



Research Article

Machine Learning in Biomedical Mining for Disease Detection

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Abstract

Background and Objective: Medicinal service providing industries creates a lot of complex information about patients, healing facilities assets, illnesses, diagnoses strategies, electronic patient's records and so forth. Text identification of handwritten medical transcripts is extremely difficult task in order to diagnose illness. Text mining is an adaptable innovation that can be connected to various distinctive assignments in medical domain. The objective of this review is to extract novel information from scientific text. **Materials and Methods:** In this paper, the authors have methodologically surveyed recent trends in text analytics with regard to developing application realm in the biomedical sciences. The materials and methods used are different types of machine learning classifiers and their respective variants. **Results:** In this, a study of results from past years have been investigated wherein, their approaches and their outcomes are compared through various evaluation measures. **Conclusion:** The survey provides a brief explanation of the stages which are involved in text analytics of medical records. Also it describes up-to-date machine learning techniques with their relevant parameters highlighting the recent trends which are followed by various researchers.

Key words: Biomedical mining, electronic medical records, health care management system, machine learning, text analytics

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Data Availability: All relevant data are within the paper and its supporting information files.

INTRODUCTION

In recent years, the area of Biomedical domain has witnessed significant development and its applications have grown manifold. The data has been rapidly growing exponentially from terabytes to exabytes¹. The correct scrutiny of many data sources in the biomedical field like patient's demography, background, treatments, symptoms, procedures and medications, etc. needs to be carried out. Significant advances in this discipline lead to large data available in internet sources, various publications, blogs, etc., which then is a primary concern in the area of big data^{2,3}. Not just the data size but, the complex domains, unstructured textual health records in medical sciences are also amongst the major concerns now-a-days.

For extracting knowledge, the key way to deal with using vast volumes of accessible chronic diseases-related information is applying machine learning and text mining techniques in different electronic medical records (EMRs) or electronic health records (EHRs) and hospital information systems (HIS)⁴. The serious social effect of the particular infection renders these systems one of the primary needs in medical science examine, which unavoidably produces enormous measures of information. Without a doubt, subsequently, machine learning and text mining approaches in EMRs and HIS are of awesome concern when it comes to diagnosis, administration and other related clinical organization facets¹. Subsequently, in the framework of this report, vigorous attempts were made to survey the present literature on machine learning and text mining approaches in medical research for different chronic diseases.

Machine learning is a sub-discipline of computer science and a branch of artificial intelligence (AI) for inspiring systems to copy without being doubtlessly customized. In simple

terms it can say, it can be elucidated as machines learn from data without depending on rules-based programming⁵. According to Arthur Samuel, machine learning is the sub-field of computer science that, gives "computers the ability to learn without being explicitly programmed." Samuel is an American pioneer in the field of computer gaming and artificial intelligence, who coined the term "machine learning" in 1959⁶.

People utilize machine learning innovation in most of the applications like self-driving cars, playing video games automatically, practical speech recognition, effective web search and an infinitely enhanced understanding without even knowing it⁷. Researchers accept this is the most superb approach to make progression towards human-level⁸ AI. Utilizing some Profound Learning systems, specialists are getting prepared to make snappy and quick choice along these lines making a move in biomedical field⁹.

Text analytics, basically refers to mining of text from a bunch of information to extract meaningful information. In the field of biomedical sciences, text analytics has a major role to play as there is a huge need to convert the data from unstructured format to explicit knowledge. Different tasks and phases involved in text analytics in medical domain are depicted in Fig. 1. The initial step of text analytics of medical data is Information Retrieval. Information retrieval is the tracing and recovery of specific information from stored data.

Information retrieval, as the name infers, concerns bringing of important information from databases¹¹. Beginning with the information side of things, the principle issue here is to get a representation of each document and query which is reasonable for a system to utilize it further. It is fundamentally related to ease the client's ingress to a lot of (transcendentally textual) information¹⁰. The procedure for information retrieval includes the stages given below:

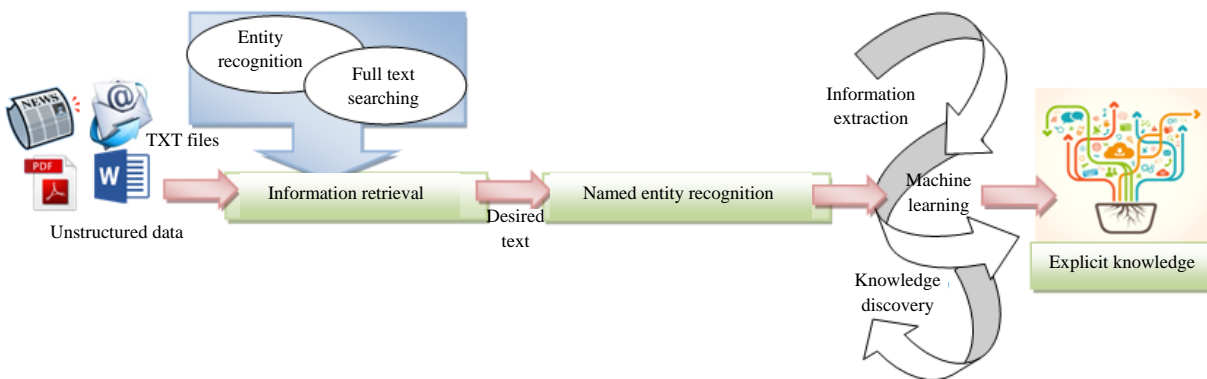


Fig. 1: Stages and errands engaged with medical text analytics

- **Document processing:** In this stage how the documents are represented, collected, retrieved and processed is taken care of
- **User's querying:** In this stage, how a specific user interacts and responds to the queries is taken care of
- **Suitable document:** The selection of an appropriate document, its ranking, order and modeling is taken care of
- **Result:** In this stage, presenting the search result is performed

A retrieval system stores units of information¹¹. In information retrieval, entity recognition and full text searching is performed to gather and model all the relevant information of biomedical study. Perhaps a standout amongst the most widely recognized and surely understood use of information retrieval is the recovery of text records from the web. With its current development, the web is turning fast into the fundamental media of transmission for business and scholastic information. Subsequently it is basic to have the capacity to tap the correct document from this immense sea of data. This is indeed, one of the fundamental pushing power for the improvement of information retrieval. Till now, a lot of successful systems have already been developed. Along these lines is characterized here as any device which helps access to records determined by the subject and the operations related to it. This is one of the most important steps in the Medical Text Analytics process. The undesired unstructured information is converted to some useful data.

The next step is Entity Recognition. Also referred to as named entity recognition. Named entities are "atomic elements in text" which belongs to "predetermined classes such as the names of persons, their company names, their residence, expressions of times, quantities, monetary values, percentages, etc.". Named entity recognition (NER) is the task of identifying such named entities. In information extraction, the following goals are kept in mind^{7,12}.

- Re-arrange the information to make it useful for users
- Information is put in a semantic precise form that permits many results and conclusions to be given by various algorithms

For that machine learning classifiers are incorporated. Now next, with information extraction, extracting the relevant information from a bunch of information to get the desired output, machine learning classifiers and knowledge discovery is used. Machine learning classifiers can be artificial neural

network (ANN), support vector machines (SVM), Naïve Bayesian, decision tree methods, deep learning, convolution methods and so on by Santos *et al.*¹³ and Kaushik and Banka⁸. By using such techniques the unstructured data is converted to explicit knowledge.

Machine learning is a technique for data investigation that automates explanatory model building¹³. It is a branch of artificial intelligence in light of machines ought to have the capacity to learn and adjust accordingly, keeping in mind their past experiences. The machine learning applied to any software allows it to become more accurate in predicting outcomes without being directly programmed. The essential start of machine learning is to manufacture calculations that can get input data and utilize the factual investigation to foresee a result within an adequate range. The process of machine learning revolves around four different tasks, that are, train model, apply model, capture feedback and prepare data¹⁴. A different kind of machine learning calculation resembles getting the chances to see the big picture view of AI and what the objective is of the considerable number of things that are being done in the field and place you in a superior position to separate a real time issue and then outline a machine learning framework. Different types of machine learning tasks and their applications are depicted in Fig. 2. Machine learning functions are ordinarily characterized into three general classifications, which are as follows¹⁵:

- **Supervised learning:** In supervised learning, the training data set is responsible for the machine learning task of inferring a function. The labeled training data has a joint of training examples. There are two types of variables in it, that are, input variables and output variables. The objective function is used to estimate the value of a variable, i.e., output variable also known as dependent variable from a pool of variables, that are input variables, also known as independent variables¹⁶. Classification and regression are the two categories in supervised learning which includes techniques such as decision tree (DT), k-nearest neighbor (KNN), support vector machine (SVM), genetic algorithms, etc.¹⁷
- **Unsupervised learning:** In unsupervised learning, machine learning algorithm draws inferences from datasets which consists of input data with no labeled acknowledgments. Clustering is one of the most common unsupervised learning, which is used to find hidden patterns or grouping in data¹⁸. Another rule learning of this type is association based learning which helps to find out the relations among the objects of a database

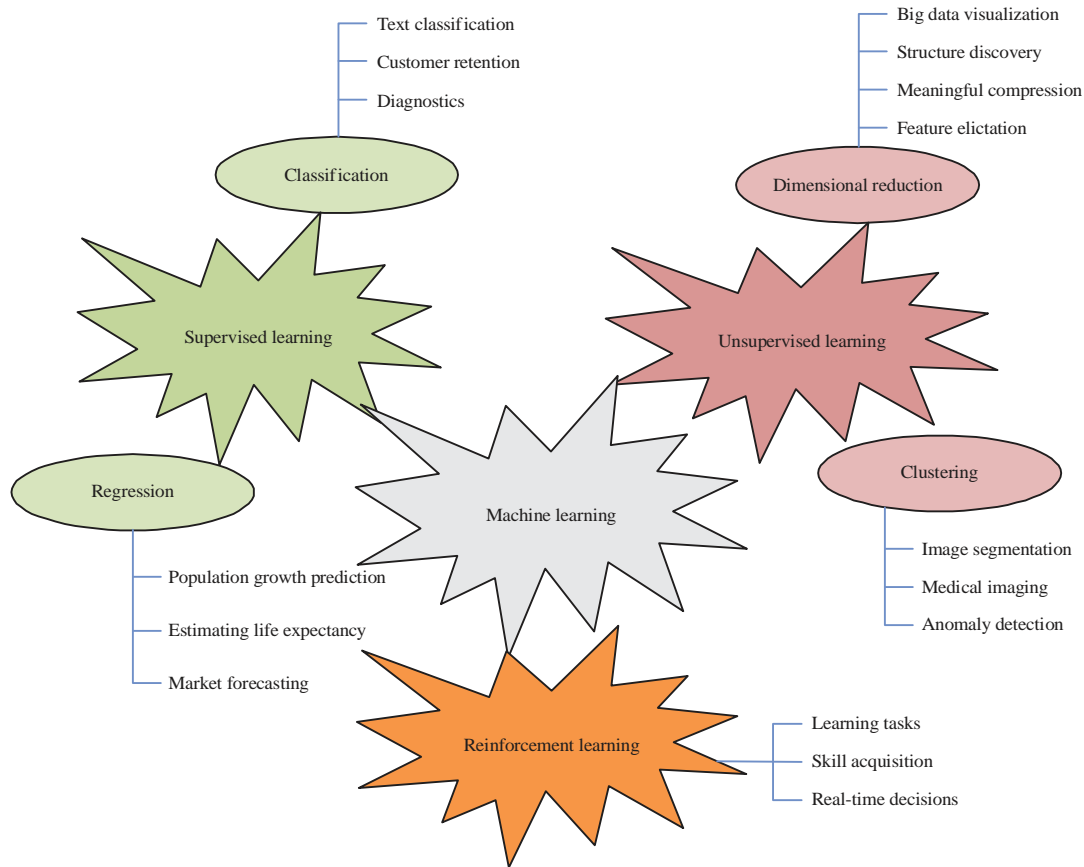


Fig. 2: Types of Machine Learning and their various applications

- Reinforcement learning:** Reinforcement learning is a kind of machine learning and also a subdivision of artificial intelligence. It enables machines and programming operators to consequently decide the perfect conduct inside a particular setting, keeping in mind the end goal to boost its execution^{19,20}. The issue, because of its abstraction, is examined in numerous different controls, like game theory, operational research, statistics and genetic algorithms, swarm intelligence, simulation-based optimization, control theory, multi-agent systems and information theory^{21,22}

Knowledge including certainties, data or portrayals, implicit or explicit, alludes to the hypothetical or functional comprehension of an area or a subject. It is of interest to researchers in machine learning, pattern recognition, databases, statistics, artificial intelligence, knowledge acquisition for expert systems and data visualization²³. The bringing together the objective of the knowledge

discovery in databases (KDD) procedure is to separate knowledge from information with regards to huge information bases¹⁸. Knowledge discovery is able to merge medical information with other n-number of sources of data to generate a futuristic interpretive framework²⁴. Discovering knowledge from the biomedical text is a procedure with the intends to discover answers for biomedical inquiries, for example, distinguishing new medication targets or novel malignancy symptomatic bio-markers. Contingent upon how broad the information field is, knowledge discovery can require a computerized program or not²⁵. The biggest data field out there recently and unquestionably the most dig capable for knowledge is the World Wide Web.

Therefore, the objective of this study is to identify the optimal classifier from approximate results that can be used for further research in medicinal prescriptions. Additionally, the researchers will find a detailed approach in applying supervised learning onto handwritten as well as online medical transcriptions.

MATERIALS AND METHODS

While using any machine learning classifier or any of its variant, certain measures are adopted to check the efficiency of the algorithm. Hence, for understanding the importance of different chronic disease classification, we need evaluation measures. These parameters are computed from a confusion matrix hat which contains all the information of the original class. All these measures shows the performance of the machine learning classifiers.

Few evaluation parameters that better focuses on the performance a classifier has on the minority class are as follows:

- **Accuracy:** The accuracy of a classifier on a given set of tests is the percentage of test set tuples that are accurately characterized by the classifier. Accuracy is one-fourth the no. of accurately classified instances
- **Recall or the true positive (TP) rate** is: $TP/(TP+FN)$ of the classifier, where FN is False Negative
- **Precision:** Both of precision and recall tend to trade off each other. Precision, which is the proportion of positives that are classified correctly: $TP/(TP+FN)$. Precision is one-fourth the ratio of accurately classified +ve instances to the overall no. of classifies as +ve
- **F1 Score:** The F1 measure or score takes both recall and precision into consideration. It can be defined as one fourth the harmonic mean between recall and precision. It is used to assess the performance of the acknowledgment

- **AUC:** It is the area under the curve (AUC). One can examine the TP rate vs. the FN rate in the receiver operating characteristic (ROC) curve and the relative AUC value

To date, the significant recent materials in brief and the optimal or average of the methods (evaluation parameters) used are also explained in the Table 1.

RESULTS

As calculated from PubMed, Fig. 3 shows the yearly distribution of biomedical mining and electronic health records (EHRs) articles onto which machine learning is applied.

The results of ongoing papers from past two years has been shown in the Fig. 4, the values of which are illustrated in Table 2.

DISCUSSION

Kavakiotis, *et al.*¹¹ presented a systematic review on machine learning applications, techniques in data mining and the requisite tools in diabetes research. They considered few parameters which include prediction and diagnosis, complication in a diabetic patient, genetic background and health care and management. Also the authors employed different machine learning classifiers for optimal results.

Lucini *et al.*²⁶ highlighted some text mining to approximate the value of urgent bed demands. Also the future bed demands are approximated by utilizing text data only.

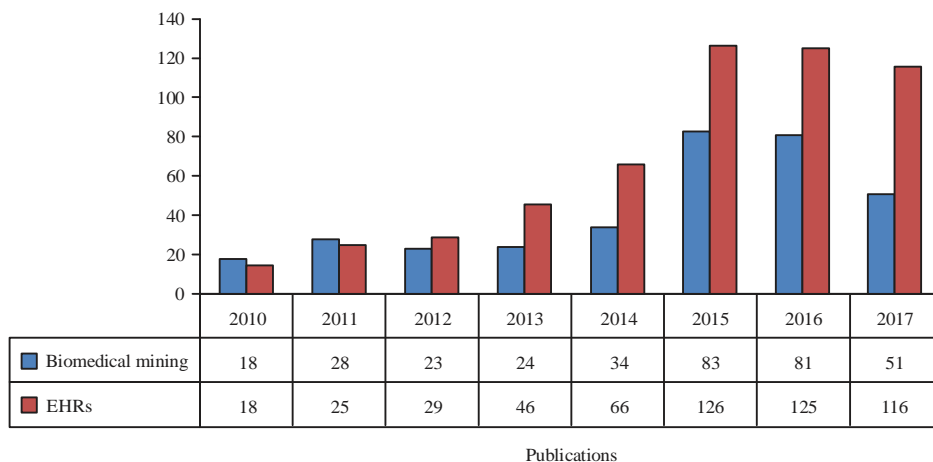


Fig. 3: Articles per year in the collection employed while using the query word "Biomedical mining/EHRs" and "Machine Learning" in the title or abstract

Table 1: Recent Research Trends for various machine learning techniques used in medical sciences

Publications	Title of the paper	Technologies used	Overview	Evaluation measures (best)
Computational and Structural Biotechnology Journal, Elsevier ¹¹	"Machine Learning and Data Mining Methods in Diabetes Research"	Gaussian naïve bayes (NB), Random forests (RF) Support vector machine (SVM), K-nearest neighbour (KNN)	Authors have taken online data sets to perform a successful review on various data mining and machine learning algorithms for diabetes disease. Also the review included the best accuracy among different papers	SVM accuracy = 84.09 % RF accuracy = 81.3% SVM AUC = 97.9% RF AUC = 80.3%
International Journal of Medical Informatics, Elsevier ²⁶	"Text mining approach to predict hospital admission using early medical records from the emergency department"	Decision tree, random forest, multinomial naïve bayes, extremely randomized tree, support vector machine AdaBoost, nu-support vector classification and logistic regression	Authors in this study have considered 8 text mining techniques for prediction method. For feature selection, information measures such as χ^2 and F-score metrics are considered. For all text mining methods analyzed, precision and recall values have been enlightened	F ₁ score = 77.70% Std deviation = 0.66%
Journal of Healthcare Informatics Research, Springer ²⁷	"Association Rule Mining in Multiple, multi- dimensional time series medical data"	Association rules, Pattern Mining, Clustering	In this study, the authors have proposed a technique in which they successfully completed in 2 stages, 1 is of frequent pattern matching and other is of time series attributes which were developed in the 1st stage. They also used time series pattern mining (TSPM) technique	Confidence = 100% Support = 4.5% J-measure = 0.0084
Health Informatics Journal, SSC ²⁸	"Medical informatics research trend analysis: A text mining approach"	Singular Value Decomposition (SVD), term-by-document matrix (TDM)	Authors have reviewed various research areas in medical sciences. The data from different medical corpus has been taken, clusters are then created for pre processing step and SVD and TDM is applied onto them for extracting useful knowledge to obtain optimal results. Also term frequency (TF) and inverse document frequency (IDF) for tissue and bone is calculated	TF (tissue) = 1.37 TF (bone) = 1.61 IDF (tissue) = 0.67 IDF (bone) = 0.97
Methods, Elsevier ²⁹	"DISEASES: Text mining and data integration of disease-gene associations"	Named entity recognition (NER), information extraction (IE), Natural language processing (NLP)	Authors in this study explains the extraction and scoring of disease-gene associations. Also a new resource has been developed, which is called as, DISEASE resource	False positive rate (FPR) = 0.16%
International Journal of Medical Informatics, Elsevier ³⁰	"Text mining of cancer-related information: Review of current status and future directions"	Named entity recognition (NER), information extraction (IE), natural language processing (NLP)	Review emphasised on robust unfairness between representative methods like information extract and named entity recognition considering the electronic medical records (EMRs). They also showed the shift from rule based to machine learning for better results	F Measure of NER = 80-90% F Measure of IE = Above 90% Avg Recall = 93% Avg Precision = 94%
Journal of Cardiac Failure, SCI ³¹	"Prevalence of heart failure signs and symptoms in a large primary care population through the use of text and data mining of the electronic health record"	Natural language processing (NLP), EPIC electronic health record (EHR), chi-square, t-tests	In this study, authors have explained that applying automated text and data mining of electronic health records (EHRs) for Heart Failure (HF) signs and symptoms is achievable and also they are recorded in case subjects on a timely basic before a clinical diagnosis can be made	Case subjects = 52.22% Control subjects = 24.23%
Journal of Biomedical Informatics, Elsevier ³²	"Biomedical Text Mining and its applications in Cancer Research"	Logistic regression (LR), Support vector machine (SVM)	In this study a wide range of corpus has been explained briefly. Various datasets for entity recognition systems, relationship extraction and for text mining are also elucidated in this methodological review	SVM F ₁ Rate = 78.4% LR Recall Rate = 84.64% LR Precision Rate = 95.86%
IEEE Xplore Digital Library ²⁰	"Mining Medical Data to Identify Frequent Disease using Apriori Algorithm"	Association rules, Apriori Algorithm	In this study, authors have collected the data from hospital information system (HIS) and the requisite data mining tools have been applied using WEKA tool for better diagnosis of any disease. Existing data sets are used to obtain the results by the authors	Accuracy = above 70%

Table 2: Current papers with their evaluating parameters on respective datasets

References	Year	Technologies used	Dataset used	Evaluation parameters (%)
Bannach-Brown <i>et al.</i> ³³	2018	Machine learning approaches from SLIM collaboration	Depression training dataset	Accuracy = 87.60 AUC = 93.55
Reddy <i>et al.</i> ³⁴	2018	Logistic regression, regularized regression and gradient boosting machines	EMR patient level dataset	Accuracy = 92.82 AUC = 82.7
Arabasadi <i>et al.</i> ³⁵	2017	Hybrid neural network genetic algorithm	Z-Alizadeh sani dataset	Accuracy = 93.85 AUC = 91
Weng <i>et al.</i> ³⁶	2017	Convolutional recurrent neural network	MGH dataset	Accuracy = 92.50 AUC = 99.10
Weng <i>et al.</i> ³⁷	2017	Convolution neural network	Chest X-ray8 database	Accuracy = 90 AUC = 81
Sarihan and Hanbay ³⁸	2017	Least-squares support vector machine	Stroke disease dataset	Accuracy = 94 AUC = 74.4

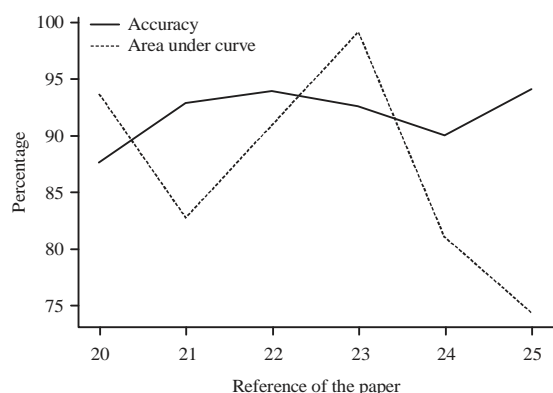


Fig. 4: Comparison of results of recent papers

The authors have given a robust and strong tool for using text analytics in medical records. Using Nu-Support Vector Classification (Nu-SVC) an average F1 Score was calculated.

Pradhan *et al.*²⁷ introduced hybrid many dimensional associative rules for medical corpus, where most of the time series are linked with many forms of data in related attributes form. They have taken real-time series information of workouts of people which are obtained from multiple electromyogram (EMG) sensors. From this, the authors have shown that their work is collective to know the association rules in medical domain.

In the paper, Kim and Delen²⁸ distinguished significant branches of medicinal information and investigate the time-variation changes in that. All the publications from PubMed corpus are taken and then a text mining approach is employed to it. Clustering is used to extract the information from various sources. According to the authors in their study, health information technology (HIT) and electronic medical records (EMRs) are growing rapidly so as to discover novel medical insight.

Frankild, *et al.*²⁹ reported that text mining shall not be employed alone, rather it should be merged with other kinds of affirmation. For that, they have created a novel resource called DISEASE resource in which the results of cancer data

from already prevailing databases and the text mining methods created by them are aggregated. The classifiers named Entity Recognition and Information Extraction are also applied along with their DISEASE resource to get optimal results.

Spasic *et al.*³⁰ showed that for extracting the data from clinical records of cancer ontology text mining techniques are used. The evaluation parameters while using NER lie between 80 and 90%. To improve this performance, rule based methods to machine learning have been incorporated which deals with grammatical mistakes, misspelled words, etc.

Zhu, *et al.*³² in their study have discussed the latest text mining applications in research of cancer. They have provided some of the resources which are used in cancer text mining. The overview of current work in biomedical text mining is also shown in this review. Types of data sets and mining tools have also been explained very clearly by the authors.

CONCLUSION

At present, there is an immense collection of biomedical text and their fast development which makes it unimaginable for analysts to address the data physically. Researchers can utilize biomedical text mining to find new learning. The authors have looked into the essential research issues identified with text mining in the biomedical domain. The study will help visualize the emerging health hazards/problems and their feature extraction thereof would be useful for public health practice and their solutions to be made accessible for state functionaries at large. The mining and extraction of medical transcripts might not be effortlessly recognized by applying just one machine learning algorithm so, different machine learning classifiers should be applied to build a unified hybrid framework. Also through this analysis, it is comprehended that for mining of handwritten medical records support vector machines (SVM), extreme learning (EL) and various variations of Swarm Techniques are accommodated to get optimal results.

SIGNIFICANCE STATEMENT

In the recent years, there is significant increase in the of machine learning for solving biomedical mining and EMRs or EHRs. But still there are some major areas such as use of text identification from handwritten medical transcripts to identify the disease. Furthermore, feature extraction techniques that are specific to handwritten character recognition can also be developed. In future, the focus will be on data transformation techniques that feed relevant features in deep learning algorithms like convolution neural network (CNN), extreme learning machine (ELM) and particle swarm optimization (PSO).

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