

Origin and Development of Hereditary Diseases

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Abstract: The human body develops on the basis of DNA, which contains genetic information. Each person's genetic material contains millions of genes, which work inextricably linked to each other. Hereditary diseases mainly occur as a result of genetic mutations, chromosomal abnormalities, or gene malfunction. These diseases are inherited, that is, they are transmitted from parents to generations, or develop as a result of the negative impact of environmental factors on genetic material. The prevalence of hereditary diseases in the world is very diverse, and they have a serious impact on human health. While some hereditary diseases, such as Down syndrome or hemophilia, are widespread, other rare diseases are often ignored by the population. The study of the origin of hereditary diseases is one of the main directions of modern medicine and genetics. Within this topic, genetic diagnostics, gene therapy, methods of prevention and treatment are developing significantly. This article provides more detailed information about the causes, development mechanisms and their significance of hereditary diseases.

Keywords: Hereditary diseases, Genetic diseases, Mutations, Genetic inheritance, Phenotype and genotype, Monogenic diseases, Polygenic diseases, Chromosomal diseases, Gene mutation, Gene therapy, Epigenetics and inheritance, Autosomal dominant and recessive inheritance.

Origin of hereditary diseases. Hereditary diseases are caused by changes in the genetic material contained in DNA. These changes can occur at the gene or chromosome level. Mutations are passed down from generation to generation, causing diseases in offspring.

Dominant inheritance: A disease can develop even if one parent inherits a recessive gene. Examples: Huntington's disease, premature hair loss, freckles, etc.

Recessive inheritance: The disease manifests itself only when a recessive gene is inherited from both parents. Example: Spinal muscular atrophy.

X-linked inheritance: Occurs as a result of genetic changes and disorders linked to the X chromosome, usually more common in men. Example: Hemophilia, color blindness.

The origin of hereditary diseases is associated with genetic changes that have been preserved during evolution.

Development of hereditary diseases. Hereditary diseases begin with changes in the genetic structure of the organism and develop under the influence of external and internal factors. The

development of the disease often depends on genetic mechanisms, environmental conditions and epigenetic changes that occur throughout life.

Environmental factors: Radiation, toxic substances or environmental stress, etc., malnutrition (for example, folic acid deficiency during pregnancy can cause fetal defects).

Phenylketonuria:

1. At birth, the child appears healthy, but the amino acid phenylalanine accumulates in the body and causes brain damage.
2. Initial symptoms: irritability, convulsions (muscle spasms).
3. Failure to follow a diet on time, developmental delay and mental retardation occur.

Down syndrome:

1. The development of the disease is caused by an extra copy of a chromosome.
2. Mental and physical abnormalities are manifested during the developmental stage.

Hemophilia:

1. The development of this disease is caused by a genetic defect in the blood clotting process.
2. Injuries increase development and reduce the quality of life.



Types and examples of hereditary diseases. Hereditary diseases are based on various genetic mechanisms and can be divided into three main categories: genetic mutations, chromosomal changes, and diseases associated with mitochondrial inheritance.

They are caused by changes in the structure or number of chromosomes.

Numerical anomalies: Changes in the number of chromosomes.

Examples:

1. Down syndrome: An extra copy of chromosome 21 (trisomy).
2. Turner syndrome: The presence of only one X chromosome (45, X).

Changes in the structure of a chromosome.

1. Leukemias resulting from large chromosome translocations.
2. Neuronal diseases resulting from the duplication of certain chromosome segments.

These diseases are associated with a combination of several genes. Often, the external environment also plays a major role.

For example:

Diabetes mellitus (type 2): Caused by genetic predisposition and unhealthy lifestyle.

Cardiovascular diseases: A combination of genetic and environmental factors.

Cancer: Develops as a result of mutations in certain genes.

Diagnostics and treatment methods. Hereditary diseases are of great importance in modern medicine, and their development is carried out in several directions. Brief information about the main directions:

Diagnostic methods:

Genetic tests:

DNA analysis (to identify mutations).

Genome sequencing (study of the entire genome).

Biochemical tests:

Determination of enzyme activity.

Measurement of metabolite levels in the blood or urine.

Treatment methods:

Pharmacogenetic therapy: Personalized drug selection (according to genetic characteristics).

Gene therapy: Correction of defective genes or changing their activity.

Gene editing using CRISPR-Cas9 technology.

Enzyme replacement therapy: Artificially introducing deficient enzymes into the body.

Cell therapy: Transplantation of healthy cells.

Treatment using induced pluripotent stem cells (iPSC).

Nutritional therapy: Special diets (for example, a phenylalanine-free diet in phenylketonuria).

The origin and development of hereditary diseases are the result of complex interactions between human genetics and environmental factors. Genetic problems can be inherited from one generation to the next or arise as a result of new mutations. Environmental conditions, lifestyle and external factors also play an important role in the development of these diseases. Diagnostic tools and genetic counseling services aimed at early detection and prevention of hereditary

diseases are becoming one of the main directions of modern medicine. At the same time, gene therapy and other innovative treatment methods may help reduce the burden of these diseases on the social and healthcare system in the future. Therefore, in-depth study of the mechanisms of hereditary diseases and work to ensure a healthy generation is an urgent task of medicine today.

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