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GENODIAGNOSTICS AS THE BASIS OF PERSONALIZED MEDICINE

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АННОТАЦИЯ: Fundamental research in genetics ensures the progress of medicine. A genetic approach brings us closer to understanding the biological nature of diseases, and the data obtained through this approach form the foundation for the further development of theoretical, clinical, and preventive medicine. Today, molecular genetic methods in disease diagnostics represent a broad and diverse group of techniques aimed at identifying polymorphisms in gene structures. The significance of molecular diagnostics in medicine is evident: it allows for the early detection of diseases, treatment with drugs proven to be safe and effective based on molecular diagnostics, integration of diagnostics and therapy, as well as monitoring of treatment and determination of disease prognosis.

ANNOTATION: Фундаментальные исследования в генетике обеспечивают прогресс медицины. Генетический подход позволяет приблизиться к пониманию биологической сущности заболеваний, а получаемые при таком подходе данные создают базу для дальнейшего развития теоретической, клинической и профилактической медицины. Молекулярной генетические методы в диагностике болезней сегодня-это большая и разнообразная группа методов, предназначенных для выявления полиморфизмов в структуре гена.

Значимость молекулярной диагностики для медицины очевидна: это раннее выявление болезни и лечение препаратами, которые считаются безопасными и эффективными на основании молекулярной диагностики; интеграция молекулярной диагностики и терапии; мониторинг лечения и определение прогноза.

KEYWORDS : gene diagnostics, molecular biology, PCR, biochips.

ANNOTATSIYA: Genetikadagi fundamental tadqiqotlar tibbiyot taraqqiyotini ta’minlaydi. Genetik yondashuv kasalliklarning biologik mohiyatini anglashga yaqinlashtiradi, ushbu yondashuv asosida olingan ma’lumotlar esa nazariy, klinik va profilaktik tibbiyotning yanada rivojlanishi uchun zamin yaratadi. Bugungi kunda kasalliklarni aniqlashda qo‘llaniladigan molekulyar-genetik usullar bu gen tuzilmasidagi polimorfizmlarni aniqlashga mo‘ljallangan keng va xilma-xil usullar majmuasidir. Molekulyar diagnostikaning tibbiyot uchun ahamiyati yaqqoldir: bu kasalliklarni erta aniqlash, molekulyar diagnostika asosida xavfsiz va samarali deb topilgan dori vositalari bilan davolash, diagnostika va terapiyani integratsiyalash, shuningdek, davolash jarayonini kuzatish va kasallikning prognozini belgilash imkonini beradi.

KALIT SO'ZLAR: gen diagnostikasi, molekulyar biologiya, biochiplar.

INTRODUCTION

The progress of medicine is ensured by fundamental developments, among which genetic research is in one of the first places. Molecular diagnostics is an actively developing field of medicine based on understanding the mechanisms of disease development at the molecular level. There are a large number of different genes in the human genome, and changes in their structure or regulation of protein synthesis can lead to the appearance and development of the disease. Genetic factors are non-modifiable risk factors for diseases, and the human genotype does not change during life, and the use of molecular diagnostic methods makes it possible to detect hereditary diseases at any stage of the body's development, including before birth (prenatal diagnosis). For the first time, the diagnosis of "Hemophilia" was made even before the

birth of a child in 1989 in St. Petersburg, at the D. O. Ott Institute of Obstetrics and Gynecology. Then, prenatal diagnostics revealed such severe genetic pathologies as cystic fibrosis, Duchenne myodystrophy, phenylketonuria, and brittle X-chromosome syndrome. The genes of the most common (about 320) and relatively rare (about 170) hereditary diseases are now known. For many late-onset diseases (Huntington's chorea, myotonic dystrophy), preclinical diagnosis of predisposition is essentially predictive. In many genetic diseases, disorders in the links of metabolism have been deciphered, according to which a hereditary pathological process develops, starting from an abnormal gene product and ending with a clinical picture at the level of metabolic and intracellular processes. Studies are often conducted only to confirm an already established diagnosis. In these cases, the task of the study is to establish a correlation between the genotype and the phenotype, that is, to predict the severity of the disease based on the mutation.

The methods of molecular diagnostics make it possible to identify not only the genes of hereditary diseases, but also the genes of predisposition to a particular disease. Among the diseases caused by the presence of predisposition genes in the genome, there are late-onset diseases that can be genetically detected already at birth, but visible symptoms develop later in life.: breast cancer, Huntington's chorea, Alzheimer's disease, familial polypous colon cancer, a number of neurodegenerative diseases and multifactorial diseases, which are also detected at birth, but manifest themselves only with unfavorable external factors: diabetes mellitus, hypertension; atherosclerosis, oncological diseases. One of the tasks of genodiagnostics is to study at the molecular level the causes of infections and the mechanisms of their development. The basis of the genetic diagnosis of infectious diseases consists of molecular research methods aimed at detecting the genetic material of infectious agents: bacteria, viruses, parasitic agents, protozoa, rickettsias. The advantage of gene diagnostics is that it makes it possible to identify a tendency to a particular disease long before its clinical manifestations, take timely preventive measures to prevent its development or alleviate its course, and apply therapy taking into account individual characteristics.

Purpose: To review the current state of genodiagnostics and its fields of application, which are inaccessible using classical methods. Materials and methods: An overview of scientific sources on the use of molecular biology in medicine is presented. Results and discussion: Since the beginning of the 21st century, biological sciences and medicine have entered a new era, characterized by the use of genetic technologies necessary to understand the structural and functional organization of the human genotype in normal and pathological conditions. The principle of PCR was described in 1983 by K. Melis— an American biochemist at Cestus [3, 5, 7], for which he was awarded the Nobel Prize in Chemistry in 1993. The method is based on multiple copying (amplification) of a specific DNA fragment using the DNA polymerase enzyme according to the principle of complementarity, which is a marker for a disease or a given type of infectious agent and allows detecting several specific DNA molecules in the presence of millions of other molecules [8]. There are qualitative and quantitative variants of PCR. Qualitative PCR can be used in the following cases: diagnosis of infectious diseases caused by certain pathogens; identification of infectious agents before quantitative PCR; diagnosis of oncological diseases; diagnosis of genetic diseases; identification of personality; screening of genetically modified food sources. The creation of new technologies has allowed us to move to a qualitatively new level — quantitative PCR. Currently, the most promising method is real-time PCR (Eng. Real-Time PCR). The essence of the method is to study the accumulation of amplification products using a special device without subsequent electrophoresis. Since the kinetics of accumulation of amplification products is related to the initial amount of the matrix, this makes it possible to accurately estimate its quantity. The

distinctive features of this method are the ability to quantify DNA/RNA of infectious agents in the test material, the absence of an electrophoresis stage, less stringent requirements for the organization of a PCR laboratory, and automatic registration and interpretation of the results obtained. The absence of an electrophoresis stage minimizes the risk of contamination with PCR products and thus reduces the number of false positive results. When deducing various mutation variants and analyzing the DNA structure, decoding its sequence remains the gold standard method in the field of DNA analysis [11-13]. Promising areas of practical use of diagnostics based on the use of PCR principles are: DNA diagnostics of benign and malignant neoplasms; study of the genetic basis of diseases, the possibility of diagnosing and starting treatment of the disease before its symptoms appear; identification of personality: forensic medicine, criminalistics; organ and tissue transplantation; determination of paternity; diagnosis of pathogens in food [8].

Early comprehensive and complete diagnosis creates the prerequisites for anti-epidemic and preventive measures. When discussing the possibilities of PCR, it is necessary to consider the use of this method to overcome the difficulties encountered in the diagnosis of pathogens of infectious diseases with a characteristic drift of surface antigens. Many pathogenic bacteria, such as gonococci, meningococci, barrels, and mycoplasmas, resist the host body's immune system because they constantly change the antigenic structure of surface determinants capable of inducing a projective immune response. The molecular mechanism that ensures the constant variability of surface structures has been well studied and is called the cassette mechanism. Antigenic drift allows the microorganism to constantly evade the attack of the immune system, prolonging the infectious process, transferring it to a chronic stage. However, with the constant change of antigenic determinants, low-water antibodies to the constant parts of the antigens begin to be produced. The bacterial cell needs to make a whining decision: to give up this fight or find some other mechanism that allows it to escape the attack of the immune system. Such a mechanism does exist. Using PCR technology, it was possible to demonstrate that in persistent forms of mycoplasmas, the expression of genes encoding the synthesis of antigenic determinants is turned off, which negatively affects their reproductive potency, but at the same time makes it possible to persist, prolonging the infectious process [15]. The use of hybridization technologies opens up new prospects for the gene diagnosis of viral hepatitis.

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