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Own Experience in the Use of Artificial Intelligence Technologies in the Diagnosis of Esophageal Achalasia

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Aim: to demonstrate an artificial intelligence model that optimises the differential diagnosis of achalasia.
Material and methods. The study included 75 patients: 52 % men (mean age 44.5 ± 17.8 years) and 48 % women (mean age 45.6 ± 16.6 years,) with a preliminary diagnosis of achalasia.
Patients were divided into four groups: type I, II, III achalasia and a group of patients whose results did not correspond to a diagnosis of achalasia according to HRM performed based on Chicago Classification version 4.0. On the basis of a set of data from 750 swallows and therefore 6750 manometric parameters, the artificial intelligence models DecisionTreeClassifier, RandomForestClassifier and CatBoostClassifier have been trained to provide a manometric diagnosis. The comparison criteria were the training time and the f1_score metric. The technical characteristics of the model (hyperparameters) were selected using the GridSearchCV method. The model with the best results was integrated into a web application.
Results. The RandomForestClassifier was chosen as the best performing model to compare. Its technical characteristics were "decision trees" and branching depth the number of which was 14 and 5 respectively. With a maximum possible value of 1.0, these hyperparameters achieved f1_score=0.91 in 27 seconds. The web application, developed on the basis of this model, is capable of analyzing manometric data and establishing one of three types of achalasia in patients. Alternatively, it can exclude the diagnosis of achalasia. The output of an image corresponding to the diagnosis is produced for each manometric type of the disease.
Conclusions. For the first time in Russia, a machine learning model based on high-resolution esophageal manometry data was developed at the V. Kh. Vasilenko Clinic of Internal Disease Propedeutics, Gastroenterology, and Hepatology of Sechenov University. The model has been applied to the creation of a web application which has the ability to substantiate the manometry diagnosis of patients. The Federal Service for Intellectual Property (Rospatent) issued a certificate of state registration of the computer program No. 2024665795 dated July 5, 2024. This artificial intelligence programme can be used in clinical practice as a medical decision support tool to optimize the process of differential diagnosis of achalasia and early detection of the disease, to determine the patient's prognosis and to select the method of further treatment.
Keywords: machine learning, artificial intelligence, achalasia, high-resolution esophageal manometry, functional diagnostics
Conflict of interest: the authors declare no conflict of interest.

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Собственный опыт применения технологий искусственного интеллекта в диагностике ахалазии кардии

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Цель: оценить значение, роль и диагностические возможности искусственного интеллекта при диагностике заболеваний пищевода, продемонстрировать модель машинного обучения, обеспечивающую оптимизацию дифференциальной диагностики ахалазии кардии.
Материалы и методы. В исследование были включены 75 пациентов (52 % мужчин и 48 % женщин, средний возраст которых составил 44,5 ± 17,8 и 45,6 ± 16,6 года соответственно) с предварительным диагнозом ахалазия кардии (АК). При проведении манометрии пищевода высокого разрешения были оценены давление покоя нижнего пищеводного сфинктера (НПС), суммарное давление расслабления НПС, давление покоя верхнего пищеводного сфинктера (ВПС), остаточное давление ВПС, латентный период дистального

сегмента, интегральная сократимость дистального сегмента, длина разрыва сокращения, одномоментное повышение давления в пищеводе, наличие перистальтических сокращений, в соответствии с которыми пациенты были распределены на 4 группы: АК I типа, АК II типа, АК III типа и группа обследованных с диагнозом, не соответствующим ахалазии кардии. На совокупности данных 750 глотков и, соответственно, 6750 манометрических параметров модели искусственного интеллекта DecisionTreeClassifier, RandomForestClassifier и CatBoostClassifier обучались устанавливать манометрический диагноз по основным манометрическим показателям. Критериями сравнения выступили время обучения и метрика f1_score. Технические характеристики модели (гиперпараметры) подбирались методом GridSearchCV. Модель с наилучшими результатами была интегрирована в веб-приложение.

Результаты. При сравнении по лучшим показателям была выбрана модель RandomForestClassifier. Ее техническими характеристиками служили «решающие деревья» и глубина ветвления, число которых составило 14 и 5 соответственно. За 27 секунд данные гиперпараметры позволили достигнуть f1_score = 0,91 при максимально возможном значении 1,0. Разработанное на основе этой модели веб-приложение при анализе данных манометрического исследования устанавливает у пациентов один из трех типов АК или исключает диагноз ахалазии кардии. Каждый манометрический тип заболевания сопровождается выводом изображения, соответствующего поставленному диагнозу.

Выводы. Впервые в России в Клинике пропедевтики внутренних болезней, гастроэнтерологии и гепатологии им. В.Х. Василенко Сеченовского Университета на основании данных манометрии пищевода высокого разрешения была разработана модель машинного обучения, примененная для создания веб-приложения и способная обосновать манометрический диагноз пациента по введенным показателям. В Федеральной службе по интеллектуальной собственности (Роспатент) получено свидетельство о государственной регистрации программы для ЭВМ № 2024665795 от 05.07.2024 г. Эта программа искусственного интеллекта может быть применена в клинической практике в качестве инструмента, обеспечивающего поддержку принятия врачебного решения с целью оптимизации процесса дифференциальной диагностики ахалазии кардии и более раннего выявления заболевания, определения прогноза пациента, а также выбора метода его дальнейшего лечения.

Ключевые слова: машинное обучение, искусственный интеллект, ахалазия кардии, манометрия пищевода высокого разрешения, функциональная диагностика

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The application of artificial intelligence (AI) technologies in therapeutic and gastroenterological practice is becoming increasingly relevant with the potential to significantly enhance the quality of healthcare.

The development of digital technologies has made it possible to accumulate a large amount of medical data in both graphic and text formats (results of instrumental and laboratory tests, medical histories, and the evaluation of the effectiveness of therapy in different groups of patients). The sheer volume of data now available requires the development of new analytical techniques. Artificial intelligence will be of significant benefit in this area. With the use of modern technologies, the speed and quality of diagnostics can be increased, as well as the ability to predict the effect of prescribed therapy in each clinical case. Furthermore, the risk factors associated with disease can be identified, allowing for the prevention of complications at an earlier stage.

Machine learning

Machine learning is a key component of artificial intelligence (AI), a method of analyzing data based on the principle of computers' ability to learn and adapt through the experience they receive [1]. In this industry there is a strong focus on the use of automated procedures. In other words, the objective of machine learning is to develop algorithms that can learn autonomously, without the need for human intervention. Machine learning can be considered a form of "programming by example" [2]. There are three main types of machine learning: supervised, unsupervised and reinforcement learning [3].

In supervised learning the "teacher" provides the program with raw data/tasks, called a "dataset", which contains the correct answers in advance. This allows the program to develop its expertise. Upon the arrival of a new task the program initiates an independent search within the database for the most appropriate response. Prior to conducting any training, it is essential to assemble a comprehensive training dataset. This dataset must be manually reviewed and labelled to ensure accuracy and quality. This should be conducted by a suitably qualified individual or a team of personnel, in the role of the 'teacher' [4].

Supervised learning is most effective when used in models designed to solve the classification problem, as it is well-suited to processing extensive amounts of data and distributing it into various categories [5]. Examples of this include the ability to recognize objects in images, such as faces, micro-specimens and endoscopic examinations. In addition, the method is suitable for text classification, allowing to highlight useful information, separating reviews into positive and negative, and tabulating values used to make diagnoses according to certain criteria. Supervised learning is also a valuable tool for making predictions based on available data. This may include selecting the most appropriate treatment plan for the patient and identifying potential complications associated with the disease.

Machine learning in gastroenterology

The use of artificial intelligence is now a standard feature in modern gastroenterology [6]. Machine learning is used in the analysis of acquired images during esophagogastroduodenoscopy and colonoscopy to detect malignant neoplasms, inflammatory processes or bleeding of the gastrointestinal tract [7] and for screening of genetic markers that determine the features of development, course and personalized approach to treatment of non-alcoholic fatty liver disease [8].

In addition, techniques that facilitate the diagnosis of functional disorders of the esophagus are receiving increased attention. The most significant application of machine learning technology to date has been in the interpretation of high-resolution manometry (HRM) and pH-Impedance monitoring [9–16].

HRM is the recognised “gold standard” for the diagnosis of esophageal motility disorders, particularly achalasia [17–24]. Achalasia is an idiopathic neuromuscular disease that manifests as functional impairment of cardia patency due to a lack of coordination between swallowing, reflex opening of the lower esophageal sphincter (LES), and peristaltic and tonic activity of the esophageal smooth muscle [25]. This disease is one of the rare esophageal motility disorders, with a prevalence of 10 cases per 100,000 population and an incidence of 1 per 100,000 population [25, 26]. Due to the low prevalence and lack of awareness of the disease among physicians, the correct diagnosis is often made too late. According to the literature, on average, this occurs 5 years after the manifestation of symptoms. Most often, these patients are mistakenly treated for gastroesophageal reflux disease. Therefore, patients presenting with complaints of dysphagia should undergo a thorough evaluation with HRM especially when diagnoses of pseudoachalasia, esophageal strictures have been excluded by barium sulphate radiological examination and esophagogastroduodenoscopy, but diagnosis of achalasia was not confirmed [24, 27]. The growing need to accelerate and improve the quality of manometric study interpretation has become a prerequisite for the development of an artificial intelligence-based model to optimize the differential diagnosis of achalasia.

Materials and methods

The study included 75 patients: 52 % men (mean age — 44.5 ± 17.8 years) and 48 % women (mean age — 45.6 ± 16.6 years,) with a preliminary diagnosis of achalasia. In accordance with the study protocol, HRM was performed on all patients using a 22-channel water perfusion catheter and “GI Solar” manometric system (Netherlands) [18].

The following parameters were identified as the foundation for the training database: resting pressure of the lower esophageal sphincter (LES), integrated relaxation pressure of LES, resting pressure of the upper esophageal sphincter (UES), residual pressure of UES, distal latency, distal contractile integral, panesophageal pressurization, presence of normal peristalsis. A total of 750 swallows and a combined total of 6,750 manometric parameters were analyzed by two independent experts (Fig. 1).

Based on the data obtained, patients were diagnosed with achalasia types I, II, and III (Fig. 2), and the results that did not correspond to the diagnosis of achalasia were placed in a separate group. As a result of interpreting the research results, among men, type I achalasia was verified in 25 % of cases, type II — in 25 %, type III — in 14 %, and the group with an unconfirmed diagnosis consisted of 36 % of patients. Among women, the following distribution by diagnoses was observed: type I achalasia — 15.4 %, type II — 15.4 %, type III — 25.6 %, not corresponding to the diagnosis — 43.6 %.

All manometric study data was uploaded to Google Colab (Colaboratory) and preprocessed using the Python 3.10.12 programming language in accordance with the Chicago Classification version 4.0. The data were divided into a training and a test sample in a ratio of 70 % to 30 %. A training sample of 70 % was used to teach the model. The parameters of manometric study were used as teaching material, with the manometric diagnosis itself becoming the desired variable. Once the training phase of the model had been completed, a test was carried out on the remaining 30 % of the data set, representing the test sample. This phase enabled us to assess the accuracy and generalising capabilities of the model on new data. A comparison was conducted between the DecisionTreeClassifier, RandomForestClassifier, and CatBoostClassifier models from the sklearn

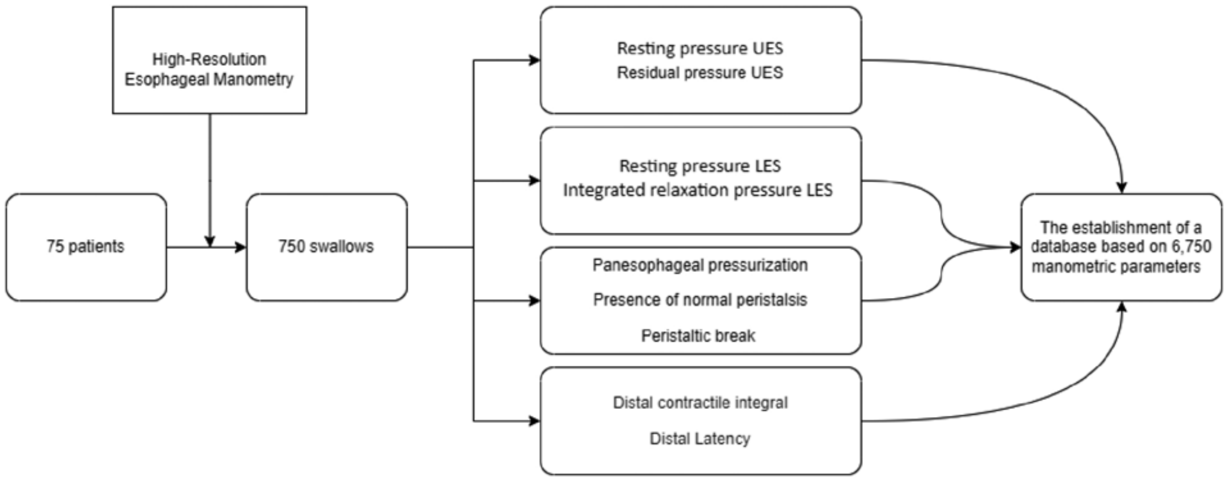

Figure 1. Research scheme: formation of a training database

Рисунок 1. Схема исследования: формирование обучающей базы данных

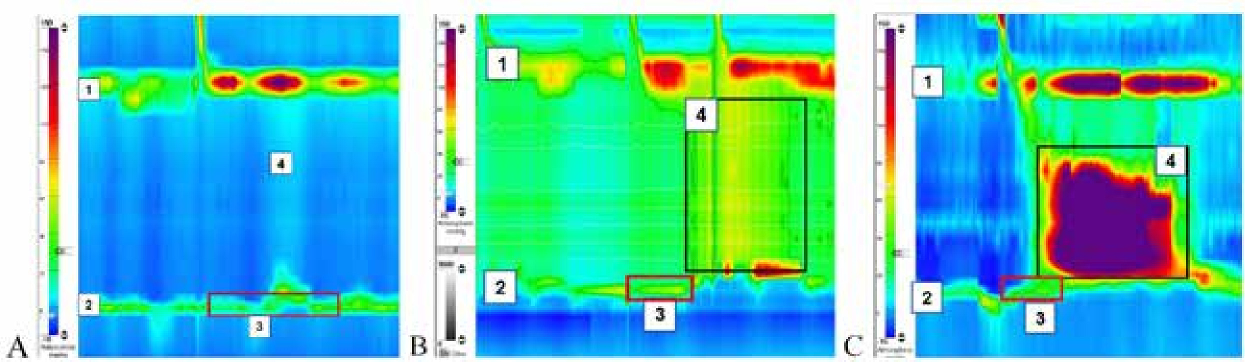

Figure 2. High-resolution esophageal manometry in patients with achalasia. A — Type I achalasia: 1 — resting pressure of the upper esophageal sphincter (UES), 2 — resting pressure of the lower esophageal sphincter (LES), 3 — absence of LES relaxation (integrated relaxation pressure (IRP) of LES is 28 mmHg), 4 — failed peristalsis; no swallows with panesophageal pressurization. Б — Type II achalasia: 1 — resting pressure of UES, 2 — resting pressure of LES, 3 — absence of LES relaxation (IRP of LES is 48 mmHg), 4 — panesophageal pressurization; failed peristalsis. B — Type III achalasia: 1 — resting pressure of UES, 2 — resting pressure of LES, 3 — absence of LES relaxation (IRP of LES is 35 mmHg), 4 — at the level of 1–14 cm above the LES, premature contraction is recorded with Distal Contractile Integral 5250 mmHg × s × cm; failed peristalsis

Рисунок 2. Манометрия пищевода высокого разрешения у пациентов с ахалазией кардии. A — I тип ахалазии кардии: 1 — давление покоя верхнего пищеводного сфинктера (ВПС), 2 — давление покоя нижнего пищеводного сфинктера (НПС), 3 — отсутствие расслабления НПС (суммарное давление расслабления НПС — 28 мм рт. ст.), 4 — отсутствие перистальтики; нет глотков с одномоментным повышением давления в пищеводе. Б — II тип ахалазии кардии: 1 — давление покоя ВПС, 2 — давление покоя НПС, 3 — отсутствие расслабления НПС (суммарное давление расслабления НПС — 48 мм рт. ст.), 4 — одномоментное повышение давления в пищеводе; отсутствие перистальтики. В — III тип ахалазии кардии: 1 — давление покоя ВПС, 2 — давление покоя НПС, 3 — отсутствие расслабления НПС (суммарное давление расслабления НПС — 35 мм рт. ст.), 4 — на уровне 1–14 см над краем НПС регистрируется преждевременное сокращение с интегральной сократимостью дистального сегмента 5250 мм рт. ст. × с × см; отсутствие перистальтики

Table. Comparison of machine learning models by performance
Таблица. Сравнение моделей машинного обучения по эффективности работы

Model / **Модель**	**F1_score**	***Time, s*** / **Время, с**
DecisionTreeClassifier	0.88	10
RandomForestClassifier	0.91	27
CatBoostClassifier	0.9	120

and catboost libraries, respectively. The selection criteria for the model were based on the training time required for the computer to learn the data and adjust its parameters to achieve the desired result. The *f1_score* metric was used to assess the quality of the classification performed, taking into account the accuracy and completeness of the analysis on the test sample. The closer the value of *f1_score* is to 1.0, the more balanced the model is (Table).

The quality of the model is primarily determined by its technical parameters, which are known as hyperparameters. In some cases, these can include the number of “decision trees” and their branching depth, as well as the number of repetitions of training cycles with a percentage of corrections to previously made decisions. The GridSearchCV from the sklearn library was employed to select the optimal parameters to achieve the highest quality level. The most successful model was therefore selected and transferred to Visual Studio Code, where it was developed into a web application using the Flask.

Results

The RandomForestClassifier was chosen as the best performing model to compare. Its technical characteristics were “decision trees” and branching depth. Using the GridSearchCV method, the optimal hyperparameters were identified as follows: number of trees — 14, branching depth of each tree — 5, which achieved the metric f1_score of 0.91. The web application, developed using the Flask function, provides a user-friendly interface for interacting with the trained RandomForestClassifier model. When the web page is opened, the boxes for entering the main parameters of the manometric examination performed on the patient are displayed. The entered data are then automatically preprocessed and transferred to the trained model, which forms a presumptive manometric conclusion based on this information. The conclusions that can be reached are type I achalasia, type II achalasia, type III achalasia and diagnosis that does not correspond to esophageal achalasia. The output of an image corresponding to the diagnosis is produced for each manometric type of the disease.

Discussion

Our scientific work has led to the development of a tool that enables faster and more accurate medical decision-making when detecting achalasia in patients, as well as optimizing the process of differential diagnosis of achalasia with other esophageal diseases. This area of research has been ongoing since 2018, when A. Frigo et al. [13] developed a medical decision support system that uses data on normal peristalsis and esophageal motility disorders to achieve an accuracy of 86 %. In 2020, a model based on the DecisionTreeClassifier algorithm was developed, achieving an accuracy level of 78 % in the differentiation of type III achalasia from types I and II achalasia [16]. In 2021, a group of scientists from the United States developed a deep learning-based model to detect pressurization and swallow type, achieving an accuracy of 0.87 and 0.64, respectively [12]. The discovery attracted significant interest, leading to the continuation of work in this area. A year later, a team of the same authors created three neural networks, which achieved an accuracy of 0.92 in manometric diagnosis [10]. Another study developed a model that can perform real-time analysis of esophageal peristalsis with 91.3 % accuracy. However, this model was not capable of providing a comprehensive manometric diagnosis [14]. In 2022, a research team from Romania developed an algorithm that can distinguish between 10 different diagnostic categories with 93 % accuracy [9]. A significant number of foreign works on this subject demonstrate the potential for further investigation in this area. A search of the eLIBRARY, Pubmed, Scholar.google databases did not yield any papers on similar studies carried out in Russia.

Our development is a semi-automated program designed to provide support for medical decision-making. The algorithm has achieved an f1_score of 0.91, which not only demonstrates its effectiveness but also outperforms the results of its foreign counterparts. Following the successful implementation of the medical decision-support system, further modifications are planned in order to refine existing limitations. The small sample size (75 patients, 750 swallows 6,750 parameters) will be increased to allow for greater statistical reliability. Additionally, the system will be expanded to accommodate four diagnoses out of seven possible, as defined by the Chicago classification version 4.0. Currently, 2,950 studies using HRM have been carried out in the laboratory for the investigation of gastrointestinal tract motility and pH-Impedance Monitoring at the V.Kh. Vasilenko Clinic of Propaedeutic of Internal Diseases, Gastroenterology, and Hepatology of Sechenov University. This could pave the way for a significant scaling up of the study by a factor of 39.

Conclusion

For the first time in Russia, the V.Kh. Vasilenko Clinic of Propaedeutic of Internal Diseases, Gastroenterology, and Hepatology of Sechenov University has conducted a study on the application of machine learning algorithms in the diagnosis of esophageal motility disorders. We have developed a system to support medical decision-making to optimize the process of differential diagnostics of achalasia and determine the patient's prognosis based on artificial intelligence, which can be integrated into clinical practice, which will speed up the work of functional diagnostics doctors in determining the types of achalasia, and with further development of the program, other motor disorders of the esophagus. The Federal Service for Intellectual Property (Rospatent) has issued a certificate of state registration of the computer program No. 2024665795 dated July 05, 2024.

The implementation of this development will enable physicians to diagnose achalasia at an earlier stage, thereby reducing the incidence of severe, complicated forms of the disease and the resulting disability of patients. Furthermore, the machine learning system is able to identify the phenotype of achalasia, which in turn determines the most appropriate course of further treatment for the patient. This will facilitate the provision of qualified medical care in a more expedient manner. The decline in the quality of life will be mitigated, and the strain on the federal funds of compulsory medical insurance will be alleviated in the future.

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