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Future Trends in Genetic Research and Their Implications for Public Health: A Literature Review

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Abstract

The development of medical genetics, which took place in the last decade, allows specialists to apply an individual approach to treatment considering the genetic characteristics of the patient

Aims: to conduct an analysis of the literature on the development of medical genetics and the use of its directions in diagnosis and treatment, as well as to clarify the ethical aspects of the introduction of genetics tools in medicine

Methodology: During the literature review, data analysis and summarization was carried out on the topic of the study using the PubMed medical search engine. The search for literary sources was limited to keywords on the research topic and covered the period 2019-2024.

Results: As a result of studying the literature, it was established that today the use of medical genetics methods is a promising direction that opens opportunities for the introduction of personalized medicine. Areas such as sequencing and genomics make it possible to study the structural and functional features of the patient's gene and help predict the development of genetic diseases, while pharmacogenomics and bioinformatics make it possible to model the effect of drugs.

Scientific Novelty: During the literature search, it was found that the implementation of medical genetics in treatment and diagnostics creates several issues related to the protection of personal data, as well as imperfect sequencing methods can cause diagnostic errors. It is important to ensure access to medical genetics methods for all population groups.

Conclusion: today society is on the threshold of a discovery in medicine, which relates to the introduction of medical genetics tools in diagnosis and treatment. The methods make it possible to treat a specific patient considering genetic characteristics, which improves diagnosis, reduces the side effects of drugs and, in general, improves the results of treatment.

Keywords: bioinformatics, personalized medicine, ethics of genetic tests, next-generation sequencing, pharmacogenomics.

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Introduction

In recent decades, genetic research has become an important part of modern medicine. Areas of medicine that make it possible to edit the genome and carry out genetic testing open up opportunities for more effective diagnosis of diseases, as well as for the introduction of innovative methods of treatment [1]. The introduction of genetic methods in diagnosis and treatment is associated with the development of such areas as genomics, genetic engineering, bioinformatics, and molecular biology, which make it possible to study heredity and variability, intergenic interaction, and study the genetic structure of patients [2]. Sequencing is considered one of the promising areas of modern genetics. This technology makes it possible to establish the sequence of genes in the entire genome, and also provides an opportunity to study rare genetic variants, which has helped in recent years to reveal the genetic cause of many diseases [3]. Sequencing has provided opportunities for the development of a new promising direction in the medicine of genetic research - genomics. With the help of genomics methods, as well as the capabilities of artificial intelligence, the concept of "personalized medicine" is gaining great importance. In the era of "precision medicine", it becomes obvious that there are no generally accepted methods of treating diseases, especially those caused by a genetic factor [4]. Deciphering the genetic code will help predict the risk of genetic diseases and develop a program of necessary preventive measures. Although the concept of "genetic code" has long been introduced into modern science, and numerous scientific studies in the field of genetics are conducted with high frequency, there is still no widespread application of this knowledge in practice. Routine screening of the fetus includes examination for the presence of chromosomal abnormalities and congenital malformations of the fetus. Down syndrome, Edwards syndrome, Patau syndrome, neural tube defects, and placental abnormalities are being studied [5]. The study of the genetic sequence by the sequencing method and the accumulation of data on the typical sequence of the location of genes in the DNA molecule makes it possible, today, not only to diagnose the genetic nature of various diseases, but also to treat these pathologies using the CRISPR method [6]. Genome editing, as a method of molecular medicine, opened up the possibilities of biotechnology in agriculture, and today it opens up the possibilities of editing genetic diseases in humans in utero. Today, in prenatal testing, non-invasive methods of diagnosing genetic diseases of the fetus using fetal DNA in the plasma of the mother's body come to the fore [7]. The modern direction of medical genetics - epigenetics studies chromatin changes that regulate gene activity without changes in the DNA sequence [8]. Achievements in medical genetics make it possible to move to personalized medicine, in which individual treatment of the patient takes into account genetic characteristics, which can improve the effectiveness of treatment [9].The possibilities of genetic testing make it possible to detect in patients the possibility of developing diseases before the appearance of clinical symptoms, which makes it possible to prevent, as well as early treatment of such diseases as diabetes. The development of medical genetics and modern technologies have created questions about the confidentiality of genetic information, discrimination based on genetic characteristics, as well as the availability of testing and genetic treatment for all segments of the population [8]

The development of medical genetics over the past decades has contributed to the emergence of such areas as genomics, sequencing, bioinformatics, genetic engineering, molecular biology, which has significantly expanded the possibilities of diagnosis and treatment of genetic diseases, but today there is a problem of preserving the patient's personal data and ensuring wide access to innovative methods examination and treatment of all patients.

Research Problem

The application of genetic methods in medicine opens up wide possibilities in the diagnosis of genetic diseases even in the prenatal period, which gives hope for their successful treatment. Such methods of genetic medicine as bioinformatics, sequencing, genomics, epigenetics, and genetic testing allow sequencing the patient's genome, comparing the sequence of genes in a DNA molecule with a typical sequence, which became possible due to the accumulation of a database, and also opens new ways for the development of personalized medicine. According to experts, personalized medicine, which takes into account the individual genetic characteristics of patients, is the future they see. But uncontrolled use of genetic engineering methods can have unpredictable, including negative, consequences. The protection of the patient's personal data remains a problem today, the limited access of patients to innovative methods of medical genetics for diagnosis and treatment creates unequal conditions for receiving medical care. For successful implementation in the medical field, it is necessary to develop means of personal data protection and create opportunities for all patients, as well as to conduct educational work among the population regarding the possibilities of medical genetics methods in order to identify predisposition to genetic diseases.

Research Focus

The conducted research is aimed at familiarizing the audience with the achievements of such areas of medical genetics as bioinformatics, genomic sequencing, genomics, epigenetics, as well as the possibility of using medical genetics tools in diagnosis and treatment. Also, the purpose of the study was to systematize information and assess the current state of development of such a field of medicine as medical genetics.

Research Aim

1. Conduct a review of the literature on the methods of medical genetics that are used today in medical research and treatment.

2. To analyze the current level of directions of medical genetics

3. To analyze the advantages of medical genetics methods in modern medicine.

4. To analyze what problems patients and specialists may face when using genetic methods in treatment and diagnosis.

5. Outline the main prospects for the introduction of medical genetics in diagnostics and treatment in modern medicine.

Research Questions

1. What are the advantages of using medical genetics methods?

2. What areas of medical genetics are currently implemented in diagnostics and treatment?

3. What problems and challenges may arise when applying the methods of medical genetics?

Research Methodology

General Background

The development of innovative areas of medicine, such as molecular biology, genetic engineering, genomics, epigenomics, bioinformatics, opens a completely new direction in medicine, which makes it possible to intervene in the human genome and correct genetic mutations that can cause genetic diseases. Genomic sequencing makes it possible to analyze entire genomes, which makes it possible to identify genetic variants that are associated with certain diseases. CRISPR technology makes it possible to make point changes in the genome and treat diseases in the prenatal period. Genetic testing opens the possibility of early diagnosis of genetic diseases, which is important for the purpose of early treatment or prevention. It is also important to solve the problems that arise during the development of medical genetics and the introduction of methods in diagnosis and treatment, regarding the protection of personal data and the availability of methods for everyone who wants to receive help.

Sample / Participants / Group

A systematic approach was used to conduct the literature review; the search was conducted using the following keywords: bioinformatics, personalized medicine, ethics of genetic tests, next-generation sequencing, and pharmacogenomics For the review, we selected 248 publications that were limited to the years 2019-2024, 112 of which were freely available. The publications that had a combination of keywords were selected for further processing: "sequencing and ethics of genetic tests" and 'personalized medicine and bioinformatics', 'pharmacogenomics and personalized medicine' There were 94 publications. After careful analysis, we selected 54 sources for further processing that were relevant and contributed to the understanding of the use of medical genetics methods in diagnosis and treatment. The approach we used made it possible to process the publications on the research topic in full and freely available in English for the last five years on the PubMed search platform [10]. (Fig. 1)

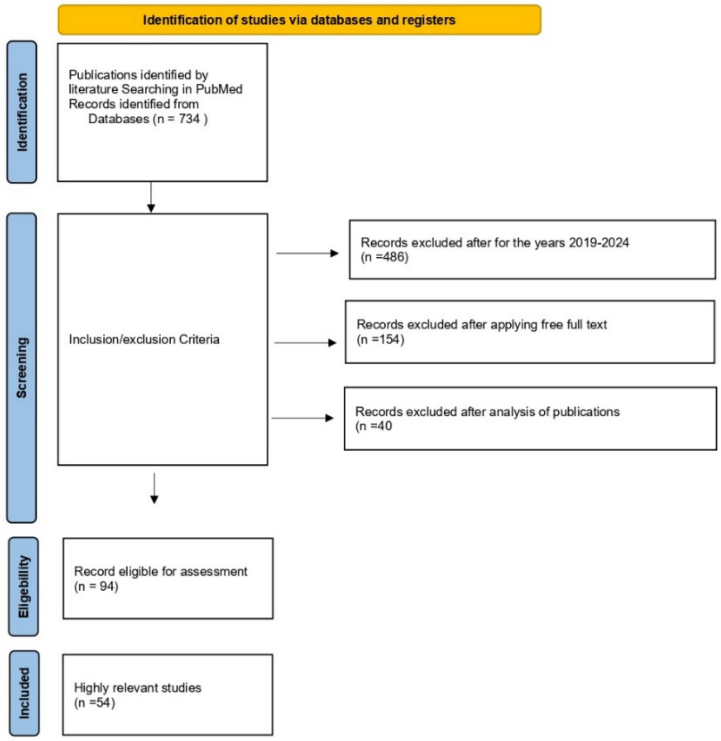

Figure 1. **PRISMA flow diagram**

Instrument and Procedures

We conducted a narrative review using a systematic methodology. When searching for sources on the PubMed platform, specific keywords were used: bioinformatics, personalized medicine, ethics of genetic tests, next-generation sequencing, pharmacogenomics. The literature search was limited to the years from 2019 to 2024 and took place in the following stages:

• The search terms "Bioinformatics" AND "Medicine", "Personalized Medicine" and "Treatment", "Personalized Medicine" and "Diagnostics", "Ethics of Genetic Tests", "Next Generation Sequencing", "Pharmacogenomics" were entered into the PubMed search. focused on the literature that examines the relationship between the methods of medical genetics, diagnosis and treatment.

• For review, we selected only articles from 2019-2024. This made it possible to consider the current state of implementation of medical genetics methods in diagnosis and treatment.

• Articles that were freely available on the platform in their full-text version were chosen for further review.

• During the work on each publication, we chose for inclusion in the article only reliable data that was confirmed.

• We carefully processed the selected articles and critically evaluated the received information, which highlighted problematic issues and determined the direction of further research. (Fig. 2).

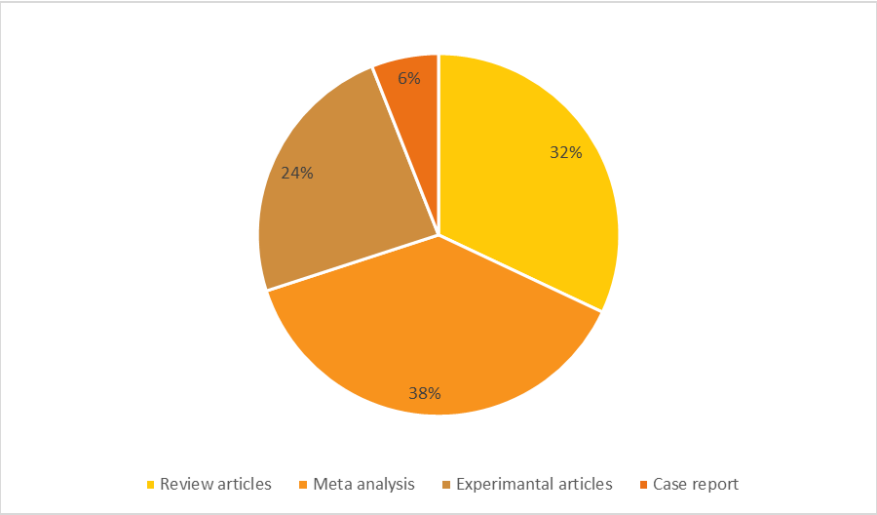

Figure 2. **Frequency distribution by types of studies**

Data Analysis

While processing the literature on the research topic, we developed theoretical knowledge about the current state of the application of medical genetics methods for diagnosis and treatment. After reviewing the literature and systematizing knowledge from scientific publications, we created a scientific narrative on the topic of the study. We described the application of medical genetics in diagnosis and treatment, identified the advantages of these methods for modern medicine, and the challenges that patients and doctors may face.

Research Results

Achievements in genetic engineering, molecular biology, which studied heredity and variability, intergenic interaction, structural and functional organization of genes opened new areas of medical genetics, such as bioinformatics, genomics, epigenetics, genome editing by CRISPR technology. Innovative methods of medical genetics make it possible to effectively diagnose genetic diseases in order to study new pathogenetic mechanisms and create individual approaches to treatment, which is the basis for the introduction of personalized medicine. We can safely say that we are at the beginning of a new era in the diagnosis and treatment of genetic diseases, which can generally improve the health of the population.

One of the methods of medical genetics, structural bioinformatics uses computer technology to create the structure of biological objects. This can be the establishment of the spatial structure of the protein and macromolecular complexes, as well as the modeling of the interaction with the ligand, which can be used to create drugs. Bioinformatics approaches in the creation of drugs are gaining importance in pharmacy today and can increase the efficiency of the drug discovery process [11]. The discovery of new medicines with the help of bioinformatics is based on the study of the type of disease and changes in the genome and epigenetic changes of a specific patient, which is an element of personalized medicine [13]. The combination of genomics, epigenetics and bioinformatics makes it possible to create personalized medicines, for example, for the treatment of oncological diseases, when the effect of the medicine will be aimed specifically at the changed cells.The possibility of creating individual drugs has become important in the search for effective drugs against COVID-19 [14]. Based on the genetic research of Sarb K. Wooller, it is known that viral infection promotes the expression of specific genes inside the cell, and bioinformatics makes it possible to create a computer model of the interaction of drugs with the genetic apparatus of the virus. The creation of drugs taking into account the genetic profile is important in the treatment of oncological diseases, especially in cases where the cells are not sensitive to the action of certain standard drug regimens [11]. Genes of tumorigenesis were determined by the method of bioinformatics, 140 such genes are known today, which is important for the creation of drugs that will have a targeted pro-oncogenic direction and will inhibit proteins that contain protein kinase domains [14]. The bioinformatics method makes it possible to differentiate between tumor suppressor genes and pro-oncogenes, which is important in order not to inhibit the function of supersor genes [15].

Using the method of molecular biology, genomics and bioinformatics, 170 mutations were found in the RNA molecule, which is present in rRNA ribosomes, the mRNA matrix, the mRNA core and tRNA [16]. Such mutations can

cause changes in the processes of regulation of gene expression, processes of transcription and protein translation. Identified changes in the nucleotide sequence of the RNA molecule make it possible to predict the appearance of certain diseases [17]. RNA modifications can be diagnosed by a sequencing method that has certain directions: sequencing as direct high-throughput sequencing, antibody enrichment sequencing, enzyme sequencing, chemical labeling sequencing, metabolic labeling sequencing, and nanopore sequencing technology [18].

Next-generation sequencing is one of the most common topics of modern research in bioinformatics and medical genetics. As a result of sequencing, it is possible to obtain millions and billions of DNA fragments. It is worth noting that such fragments are not necessarily direct copies of loci [19]. The key task of medical genetics today is deciphering these fragments. The analysis of biomolecules can be carried out using the methods of molecular marker design, as well as the method of DNA amplification. A great prospect is seen today in the combination of genomics, transcriptomics, etc. for improved decoding of biological molecules [20].

In DNA sequencing, counting the number of molecules is important. This can be done using the SQUICH program, which is the theoretical basis for counting the number of molecules. The first sequenato nanopore MinION was applied in 2014. The technique is based on the fact that a constant ion flow is created through the nanopore of the plasma membrane by creating a constant voltage [21]. Negatively charged DNA and RNA molecules pass through the nanopore from the cis side, which has a negative charge, to the trans side, which is positively charged. During the passage of the ion flow through the nanopore, certain changes may occur, which is associated with the peculiarities of the sequence of nucleotides, which are automatically deciphered with the help of special devices, which allows sequencing of DNA and RNA molecules [22]. Sequencing is carried out using automatic sequencers, where fluorescently labeled primers are used. Special fluorescent labels are connected to four nucleotides of the DNA sequence. Deciphering the sequence of nucleotides becomes possible after scanning the gel with a laser beam, which triggers the phenomenon of fluorescence [23].

The possibility of establishing the nucleotide sequence in the DNA molecule by the sequencing method opens up prospects for the use of biotechnologies in the treatment of genetic diseases. For example, the method of gene therapy - CRISPR can cut out problem areas, and also allows the identification of new targets for the use of drugs. Sequencing is a basic direction for the development of personalized medicine, for early diagnosis of genetic diseases, as well as for targeted therapy. The sequencing method expands the possibilities of reproductive medicine and can be used in pregnancy planning to assess the risk of transmission of hereditary diseases.

Nanopore sequencing technology was discovered in the 80s of the last centuries. This technology was developed on Staphylococcus aureus, the nanopore diameter ranged from 1.4 to 2.5 nm, and the motor protein that was identified in the sequencing technology was α-hemolysin [24]. This method made it possible to sequence DNA molecules, but it had limitations and complex sequences were not deciphered by this method [20]. The sequencing method was improved by including enzymes to slow down the rate of passage of the DNA molecule through the pore using phi29 DNA polymerase, which greatly improved the process of reading the sequence in the DNA molecule. Experts also note that sequencing molecules several times can improve the accuracy of information and reducing the size of the DNA molecule using ultrasound, transposase cleavage [19].

Yun Su's research examines the use of genetic sequencing in the diagnosis of neurodegenerative diseases. Pathogenetic mutations were determined using short-read sequencers and, for example, in Huntington's disease, expanded trinucleotide repeats were established, which indicate the genetic nature of the disease. Sequencing also revealed a trinucleotide repeat of CGG in the 5 UTR on the X-chromosome in X-linked mental retardation [24].

In recent years, sequencing technology has received many innovations that are used in the genetic diagnosis of patients worldwide [26]. Specialists note that repeating the polymerase reaction during sequencing of a DNA molecule can produce false results, as well as displacement of chains can also be the result of sequencing errors [27,28].

The role of genetics today is difficult to overestimate. Many studies have proven that the most common nosologies of the cardiovascular system today have a high percentage of heredity. Hereditary diseases of the cardiovascular system include: cardiomyopathy, cardiovascular insufficiency, rhythm disorders, as well as hereditary hypercholesterolemia, which is a direct factor in CHD [29]. According to some data, the analyzed genetic risks of cardiovascular diseases can significantly reduce mortality. On the part of the medical staff, it is important to be ready to timely assess the risk of cardiovascular disease in the patient and take the necessary measures.A study was

conducted to assess the awareness of doctors regarding the benefits of genetic testing and to determine the prevalence of genetic methods among medical personnel [30].

For convenience, the study was divided into three parts. The first part was to determine the use of existing methods. The second part is an analysis of factors that motivate medical personnel to use genetic methods in their practice. The third part is proposals and wishes for the improvement of genetic methods in cardiology. According to the results of the study, it was established that practically all survey participants were informed about the role of genetics in cardiology, but not all clinicians confirmed the need to use these methods in practice, because they did not belong to their field of activity. In view of this fact, there may be a suspicion that the performance of tasks by doctors only within the competence of their job instructions may lead to insufficient diagnosis [31].

Most of the survey participants recognized that the focus of attention during solving a clinical problem was focused on conducting the necessary laboratory tests. When laboratory tests became more accessible, and the time spent on them decreased, the question arose about the use of medical genetics methods in practice as preventive measures. The value of such studies can hardly be overestimated, as they can provide the necessary information about future generations, if the patient has children, and avoid the debut of the disease in them [29].

A key problem identified during the study was the lack of knowledge of genetics among medical personnel, due to which they had a need to turn to specialists when investigating a particular clinical case. There is also a tendency for doctors to ignore the need to refer to specialists in medical genetics due to the importance of focusing on the modification of existing parameters such as: elimination of symptoms of cardiovascular diseases, achievement of target values of laboratory indicators and maintenance of blood pressure at a given level, modification of the patient's lifestyle, selection of appropriate treatment methods [30]. A very common feature encountered by researchers in the analysis of clinical cases is the attitude of clinicians to diagnosis and treatment, the algorithm of which will not deviate from the protocol order. It is undeniable that the diagnosis of genetic code violations requires financial resources and is not always economically beneficial for the patient.However, a clear definition of the genetic abnormality and establishment of the cause of the defect in the phenotype, for example, a defect in enzyme systems, can help in selecting the necessary treatment [30].

The solution to the problem of insufficient competence among medical personnel in the field of genetics can be advanced training courses, as well as participation in research that proves the importance of applying genetic methods in practical cardiology. The theme of confrontation between supporters and opponents of medical genetics in cardiology practice may be that many representatives of the medical field do not have a strong desire to possess knowledge of medical genetics at an expert level, explaining this by the fact that a clinical case should be subject to a clear analysis, which will be carried out according to an established algorithm. and the methods of medical genetics can be applied only in those cases where there is a clear clinical picture of genetic nosology, while the routine conduct of genetic studies requires a large amount of time and has questionable expediency [32].

Hypertrophic cardiomyopathy caused by genetic factors can be considered a big problem in cardiology. This pathology requires strict lifestyle modification, conservative treatment in the form of drugs that affect pre- and postload on the heart, and sometimes heart transplant surgery. Clinically, cardiomyopathy is manifested by spontaneous arrhythmias and heart failure of the left ventricular type. It is believed that the genetic nature of cardiomyopathy is quite diverse. Approximately 30 percent of the disorders occur in genes encoding sarcomeres [16]. In many cases of cardiomyopathy, the etiology cannot be identified, since the sarcomere defect can be caused by a monogenic defect and can also occur as a result of genetic disorders that do not occur according to Mendel's laws [32].

For the treatment of hypertrophic cardiomyopathy, researchers consider gene editing promising. The mechanism of this method consists in the ability of endonucleases to influence the synthesis of DNA, which is capable of repairing errors in the genome. In scientific sources, it is often claimed that this method made it possible to better understand the pathophysiological processes of cardiomyopathy [30]. The methods of influencing the genetic code also include gene transfer, which consists in the fact that certain genes responsible for the synthesis of necessary proteins can be transferred to the genome, replacing defective genes. This method can at least modify the disease and is suitable for the treatment of various types of cardiomyopathies. Also worthy of attention is the method of blocking certain alleles in genes using RNA. It consists in the fact that protein coding by a defective gene stops, because RNA molecules are attached to it, which stops the synthesis of an abnormal protein [14].

During the experiment on mice, homozygous and heterozygous, it was established that the gene editing method has a certain effect. Homozygous mice were diagnosed with hypertrophic cardiomyopathy, which progressed until they were 9 months old. Homozygous mice were diagnosed with a severe form of hypertrophic cardiomyopathy, because of which the mice could live for a maximum of 1 week [33]. A double adeno-associated viral vector in combination with a T-troponin promoter to achieve specificity is key in gene therapy. According to the results of the study, it was established that a large percentage (35%) of transcription correction was achieved, which is a convincing proof of the effectiveness of the gene editing method [14].

Genome editing in cardiomyopathy can be a promising direction of treatment and diagnosis. The development of cardiomyopathies is associated with acquired and hereditary factors, which is associated with gene mutation. Establishing the genetic nature of cardiomyopathy makes it possible to use CRISPR-Cas9 technology to correct problematic areas of the DNA molecule. Currently, the possibility of prenatal or preimplantation genome correction for cardiomyopathies is being considered. At the same time, it is necessary to remember that the technique has certain limitations, which are associated with the appearance of unwanted mutations.

As for the method of gene transfer, they are aimed mainly at the MYBPC3 site, changes in which are believed to play a major role in the mechanisms of hypertrophic cardiomyopathy. The most successful step is to replace the deficient protein. This task can be achieved using the process of RNA trans-splicing, which takes over the function of producing the missing protein. However, it is noted that this method is not effective in achieving visible clinical results, which limits its use. Another method of gene replacement is to insert a copy of MYBPC3 DNA. The proof of the effectiveness of this method is the improvement of the functioning of myofilaments. The method of silencing certain alleles can also be effective in stopping the progression of hypertrophic cardiomyopathy [14]. However, they note that this effect is not long-lasting, which is why the use of allele silencing methods is not used very often. The signal path modulation method is aimed at controlling the processes of heart muscle contraction and relaxation. It is believed that one of the causes of hypertrophy of cardiomyocytes is an insufficient level of SERCA2a, therefore methods aimed at increasing the expression of this gene are potentially effective. Inhibition of phospholamban, which is an endogenous inhibitor of SERCA2a, is one of the key stages of this method [34].

Today, methods of gene therapy in cardiology, in the section of cardiomyopathy, are at the experimental stage. A large number of clinical studies are necessary for the implementation of these methods in medical practice. In addition, the researchers note that gene therapy may pose certain risks from the point of view of the body's immune response. AVV vectors, which are widely used in cardiac genomics, can become potential foreign agents for the body's immune system, which can lead to an immune response. The question of the duration of the introduction of gene preparations into the target organs is controversial. On the one hand, because of long-term administration of these drugs, an immune response may develop. On the other hand, simultaneous administration of a large dose of the drug threatens with symptoms of general intoxication, severe hepatotoxicity, as well as the development of renal failure and thrombocytopenia [14].

According to research, drugs can cause fatal consequences. Medicines can have different effects depending on gender, age, race, individual characteristics, routes of administration. Pharmacogenomics enables doctors to choose drugs depending on the individual, genetic characteristics of patients in the treatment of cardiovascular, oncological, and nervous system diseases [35]. The most adverse reactions are caused by drugs for the treatment of cardiovascular diseases. According to experts, diseases of the cardiovascular system and disorders of the central nervous system make up 80% of all diseases, and side effects of drugs are noted in 40% of patients, which are associated with genetic predisposition. The use of chemotherapy drugs such as cisplastin is associated with hearing loss in children with genetic variants of TPMT (rs12201199) and ABCC3 (rs1051640) [36].

Innovative methods of medical genetics create opportunities for the development of personalized medicine, which aims to treat the patient taking into account genetic characteristics, which can reduce the side effects of drugs and choose the most optimal treatment methods, and the use of genetic testing will make it possible to apply treatment or create preventive measures in the presence of genetic tendencies.

According to recent studies, adverse reactions to oics are related to gender and are more often observed in women receiving treatment for cardiovascular diseases. A higher frequency of adverse reactions is associated with treatment with an angiotensin-converting enzyme (ACE) inhibitor, digoxin, or mineralocorticoid receptor antagonist [37].

Gene therapy is possible only under conditions of intervention in the genome. Genome intervention is the introduction of healthy genes into the body or the correction of defective sequences. However, ethical issues remain unchanged. It is important to establish the risks of genetic therapy and to determine ethically acceptable approaches to the experiment.

Maintaining biological security is also a key task. It is necessary to weigh all the risks of carrying out genetic experiments. It is worth noting that interfering with the genome has unpredictable consequences. Ethical aspects consist in the control of research in the field of medical genetics, the determination of ethical standards, which were considered the boundary between what is allowed and what is not allowed. Certain aspects of genetic engineering require analysis from a social point of view. Specialists in genetic engineering must consider the opinion of the leading intellectual community. It is also important to know the opinion of the public regarding these technologies, to consider the influence of mass media on the establishment of views, to be able to convey to society the importance of modern technologies of genetic engineering in the prevention of the occurrence of hereditary diseases and anomalies.

Discussion

Our literature search demonstrated that the introduction of medical genetics tools into medicine in recent decades has significantly improved the diagnosis of genetic diseases and opened the way to new treatment methods. Such areas of medical genetics as bioinformatics, genomics, pharmacogenomics, genomic sequencing, epigenetics, genetic testing, and screening open opportunities for medicine, especially for the implementation of personalized medicine, which is considered by specialists as the medicine of the future. How D research examines the benefits of personalized medicine. Experts have indicated that it is necessary to conduct a full examination of healthy individuals in order to have a database that will allow establishing the reasons for the transition from a healthy state to a diseased state [38]. The accumulation of a genetic database, digitized X-ray and histological images, and artificial intelligence create opportunities for accurate diagnosis and the ability to treat a specific patient. The use of such methods as bioinformatics in diagnostics, namely proteomics, makes it possible to establish the tertiary structure of proteins and possible interaction with plasma membrane ligands through computer modeling, which is important for studying the effects of drugs and creating new methods of pharmagenomics [39]. According to experts, side effects of drugs are noted in 40% of people who receive treatment for cardiovascular and central nervous system diseases. Pharmacogenomics makes it possible to create personalized medicines, and also makes it possible to study the effect of drugs on the human genome and to study the mechanism of side effects. Bioinformatics studies therapeutic targets for drugs taking into account the patient's genetic profile [40]. Anmar Al-Taie's research examines the issue of creating drugs against COVID 19 and the expert notes that one drug does not fit all, which explains the unequal effectiveness of molnupiravir in the treatment of COVID 19 [41]. Ramdesivir also demonstrated selective effectiveness, so the genes rs2306283 and c.388A were identified in Africans, Asians and Caucasians, which are associated with a decrease in the speed of transport, which is important in the insensitivity of the virus to drugs [42]. The use of lopinavir also has certain characteristics among representatives of different races, for example, the Asian population has a different frequency of the variant 3435C > T allele than the African and Caucasian populations [41], which may explain the side effect that is associated with drug accumulation. Atazanavir can cause hyperbilirubinemia, which is associated with the homozygous state. The selection of drugs taking into account the individual characteristics of patients can reduce side effects and increase the effectiveness of drugs [43]. The improvement of sequencing technology according to Gilad D Evrony allows to detect features of the DNA molecule in the form of mutations or modifications, which can help establish a certain nucleotide sequence that has been established as a carrier of the disease [44]. In recent decades, the sequencing method has received new and improved methods, but even today there is a possibility of errors, which is associated with the displacement of molecules during translocation through nanopores, as well as the rapid translocation of the DNA molecule. Genome sequencing opens up opportunities for genome editing using the CRISPR technique. In the researches of Paul B, the mechanisms that explain the possibility of modifications in the DNA molecule are considered [45]. CRISPR technology as molecular scissors, where a simple exchange of the guide RNA sequence is sufficient to re-engineer the specificity of the nuclease, has paved the way for a revolution in the life sciences. Today, Cas9 and Cas12a are considered, which are used for genome editing [45].

In the work of Mandrioli M., the ethical issues of the introduction of gene methods in diagnosis and treatment are considered, so the author notes that the CRISPR-Cas9 method, which makes it possible to treat point mutations,

genetic diseases, etc. cannot be patented by commercial pharmaceutical companies, but should be available to anyone who needs such help [46].

The application of medical genetics in recent years has received significant progress, which is connected with technical possibilities. The use of sequencing allows the detection of genetic mutations in the early prenatal stages or even before implantation and the timely correction of detected mutations using CRISPR-Cas9 technology. Prenatal diagnosis, which was previously based on the diagnosis of amniotic fluid and is an invasive method. Today, prenatal blood of the mother can be used to detect genetic abnormalities of the fetus, which is a non-invasive method and therefore has no side effects. The sequencing method allows couples to assess the risks of the appearance of hereditary diseases and carry out a test for carrying abnormal genes. Thus, it is possible to detect the carrier of genes for such recessive diseases as cystic fibrosis and spinal muscular atrophy. The use of direct sequencing makes it possible to detect mutations in certain genes, which are the cause of the appearance of a genetic disease, and it is also possible to conduct a karyotype analysis, which will reveal the appearance of chromosomal abnormalities in the form of deletions, translocations, duplications, and others. Pharmacogenic tests make it possible to detect the reaction to the action of drugs, taking into account genetics. The methods of medical genetics make it possible to create personalized medicines taking into account the patient's genetic profile, as well as to avoid the side effects of medicines. Genetic tests allow for early diagnosis, personalized treatment, risk prediction, create conditions for better diagnosis and treatment for doctors, and for patients the opportunity to overcome the disease with the least side effects.

Conclusions and Implications

The conducted research allows us to draw conclusions about the importance of the development of medical genetics, and it can be argued that today medicine is on the verge of introducing personalized medicine, which takes into account the individual genetic characteristics of the patient. Using the method of pharmagenomics and bioinformatics, it is possible to establish the specifics of the effect of drugs on the patient's body and the possibility of side effects, taking into account the human genome and gene polymorphisms. The sequencing method makes it possible to establish the nucleotide sequence of DNA and RNA and to establish the areas of mutations and modifications, and CRISPR/Cas9 technologies make it possible to cut out problem areas even prenatally, as well as establishing a genetic predisposition to diabetes, ulcers, and bronchial asthma makes it possible to start treatment in time. Today, there are still problems related to the protection of personal data, the possibility of errors during sequencing, which can cause a new gene mutation, which indicates no prospects for research in these directions.

Limitations

The application of medical genetics methods can create certain challenges that are associated with genome correction, which can be the cause of genetic discrimination. The methods of medical genetics, which allow obtaining almost all information about the patient, currently do not have sufficient protection of personal data, which can become a problem when the information reaches criminals. Genetic tests are not perfect and can give false positives or false negatives. the application of technology for the correction of abnormal DNA regions has its own technological limitations. There are risks of unsuccessful correction, which will cause the appearance of new mutations. Pharmacogenomics and bioinformatics, which study the effect of drugs and the possibility of point correction, also have certain technological limitations, which is associated with the imperfection of methods of delivering correctors of genetic information. Not everyone can use the methods of medical genetics due to the high cost, which also creates inequality in the provision of medical services.

Suggestions for Future Research

The methods of medical genetics, which have made significant progress in recent years, need constant improvement for their safe use in the future. The sequencing method, which allows reading DNA and RNA molecules, has certain limitations, as it sometimes gives false results and needs further improvement. The development and improvement of methods of delivery of genetic editors is promising. Medical genetics is closely related to the possibilities of technological progress, so the use of innovative methods to improve the possibilities of medical genetics for diagnosis and treatment is promising. The use of artificial intelligence to analyze genetic data can improve the method of diagnosis and help the doctor decide on the right treatment. An important issue that needs to be resolved is the reduction of the cost of genetic tests and the possibility of their use among all segments

of the population, which is also a promising direction in terms of finding means to increase the production of genetic tests.

Declarations

Author Contributions

Conceptualization, Iryna Orlova and Olha Abramchuk; methodology, Iryna Orlova; software, Iryna Orlova; validation, Iryna Orlova, Olha Abramchuk and Ivanna Babik; formal analysis, Iryna Orlova; investigation, Iryna Orlova; resources, Iryna Orlova; data curation, Iryna Orlova; writing—original draft preparation, Iryna Orlova; writing—review and editing, Iryna Orlova; visualization, Iryna Orlova; supervision, Iryna Orlova; project administration, Iryna Orlova and Marta Dats-Opoka.

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Conflicts of Interest

The authors declares that there is no conflict of interests regarding the publication of this manuscript.

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