

GENETIC MUTATIONS AND THEIR CONNECTION WITH HEREDITARY DISEASES

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Abstract: This article analyzes the types of genetic mutations, their causes, and the hereditary diseases that develop as a result of such mutations. The effects of mutations on key biological processes in the human body, the anatomical changes they cause, and the mechanisms by which they are passed from one generation to the next are scientifically explored.

Keywords: genetics, mutation, DNA, chromosome, hereditary disease, autosome, dominant, recessive, anatomical disorder

General Understanding of Genetic Mutations. Genetic mutations are changes that occur in the genetic material of DNA. Every human cell contains a set of genes that control development, structure, metabolism, and many other essential functions. Any alteration, especially inherited mutations, can lead to deviations from normal health. This article thoroughly explores the mechanisms by which mutations lead to hereditary diseases.

Types of Mutations. Mutations are changes that occur in hereditary material and can be classified into three main types:

1. **Point Mutations**
These involve the alteration or misplacement of a single nucleotide. For example, in Sickle Cell Anemia, a change in a single nucleotide leads to an abnormally shaped hemoglobin molecule.
2. **Chromosomal Mutations**
These involve structural changes in chromosomes (such as deletions, duplications, inversions, and translocations). For instance, Cri-du-chat syndrome results from a deletion in chromosome 5.
3. **Genomic Mutations**
These include changes in the number of chromosomes (aneuploidy). Down syndrome is an example, caused by the trisomy of chromosome 21.

Causes of Mutations
Mutations can arise due to the following factors:

- Natural Causes: Errors during DNA replication in cell division.
- External Factors: Ultraviolet radiation, ionizing radiation, chemical substances (mutagens), and viruses.
- Inherited Factors: Genetic changes passed down from parents.

Relationship Between Hereditary Diseases and Mutations
Below are examples of hereditary diseases caused by mutations:

1. **Phenylketonuria**
Caused by a mutation in the gene encoding the enzyme phenylalanine hydroxylase, which disrupts the breakdown of the amino acid phenylalanine and severely affects the nervous system.
2. **Hemophilia**
A recessive X-linked disorder resulting from a deficiency in proteins involved in blood clotting.

3. Cystic Fibrosis
Caused by a mutation in the CFTR gene, leading to malfunctioning mucus glands. The thick mucus accumulates in the respiratory tract and leads to frequent infections.

4. Down Syndrome
Arises due to an extra copy of chromosome 21 (trisomy). Individuals with this syndrome typically have distinctive facial features and developmental delays.

Anatomical Effects of Mutations. Genetic mutations not only occur at the molecular level but also significantly affect anatomical structures:
- Cardiovascular System: In Marfan syndrome, elongation of heart valves and dilation of the aorta are observed.

- Skeletal System: Achondroplasia leads to short stature and abnormal bone growth.

- Nervous System: Tay-Sachs disease results in the destruction of nerve cells.

- Visual and Auditory Systems: Conditions like retinitis pigmentosa and hereditary deafness are linked to genetic mutations.

Mechanisms of Hereditary Transmission of Genetic Diseases.
The inheritance patterns of genetic disorders include:

- Autosomal Dominant: The disease develops with only one mutated gene (e.g., Huntington's disease).

- Autosomal Recessive: Both parents must pass on the mutated gene (e.g., thalassemia).

- X-linked Recessive: More commonly affects males (e.g., hemophilia).

- Mitochondrial Inheritance: Passed only from the mother through mitochondrial DNA.

Modern Diagnostic and Treatment Approaches
Today, genetic mutations can be detected using the following methods:

- Genetic Screening: DNA testing during pregnancy or postnatally to assess the risk of genetic diseases.

- Prenatal Diagnosis: Identification of genetic abnormalities through amniocentesis and chorionic villus sampling.

- Gene Therapy: Includes techniques for correcting faulty genes or introducing healthy ones.

Conclusion

Genetic mutations directly impact human health. The hereditary diseases caused by these mutations not only reduce quality of life but may also shorten life expectancy. Therefore, genetic testing, counseling, and preventive measures play a vital role in modern medicine. Expanding genetic knowledge is crucial in shaping a healthy future generation.

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