Medical Diagnostic Decision Support Systems Based on Artificial Intelligence Algorithms

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Abstract—The idea of using AI algorithms for diagnosing and detecting pathologies in the human body is currently very popular. The reason is that the artificial intelligence technologies available to researchers make it possible to analyze information almost intuitively, to detect similarities between non-obvious patterns, and find an effective solution through training on the existing research and practical experience of specialists. As a result, diagnostic decision support systems based on artificial intelligence algorithms are increasingly being used in practice. This review examines the main artificial intelligence algorithms used in modern diagnostic decision support systems and illustrates their application for the diagnosis of several key socially significant diseases. Key issues related to the application of artificial intelligence systems in disease diagnosis are also discussed.

Keywords—clinical decision support systems, cognitive computing, deep learning, diagnostic decision support systems, machine learning.

I. INTRODUCTION

Clinical decision support systems (CDSS) are a natural stage in the development of medicine.

Information about the first CDSS began to appear in the 1970s. These were predominantly products used for academic purposes and not intended for real clinical applications [1, 2]. Systematic work in this area aimed at application in practical health care began in the 1990s – 2000s, and in the last decade, there has been a sharp, almost exponential increase in interest in this topic (Figure 1). This growth naturally coincided with several technological breakthroughs in pharmaceuticals, biology, medicine, information, and computer technologies. An important factor in the development of CDSS was the accumulation of huge amounts of biological and clinical data, the processing of which is already unthinkable without the involvement of modern computer algorithms. Increasing demands on the quality of healthcare services amid increasing economic pressure on healthcare systems have become another powerful incentive for the implementation of CDSS [3, 4].

Figure 1. The Annual Number of Scientific Publications in the PubMed Database Containing the Phrase "Clinical Decision Support System" in the Title or Abstract. Around the Turn of 2008–2010, There Started an Exponential Growth of Interest in This Topic.

Source: PubMed database (<https://pubmed.ncbi.nlm.nih.gov>)

Due to these factors, in recent decades, dozens of different CDSS have been actively developed and put into practice in a wide variety of therapeutic areas. The tasks performed by the CDSS are very diverse. They include diagnosis of diseases, control and monitoring of patients, prescription of medications, and much more [3]. Most CDSS use web applications that can be administered from a computer, smartphone, or other devices.

CDSS can be classified into various categories and types [4]. Early CDSS were rule-based, and the rules were based on literature, clinical practice, or patient information.

However, an obvious modern trend is the active implementation of artificial intelligence (AI) algorithms in CDSS (Figure 2). AI-based CDSS are typically composed of three main components (1) knowledge base with relevant clinical, biological, and chemical data; (2) AI algorithm used to model the decision; (3) inference engine which uses AI-determined decisions and applies them to the patient's clinical data to generate a recommendation. The latter is presented to the end-user (e.g. physician) through the communication interface.

Figure 2. Key Components of AI-Based CDSS

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For decision-making in medicine, today the gold standard is a multicenter, randomized, blinded study with a large sample, which allows obtaining statistically significant results [5]. However, the study result is influenced by many additional factors that can lead to systematic errors. An analytical approach to medical research does not always provide reliable results.

The statistical approach to clinical trials has some problems inherent in the methodology [6]:

- If all values that are beyond a certain statistical limit (for example, beyond the 80% cut-off point) were considered pathological, then the prevalence of all diseases would be the same and amount to 20%.
- There is no complete correspondence between the degree of statistical "unusualness" of the measured value of the indicator and the disease.
- The risk of the disease correlates with the indicators of many clinical and laboratory tests over the entire range of their possible values (ie, at different cut-off points of the norm). For example, for high normal and low normal cholesterol levels, the risk of coronary heart disease differs several times. The same happens for the levels of blood pressure.
- Some extreme, clearly unusual values of the indicator for a particular patient may be more informative than the usual values of this indicator.

As a result of the analytical approach to clinical trials, systemic errors at the stage of selection, treatment, dropout, and evaluation of outcomes are multiplied, which can ultimately neutralize the value of research findings. A large amount of new knowledge in the fields of cloud technologies, radionics, PACS (Picture Archiving and Communication System, DICOM image transmission and archiving systems) and methods of complex dynamic studies, new algorithms for diagnosing lung diseases using several scanners and technologies (computer tomography (CT), magnetic resonance, positron emission tomography, acoustic radiation force imaging, as well as the flow of general clinical data, contribute to the professional burnout of doctors trying to assimilate this layer of diagnostic information [7]. Without formalization of the knowledge base, the data is lost irretrievably and becomes useless in the treatment of the patient.

In contrast to the analytical approach, the integrated version of the analysis of clinical observations allows taking into account many interdependent parameters, forming an electronic patient model with clinical recommendations for treatment. One of the technologies for integrally making decisions is the use of AI algorithms.

The idea of using AI algorithms for diagnosing and detecting pathologies in the human body is currently very popular. The reason is that the available AI technologies make it possible to analyze information almost intuitively, to detect similarities between non-obvious patterns, and find an effective solution through training on the existing research and practical experience of specialists. As a result, a variety of CDSS has emerged and is actively developing, known as diagnostic decision support systems (DDSS) [8].

In this mini-review, we take a quick look at the main AI algorithms used in modern DDSS and illustrate their application for solving some pressing healthcare problems. We will also discuss some of the challenges associated with the use of AI in disease diagnosis.

II. BASIC AI ALGORITHMS IN DDSS

Developers approach the functioning of AI systems from different philosophical positions [9]. Symbolists use methods based on high-level "symbolic" (human-readable) representations of tasks, logic, and search. Connectionists copy the structure of a neural network as much as possible; this group relies on feedback to solve a problem. Evolutionists take evolutionary biology as their starting point; this group relies on genetic programming to solve a problem. Bayesians rely on statistical inference to solve a problem. Analogists choose psychology as their starting point; to solve any problem, this group relies on multi-core machines and their interactions with each other according to certain laws. Someday these approaches will reach a singularity point, which could lead to the emergence of a truly powerful AI.

The main AI algorithms currently used in DDSS are machine learning methods [10]. Machine learning uses large amounts of data and reveals subtle interdependencies between variables. As a result, it can detect hidden patterns in data series, analyze the influence of input parameters on outputs, generate new hypotheses, and direct research in the most promising directions.

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Machine learning algorithms fall into two main categories: supervised and unsupervised ones [11]. In supervised methods, both input and output parameters of the training set are taken into account when building a model.

Such models are widely used in the construction of classification and regression models. Unsupervised learning does not require labeled data. It is aimed at revealing hidden patterns present in the studied samples and is often used to explore data and create new hypotheses.

TABLE I
KEY AI ALGORITHMS USED IN MODERN DDSS

ML types	Description	Examples of algorithms
Supervised	A labeled training dataset is required. The system is trained with human feedback. Applications include Classification, Regression, and Prediction; ideal for modeling disease prognosis or treatment outcome.	Artificial Neural Network (ANN), Support Vector Machine (SVM), Random Forest (RF), Decision Tree (DT), Gradient Boosting (GB), Fuzzy Logic (FL), etc.
Unsupervised	Non-labeled data are used for model generation. Applications include mainly pattern recognition; ideal for modeling disease mechanisms, identifying hidden patterns in genotype or phenotype data.	Clustering methods, Self-Organizing Map (SOM), etc.

Deep learning is a modern subfield of machine learning that mimics the work of the human brain. Typically, for this, several layers of artificial neural networks are used to create predictive models based on training datasets. Models based on deep learning algorithms have many parameters and layers. In biomedical applications, deep learning represents a modern, high-performance alternative to more traditional machine learning methods [12]. When developing DDSS, one of the most popular deep learning algorithms is convolutional neural networks (CNNs). Deep learning algorithms are especially successful in the field of image analysis and recognition [13].

Cognitive computing, another type of AI technique, includes self-learning systems that use pattern recognition and natural language processing on semi-structured or unstructured data. Cognitive computing mimics the thought processes of the human brain to create automated computer models that can solve complex problems without human intervention. Currently, cognitive computing is considered a promising innovative methodology, potentially revolutionizing the field of support for medical decisions in healthcare [14].

III. DDSS IN VARIOUS FIELDS OF MEDICINE

This section contains actual examples of DDSS for the diagnosis of some socially significant diseases (Table 2).

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TABLE II
EXAMPLES OF REPORTED AI-BASED DDSS

field of medicine	program/algorithm	application	reference
oncology	IBM Watson for Oncology	a cognitive computing technology configured to support cancer research and treatment	Chen et al. [15]
	MLP, RF, DT, and SVM classifiers	prediction of breast cancer survival in patient records	Kalafi et al. [16]
	CNN classifier	diagnosis of neoplasms of the gastrointestinal tract using endoscopic images	Horie et al. [17]
	Gastrointestinal AI Diagnostic System (GRAIDS) based on a deep learning algorithm	diagnosis of upper gastrointestinal cancers using clinical endoscopy data	Luo et al. [18]
	a deep neural network classifier	classification of skin cancers	Esteva et al. [19]
cardiovascular diseases	CNN classifier	detection of arrhythmias on electrocardiograms	Hannun et al. [20]
	DT and GB classifiers	prediction of cardiovascular disease risk using routine clinical data	Weng et al. [21]
	SVM classifier	prediction of the development of restenosis in stents	Cui et al. [22]
	classifiers based on machine learning algorithms, including deep learning	early diagnosis of heart failure	Choi et al. [23]
	classifier based on a deep neural network	diagnosis of intracranial hemorrhage using computed tomography scans	Arbabshirani et al. [24]
neurology	FL classifier	diagnosis of motor, sensory, mixed neuropathies using clinical symptoms and diagnostic test results	Kunhimangalam et al. [25]
	FL expert system	determination of the severity of carpal tunnel syndrome using the results of nerve conduction studies	Kunhimangalam et al. [26]
endocrinology	classifier based on a deep neural network	diagnosis of type 2 diabetes mellitus using data from non-invasive sensors	Spänig et al. [27]
	CNN classifier	detection of diabetic retinopathy using retinal images	Gulshan et al. [28]
	SVM classifier	diagnosis of type 2 diabetes mellitus using patients' clinical data	Kuo et al. [29]

A. Oncology

Perhaps the most famous commercial product of this type is the IBM Watson system [15]. Its hardware part consists of 90 servers with eight-core processors and a total amount of random access memory (RAM) over 15 terabytes. This system was created to search for symptoms of oncological diseases in the database of case histories.

When a match or close values are found, the system issues the treatment that was prescribed to a similar patient. Despite the impressive resources invested in the development and promotion of the IBM Watson system, its practical application raises some questions related to the adequacy of the decisions made (see the next section).

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Kalafi et al. used several different machine approaches to predict breast cancer survival in 4,902 patient records [16]. The multilayer perceptron (MLP), RF, DT, and SVM classifiers were able to predict survivorship, respectively, with 88.2%, 83.3%, 82.5%, and 80.5% accuracy.

In the diagnosis of cancer, AI is currently used to the greatest extent in the analysis of biomedical images, including X-ray, CT, ultrasound images, etc. With the help of the methods described in the literature, pathologically altered tissue can be effectively distinguished from normal tissue, and the diagnostic accuracy in most cases exceeds even that of a highly qualified human expert.

As an illustration, let us look at the works devoted to the diagnosis of neoplasms of the gastrointestinal tract. Horie et al. demonstrated the diagnostic ability of the convolutional neural network to detect esophageal cancer, including squamous cell carcinoma and adenocarcinoma [17]. They used 8,428 esophageal cancer images from 384 patients at the Cancer Institute Hospital, Japan as a training sample. To assess the diagnostic accuracy, 1,118 test images of 47 patients with 49 types of esophageal cancer and 50 patients without esophageal cancer were used. The developed model was able to analyze endoscopic images in a short time with high efficiency. Thus, in the test sample, it found cases of esophageal cancer with a sensitivity and accuracy of more than 98%.

An even more ambitious example is the work of Chinese researchers who developed the Gastrointestinal AI Diagnostic System (GRAIDS) based on a deep learning algorithm for diagnosing upper gastrointestinal cancers by analyzing data from clinical endoscopy [18]. This multicenter diagnostic study was conducted in six hospitals in China. The study involved patients with malignant lesions of the upper gastrointestinal tract, including histologically confirmed esophageal cancer and gastric cancer. The images were randomly (8: 1: 1) divided into training and internal validation datasets for the development of GRAIDS, as well as a test set to evaluate the effectiveness of diagnostics. The GRAIDS scores were compared with those of endoscopists at three skill levels: expert, competent, and trainee. A total of 1,036,496 endoscopic images from 84,424 people were used to develop and test GRAIDS. GRAIDS achieved diagnostic sensitivity similar to that of an expert endoscopist (94.2% versus 94.5%) and superior to that of a competent (85.8%) and trainee (72.2%) endoscopist. The described study is a good, but still quite a rare example of systematic clinical validation of a predictive system based on AI.

AI is used to classify images of skin lesions [19]. AI has been proven to diagnose such diseases (including melanoma) as accurately as expert dermatologists. Given the universal availability of smartphones with the ability to receive and send skin imaging, these advances potentially provide universal access to diagnosis at the dermatologist level. AI also achieved the level of accuracy of a medical professional in interpreting mammograms in breast cancer screening (the area under the curve (AUC) for AI equals 0.840 compared to 0.814 for doctors) [19]. In addition, deep neural networks were able to detect enlarged lymph nodes or colon polyps on CT scans [30]. Numerous successful applications of deep learning in biomedical image analysis have been analyzed in some review publications (e. g., [31]).

The current status of the development of AI systems for the diagnosis of oncological diseases has been analyzed in several reviews [32, 33].

B. Cardiovascular Pathology

The literature describes numerous examples of AI-based diagnostic systems that surpass the assessments of qualified clinicians in the diagnosis of cardiovascular diseases.

A Stanford team of researchers has developed a deep CNN that outperforms a qualified cardiologist in detecting arrhythmias on an electrocardiogram [20].

Weng et al. used the DT and GB algorithms to predict the risk of cardiovascular disease in a large sample of approximately 380,000 patients in the UK [21]. AI methods were found to outperform the algorithms for assessing these risks recommended by the American College of Cardiology and the American Heart Association by 1.7% and 3.6%, respectively.

Cui et al. demonstrated the usefulness of the SVM algorithm for predicting the development of restenosis in stents [22]. Their model was 90% accurate using the levels of some blood plasma metabolites as inputs.

Choi et al. described the successful application of several machine learning algorithms, including deep learning, for the early diagnosis of heart failure [23].

A deep neural network was trained on more than 37,000 head computed tomography scans for intracranial hemorrhage and subsequently evaluated on 9,500 unseen cases, reducing time to diagnosis of new outpatient intracranial hemorrhage by 96% with an accuracy of 84% [24].

The use of AI-based systems in cardiology has been analyzed in detail in some review publications [34, 35].

C. Neurology

Kunhimangalam et al. developed an effective DDSS for the diagnosis of peripheral neuropathy using an FL algorithm [25]. With 24 inputs that include clinical symptoms and diagnostic test results, the system achieves 93.3% accuracy in diagnosing motor, sensory, mixed neuropathies, or normal cases.

Another FL expert system was developed to determine the severity of carpal tunnel syndrome (CTS), a peripheral nerve disorder [26]. The results of a nerve conduction study obtained from Kannur Medical College, India were used as training data. The system uses 13 input fields, which include data from diagnostic tests and clinical symptoms, and the output parameter is the severity of the disease. The diagnoses obtained coincide by 98.4% with the expert's assessment and provide high sensitivity and specificity.

In both of these cases, the FL algorithm was used. FL, a multivalued logic similar to human thinking and interpretation, has been actively used in medicine in the last three decades for the interpretation of clinical data, diagnosis, and treatment selection. Various FL-based DDSS have been developed in the field of diabetes, oncology, cardiovascular, central nervous system (CNS), and some other pathologies [36].

D. Diabetes

Diabetes is another major global health problem that AI systems can help solve.

Spänig et al. developed a DDSS based on non-invasive sensors and deep neural networks that interacted with the patient using a speech recognition system. The system can effectively diagnose type 2 diabetes mellitus [27].

The Google team has developed a deep CNN capable of detecting diabetic retinopathy (damage to the blood vessels in the eye) with high sensitivity and specificity [28]. When creating the model, a training set consisting of 130,000 retinal images was used. The effectiveness of the expert system was at the level of US-certified ophthalmologists.

Iranian scientists (published in 2019) carried out a meta-analysis of studies on the use of AI for the screening and diagnosis of type 2 diabetes mellitus [37]. Machine learning methods were applied in 71% ($n = 22$) of the total number of analyzed papers ($n = 31$). The most commonly used machine learning methods were SVM (21%) and naive Bayesian (19%). The most significant inputs used in the studies reviewed were body mass index, blood sugar, blood pressure, HbA1c, triglycerides, low-density lipoproteins, high-density lipoproteins, and demographics.

A recent study by Taiwanese scientists aimed at diagnosing type 2 diabetes mellitus using a multiclass prediction model with a minimum number of input variables [29]. Data from 149 patients from Taiwan were analyzed. Decision trees, deep neural networks, and ensemble algorithms were used to build predictive models. The effectiveness of the diagnosis was assessed using several parameters. The best results were achieved using an SVM model based on 10 input parameters.

A review of modern AI solutions that contribute to the fight against diabetes is presented in several works [38, 39].

IV. POTENTIAL PROBLEMS OF AI- BASED DDSS

In the examples presented in the previous section, we have illustrated the numerous possibilities associated with the use of AI systems in DDSS. However, some problematic aspects should also be noted that hinder the development of such solutions. Further, the main problems that are discussed in the literature on this topic will be indicated. Some of them are solely related to the specific nature of AI methods, while others are more general.

A. The Limited Accuracy of Forecasts

The most important criterion for introducing AI technologies into medical practice is improving the quality of patient care. Unfortunately, even high-budget commercial solutions do not always meet this criterion. IBM's Watson for Oncology is a good example. Designed to support clinicians in cancer diagnosis, this AI-powered DDSS was released in 2013 and has been implemented in hospitals in several countries. However, in contrast to the estimates of the developer company, the actual diagnostic accuracy was not very high. As a result, the initial atmosphere of triumph was replaced by disappointment. For example, as a result of the discovered problems, MD Anderson Cancer Center, one of the leading oncological clinics in the United States, abandoned attempts to implement Watson for Oncology in 2017, having spent 62 million USD on this project [40].

An analysis of the problems encountered with Watson for Oncology demonstrated the need for systematic clinical studies of DDSS expert systems, similar to those conducted for medications and medical devices. Such studies, based on the principles of evidence-based medicine, will determine how effective and safe decisions made with the support of DDSS are [41].

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B. The "Black Box" Effect

Most AI algorithms operate in a "black box" mode, which prevents the end-user from intuitively interpreting the recommendations received. This is the essential difference between AI-based DDSS and expert systems based on simple and intuitive rules. Given that the end-user has to make very responsible decisions related to the health and safety of the patient, this circumstance should be considered a rather serious problem.

C. Critical Dependence of AI Solutions on the Quality of the Training Sample

A feature of any predictive model based on the use of AI is the presence of a training sample. At the same time, the quality of the training sample critically affects the accuracy of the recommendations issued by DDSS. Important criteria for the quality of training samples discussed in the literature are their sufficient size, data reliability, absence of bias in data collection, a unified data presentation format, absence of significant imbalances between positive and negative samples (for classification models), etc. Unfortunately, DDSS developers do not always pay due attention to these aspects.

D. The Need for Thorough Testing of DDSS

When creating a DDSS, one needs to pay special attention to thoroughly testing it. To properly validate the clinical accuracy of AI algorithms, test data must meet several criteria: (1) no bias in the set of test data to be based on target patient populations in well-defined clinical scenarios; (2) the test sample must contain clinical data from hospitals other than the institution from which the data for AI training was collected; (3) test data must be collected from several different institutions; (4) prospectively collected data should be used whenever possible. It should be noted that most of the DDSS developed so far have not been validated in this way, which raises serious concerns about their effective and safe use.

E. Impossibility or Difficulties in Improving the Model

The indisputable advantage of DDSS is the ability to efficiently replenish the training sample to retrain and improve the accuracy of the diagnostic model. Unfortunately, not all AI algorithms used in DDSS provide this capability.

F. Limited Data Entry Capabilities

In particular, we are talking about the impossibility of using speech data input in most systems described in the literature. In some cases, such an option can provide additional opportunities to increase the usability of DDSS.

G. The Effect of Fatigue from Warnings

Some DDSS, as a result of their calculations, can produce many different warnings or recommendations, including unimportant ones. This effect often leads to the fact that the user of the system begins to reject them, regardless of their importance [42]. A potential solution to this problem is to identify critical warnings for the specific use of DDSS that will have the greatest impact on the patient.

H. A Negative, Relaxing Effect on Users

Until the advent of DDSS, the diagnosis of diseases was exclusively the area of responsibility of the medical professional. Such professionals constantly improved their knowledge and skills, which led to an increase in the accuracy of diagnostic solutions. However, when introducing computer diagnostic systems, users may get the impression that checking the accuracy of machine diagnostics is not required (unfortunately, this is far from the case). As a result, users increasingly trust the accuracy of computer solutions, which in the long term can lead to the loss of their diagnostic skills or the inability to assess the consequences of computer predictions [43]. This effect can be compared to the habit of using a calculator for mathematical calculations, which gradually leads to a deterioration in mental calculation skills.

J. High Cost

Clinical diagnostic systems, including those based on AI methods, are usually very expensive, which still significantly slows down their introduction into clinical practice. Despite the certain advantages that they provide to health systems, in many cases, specific studies do not confirm their cost-effectiveness in comparison with traditional diagnostic methods (see, for example, [44]).

V. CONCLUSION

An obvious trend in the modern stage of development of DDSS is the increasingly widespread use of AI algorithms.

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At the same time, along with traditional algorithms, modern approaches related to deep learning and cognitive computing are increasingly being used. Modern systems are still not devoid of certain drawbacks that hinder their active implementation in practical health care. However, a growing number of successful examples of the development of effective DDSS in various therapeutic areas demonstrate their enormous and continuously growing potential.

In this regard, the following question seems to be extremely relevant: in which branches of medicine will AI be most in-demand in the coming years? A few years ago, it was widely believed that AI could soon replace medical professionals in the departments of radiology or pathology, who are engaged in decoding visual diagnostic data. The prevailing understanding today is that AI-based diagnostic tools, including DDSS in radiology and pathology, are unlikely to replace humans in the coming years. Rather, they will play the role of valuable virtual assistants providing more accurate and efficient patient care.

At the same time, in the treatment of common chronic diseases or primary care, AI can be effectively used as a virtual assistant for patients and the population [45]. For example, such systems could provide effective support for patients who decide to see a doctor for their minor health problems, or patients who need a repeated prescription for daily medication. In these cases, AI systems can provide effective and accurate recommendations that can prevent hospital visits. A similar scenario is possible when a patient evaluates the need for an emergency department visit. For example, an AI system would be very useful, since it can make recommendations about whether the patient should immediately go to the emergency room or visit the doctor's office on the next day using photos of skin taken on a smartphone by a patient who has a skin rash and fever in the evening or at night. In this case, the number of patients visiting the emergency department at night may decrease. In these cases, AI algorithms provide recommendations directly to patients and allow them to independently make informed decisions about the necessary actions. It should be noted that the work on the creation of such AI algorithms is still lagging behind the development of professional DDSS [45]. However, there is every reason to believe that AI tools for direct patient consultation could become one of the main directions in the development of AI systems in medicine.

In this case, professional physicians will be responsible for the clinical validation of such instruments, as well as providing reliable information about them.

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