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HEREDITARY DISEASE

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Annotations: The interest shown by scientists around the world to human heredity is not accidental. Currently, about 2000 hereditary diseases and genetically determined syndromes are known. Their number is constantly growing, dozens of new forms of hereditary pathology are described every year. At the present stage of development of medicine, recognition of various hereditary diseases and genetically determined syndromes is of exceptional importance.The lack of a generally accepted classification of hereditary diseases, their large polymorphism and the large number of phenocopies create difficulties in the diagnosis and differential diagnosis of heredial forms of pathology.
Keywords: hereditary,genetics,syndromes,

Hereditary diseases are diseases caused by chromosomal and gene mutations. The science that studies the phenomena of heredity and variability in human populations is genetics. It is often believed that the term "hereditary disease" and "congenital disease" are synonyms. However, unlike congenital diseases that arise at the birth of a child, hereditary diseases are already caused by hereditary and exogenous factors.Hereditary diseases - diseases, the occurrence and development of which is associated with various defects and disorders in the hereditary apparatus of cells. At the heart of hereditary diseases are mutations: chromosomal, gene and mitochondrial.

With hereditary diseases, genetic disorders of a different nature and localization can occur. These diseases can be associated with abnormalities in nuclear (chromosomal) or mitochondrial DNA. They can develop as a result of gene (point) mutations (transitions, transversions , mutations of shift of the reading frame , or rather gross changes in the structure of chromosomes or DNA (deletions, duplications, inversion, translocation, transposition), as well as due to genomic mutations (changes in the number of chromosomes). Accordingly, hereditary diseases are classified as genetic, chromosomal, mitochondrial.

Hereditary diseases are also classified according to the type of inheritance. For a significant part of hereditary diseases, the type of inheritance is established - pathological signs, as well as normal, can be inherited autosomal dominant, autosomal recessive and sex-linked (X-linked dominant, X-linked recessive and Y-linked types of inheritance). The term "autosomal" indicates that the mutant gene is located in the autosome, "X-linked" - in the sex X-chromosome, and "Y-linked" - in the sex Y-chromosome. Isolation of dominant and recessive types of inheritance is essential from a medical point of view, since with the dominant type of inheritance, the clinical manifestation of the disease is found in homo- and heterozygotes, and with recessive - only in homozygotes, that is, much less often. The main

methods used to establish this or that type of inheritance are clinical and genealogical, based on the analysis of pedigrees, and more accurate segregation analysis, the object of which, as a rule, is the so-called "nuclear families" (that is, parents and children).The problems of heredity have been of interest to people for many centuries. So, for example, a disease such as hemophilia has been known for a long time. In this regard, marriages between blood relatives were prohibited. Thanks to the progress of medical genetics and the expansion of ideas about the nature of inheritance of various diseases and the influence of environmental factors on the manifestation of mutant genes, the ways of treating and preventing N. z have become much clearer. Basic principles of treatment: exclusion or restriction of products, the transformation of which in the body in the absence of the necessary enzyme leads to a pathological state; replacement therapy with a deficient enzyme or the normal end product of a perverse reaction; induction of deficient enzymes. Great importance is attached to the factor of timeliness of therapy, which should be started before the development of severe disorders in patients. Some biochemical defects can be partially compensated with age. In the future, great hopes are pinned on genetic engineering, which means targeted intervention in the structure and functioning of the genetic apparatus - removing or correcting mutant genes, replacing them with normal ones.Many scientists have put forward their hypotheses about the occurrence of hereditary pathologies. Their assumptions were not always based on scientific observation. Only in the 20th century, with the development of genetics, scientific evidence was revealed. Progress in the medical field has led to a relative increase in the proportion of genetically determined pathologies. To date, more than 3500 hereditary human diseases have been identified. About 5% of babies are born with genetic or congenital diseases.Thanks to the progress of medical genetics and the expansion of ideas about the nature of inheritance of various diseases and the influence of environmental factors on the manifestation of mutant genes, the ways of treatment have become much clearer, and most importantly, the prevention of hereditary diseases. Basic principles of treatment: exclusion or limitation of products, the transformation of which in the body in the absence of the necessary enzyme leads to a pathological state; replacement therapy with an enzyme deficient in the body or a normal end product of a distorted reaction; induction of deficient enzymes. The factor of timeliness of therapy is of great importance. Therapy should be started before the patient develops severe disorders in cases where the patient is still phenotypically normal. Some biochemical defects can be partially compensated with age or as a result of intervention. In the future, great hopes are pinned on genetic engineering, which means targeted intervention in the structure and functioning of the genetic apparatus - the removal or correction of mutant genes, replacing them with normal ones.But the most important task of medical genetics is the prevention of hereditary diseases, the timely prevention of the birth of sick children. Prevention of hereditary diseases is carried out mainly through medical genetic consultations and regional diagnostic centers.

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