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DIAGNOSIS AND TREATMENT OF GENETIC DISEASES IN CHILDREN: NEW COMPUTER TECHNOLOGIES AND ARTIFICIAL INTELLIGENCE

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Abstract
This article discusses modern methods of diagnosis and treatment of genetic diseases in children using the latest computer technologies and artificial intelligence systems. Special attention is paid to the role of digital medicine, genomic analysis and bioinformatics in improving diagnostic accuracy, predicting the course of the disease and developing personalized approaches to therapy. The prospects for further implementation of artificial intelligence in pediatric genetics are noted, which opens up new opportunities for improving the quality of life of patients.

Keywords: Genetic diseases, children, diagnostics, artificial intelligence, bioinformatics, personalized therapy, digital medicine.

Introduction

Genetic diseases are one of the most important problems of modern medicine. According to the World Health Organization, more than 7% of newborns worldwide have congenital or hereditary pathologies of varying severity. Most of these diseases manifest themselves in childhood and often lead to disability or death.

In recent years, the development of molecular biology, genomics, and computer technology has opened up new opportunities for understanding the mechanisms of hereditary disorders and their correction. Modern diagnostic methods make it possible to identify genetic defects at an early stage of development, and the use of artificial intelligence significantly speeds up and simplifies the interpretation of complex biological data.

The purpose of this work is to analyze modern approaches to the diagnosis and treatment of genetic diseases in children, with an emphasis on the use of computer technology and artificial intelligence.

1. Epidemiology and significance of genetic diseases in children

Genetic diseases occur on average in one in twenty newborns. At the same time, about 30% of child mortality is associated with hereditary pathologies. The most common are chromosomal abnormalities (for example, Down syndrome), monogenic diseases (cystic fibrosis, phenylketonuria, spinal muscular atrophy) and multifactorial disorders caused by a combination of genetic and external factors.

Early detection of such pathologies is of great importance, since timely diagnosis and an individual approach to treatment can significantly improve the quality and life expectancy of a child.

Modern medicine focuses on predictive pediatrics, a field that combines genetic information with data on the development of a child and his environment to predict possible diseases.

2. Modern diagnostic methods

The diagnosis of genetic diseases has gone from simple cytogenetic tests to complex molecular and bioinformatic systems.

2.1 Traditional methods

Previously, the following methods were used for diagnosis:

• Karyotyping is the detection of chromosomal abnormalities.

• PCR (polymerase chain reaction) — identification of individual mutations in specific genes.

• Molecular genetic analysis — identification of hereditary defects in the coding regions of DNA.

However, these methods are limited: they do not allow to cover the entire genome and require considerable time.

2.2 Latest technologies

With the involvement of new generation Researchers (NGS), it became possible to continuously monitor millions of DNK participants, which significantly accelerated these security issues.

Mass (WGS) and mass (WES) sequencing can detect even rare and unknown mutations.

The data obtained during sequencing is processed using bioinformatic platforms, where artificial intelligence algorithms are used to automatically search for significant mutations. Face2Gene programs and similar systems that use machine vision to analyze patients' facial features and identify rare syndromes from photographs are also being actively implemented. 3. The role of artificial intelligence in diagnostics and bioinformatics

The use of artificial intelligence (AI) in medical genetics has become one of the most significant achievements of recent years.

AI is used to automatically decode sequencing results, search for mutations, predict the course of the disease, and select the optimal treatment.

Machine learning algorithms can analyze huge amounts of data, including genomic, biochemical, and clinical parameters.

Some argue that field research with international databases such as KlinVar, OMIM, and Gnomad significantly improves diagnostic efficiency.

It is especially important to use AI in cases where the disease is complex or rare. For example, if the phenotypes are unclear, the AI can offer the doctor a probable diagnosis based on thousands of similar cases from the database.

This significantly improves the accuracy of diagnosis and reduces the time between the onset of symptoms and the start of treatment.

In addition, neural network and deep learning technologies are used to analyze images such as photographs of faces, X-rays, or MRI results.
Programs like Face2Gene and DeepGestalt can help recognize a rare genetic syndrome with up to 90% accuracy, making them invisible in psychiatric practice.

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4. Modern approaches to treatment

Modern medicine is gradually moving from symptomatic treatment to personalized therapy based on the individual genetic characteristics of the patient.
The main areas of treatment for genetic diseases include gene therapy, genome editing, cellular technologies, and digital treatment control.

4.1 Gene therapy

Gene therapy is aimed at correcting a defective gene or replacing it with a normal copy.
Examples of successful applications include the treatment of spinal muscular atrophy using the drug Zolgensma, which restores the lost SMN1 gene.
Such technologies make it possible to eliminate the very cause of the disease, not just its manifestations.

4.2 Genome editing

CRISPR/Cas9 technology makes it possible to simply "cut out" specific information and replace it with others.

This method is being actively studied in the treatment of diseases such as sickle cell anemia, beta-thalassemia, and cystic fibrosis.

Despite the fact that these methods are still limited to laboratory studies, they have great potential for future clinical applications.

4.3 Cell therapy

The use of stem cells makes it possible to repair damaged tissues and organs.

For example, in some hereditary diseases of the blood or immune system, bone marrow transplantation can lead to a complete recovery of the child.

4.4 The role of AI in the selection of therapy

AI helps predict the effectiveness of drugs for a particular patient.

It analyzes data on metabolism, the genetic profile and the characteristics of the disease in order to select the optimal drugs and dosages.

This reduces the risk of side effects and increases the effectiveness of treatment.

5. Digital technologies and telemedicine

The development of telemedicine and digital platforms has greatly simplified the monitoring of children with genetic diseases.

Parents can provide doctors with information about the child's well-being through special applications, and doctors evaluate the results of treatment in real time.

Digital medical records allow you to store your entire medical history, test results, and genetic tests in one place.

This facilitates interaction between specialists and speeds up decision-making.

Cloud databases, which store anonymized genetic data of millions of patients, also play an important role.

This creates the basis for international cooperation and scientific discoveries in the field of child genetics.

6. Ethical and legal aspects

With the development of digital technologies, there is a need for strict compliance with ethical standards. Genetic information is personal data, so it must be protected from unauthorized access.

In addition, it is necessary to regulate the use of artificial intelligence in medicine in order to avoid diagnostic errors and discrimination of patients.

All research involving children should be conducted only with parental consent and under the supervision of ethical committees.

The issue of equitable access to new technologies is also being discussed: it is important that advanced diagnostic and therapeutic methods are available not only in developed countries, but also in regions with limited resources.

7. Development prospects

The integration of AI with medical genomics is expected to be actively developed in the coming years.

There will be smart medical decision support systems that will be able not only to analyze data, but also to make clinical recommendations.

In addition, the role of non—invasive diagnostic methods will increase, for example, fetal DNA analysis based on maternal blood.

The creation of international genetic biobanks will allow scientists to share data and develop new treatments.

Nanomedicine is also actively developing, aimed at delivering drugs at the cellular level with an accuracy of one molecule.

8. Conclusions

The use of computer technology and artificial intelligence in the diagnosis and treatment of genetic diseases in children is one of the most promising areas of modern medicine.

These technologies make it possible to identify hereditary diseases at an early stage, pinpoint their causes and select personalized treatment.

The introduction of AI and digital medicine in pediatrics contributes not only to improving diagnostic efficiency, but also to improving the quality of life of patients and their families.

The future of pediatric medicine lies in combining technology, genomics, and human experience.

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