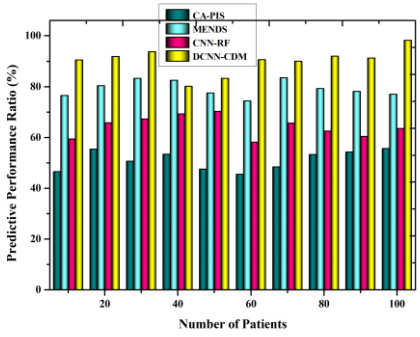

Figure 5. Predictive Performance Ratio

5. Conclusion and future scope

This paper presents DCNN-CDM through pharmacogenomics and an improved predictive analytics model to enable informed real-time decision-making at the point of care. DCNN has the advantage over other programs in analyzing large and complex data sets promoting wellness and chronic disease management. Improving the implementation of wellness programs related to chronic illnesses may be achieved by identifying groups of individuals in distinct behavioural risk categories. For instance, to create programs that mitigate and prevent the emergence of diabetes and similar chronic diseases, it is necessary to identify people at risk of contracting the disease. One of predictive analytics' many strengths is its ability to combine and evaluate information from several sources, including EHRs, wearable devices, medical imaging, patient-reported results and genetic data. By integrating various data, predictive models may find hidden patterns, correlations, and risk factors that can diagnose chronic illnesses early. Compared to conventional care models, pharmacogenomics may aid in the

future and accurate selection of prescription regimens. When pharmacogenomics is integrated into patient care, pharmacists have a unique chance to provide insightful recommendations

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