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«MEDICINE»

TOPICAL ISSUES OF MEDICAL GENETICS OF THE XXI CENTURY

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ABSTRACT

The article presents topical issues of medical genetics, its methods, recent achievements and development prospects. The successes of research in the field of human medical genetics are of practical importance for all mankind. Expanding knowledge about genetic diseases helps to improve their diagnosis, find new therapeutic approaches and, moreover, prevent their occurrence. The article outlines some interesting aspects of the prospects for the development of medical genetics.

Keywords: medical genetics, heredity, DNA, gene, genome, biology, chromatin, nucleotide.

Human genetics and such fundamental disciplines as medical biology, anatomy, physiology, biochemistry form the basis of modern medicine. The place of genetics among the biological sciences and the interest in it are determined by the fact that it studies the basic properties of organisms, namely heredity and variability. Heredity and variability in humans are the subject of study of human genetics at all levels of its organization: molecular, cellular, organismal, population. Human genetics owes its success to a large extent to medical genetics, the science that studies the role of heredity in human pathology [1].

In the DNA molecule, on which the life program is written, and more specifically, the entire synthesis process, the structure and function of proteins, as the main elements of life. In addition to preserving the life program, the DNA molecule performs another important function – its self-reproduction, copying, and creates continuity between generations. Once having arisen, life reproduces itself in a huge variety, which ensures its stability, adaptability to various environmental conditions and evolution [7].

Medical geneticists can delve deeper into population and evolutionary processes, study hereditary diseases, the problem of cancer, and much more. Modern genetics has provided new opportunities for studying the activity of an organism: with the help of induced mutations, almost any physiological process can be turned off and on, protein biosynthesis in a cell can be interrupted, morphogenesis can be changed, and development can be stopped at a certain stage. The development of molecular biological approaches and methods has allowed medical geneticists not only to decipher the genomes of many organisms, but also to design living beings with desired properties [6].

Medical genetics studies the role of heredity in human pathology, the patterns of transmission from generation to generation of hereditary diseases, develops methods for diagnosing, treating and

preventing hereditary pathologies, including diseases with a hereditary predisposition. This direction synthesizes medical and genetic discoveries and achievements, directing them to fight diseases and improve people's health. Medical genetics is an important part of theoretical medicine, considering the following questions in connection with pathology: what hereditary mechanisms support the homeostasis of the body and determine the health of the individual; the importance of hereditary factors in the etiology of diseases; what is the ratio of hereditary and environmental factors in the pathogenesis of diseases; what is the role of hereditary factors in determining the clinical picture of diseases; does the hereditary constitution affect the process of a person's recovery and the outcome of the disease; how heredity determines the specifics of pharmacological and other types of treatment. As a theoretical and clinical discipline, medical genetics continues to expand rapidly in various directions: the study of the human genome, cytogenetics, molecular and biochemical genetics, immunogenetics, developmental genetics, population genetics, and clinical genetics. [eight]. population genetics, clinical genetics. [eight]. population genetics, clinical genetics. [eight].

During the implementation of the Human Genome Project, many new research methods have been developed. The same methods of analysis can be used for other purposes: in medicine, pharmacology, forensics, and so on. Every thousandth child in the world is born with some kind of hereditary defect. To date, about 15 thousand different human diseases are known, of which more than 4 thousand are hereditary. Mutations responsible for diseases such as hypertension, diabetes, certain types of blindness and deafness, and malignant tumors have been identified. The genes responsible for one of the forms of epilepsy, gigantism, etc. were discovered. The molecular basis of heredity was discovered, the genetic code was deciphered; new artificial genes are created; viruses are grown in test tubes; identical twins of frogs and sheep are created from the cells of a mature organism; human cells are fertilized in test tubes; women are transplanted with embryos; doctors treat many hereditary diseases; hybrids of rats and mice are grown [7].

Every year we get closer to preventing the development of hereditary diseases at the stage of fetal development. The main tool capable of such magic is gene editing using CRISPR technology. It is she who allows you to literally get into DNA, remove or transform the necessary genes [5].

According to the forecasts of medical geneticists, by the end of the second decade of the 21st century, genetic vaccines will replace the usual vaccinations, and doctors will have the opportunity to permanently end such incurable diseases as cancer, Alzheimer's disease, diabetes, and asthma. In this direction, scientific research is being carried out, which has its own name – gene therapy. According to some forecasts, around 2021, exceptionally healthy children will be born: already at the embryonic stage of fetal development, geneticists will be able to correct hereditary problems. Scientists predict that in 2055 there will be attempts to improve the human species. We will read everything that is written in our chromosomes, and we will learn to understand it, we will use this to correct all the errors found. By this time, we will have learned to design people of a certain specialization: mathematicians, physicists, artists, poets, and maybe even geniuses. Man's dream will come true: the aging process, of course, can be controlled, and there it is not far to immortality [4].

Conclusion

The development of medical genetics is a continuously expanding front of research. A lot has already been done in this area, and every day the cutting edge of science is approaching the goal – unraveling the nature of the gene. To date, a number of phenomena characterizing the nature of the gene have been established. A gene in a chromosome has the property of self-reproducing; it is able to mutate; it is associated with a certain chemical structure – DNA; it controls the synthesis of amino acids and their sequences into a protein molecule. [2]. In connection with recent studies, a new understanding of the gene as a functional system is being formed, and the effect of the gene on determining traits is considered in an integral system of genes – the genotype. The opening prospects for the synthesis of living matter attract great attention of geneticists, biochemists, physicists and other specialists. With the help of the latest cytological methods, cytogenetic, in particular, extensive studies of the genetic causes of various diseases are being carried out, thanks to which there is

a new branch of medicine – medical cytogenetics. Sections of genetics related to the study of the effect of mutagens on the cell are directly related to preventive medicine.

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