

Genetic Mutations: From Inheritance to Impact in Humans

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DESCRIPTION

Genetic mutations are changes in the Deoxyribonucleic Acid (DNA) sequence that can affect the normal functioning of genes. They play a fundamental role in evolution, but they can also have significant impacts on human health. Some mutations are harmless and even beneficial, while others can lead to genetic disorders or contribute to the development of diseases like cancer. Insight into the origins, types and effects of genetic mutations in humans is crucial for advancing medical science and improving the treatment of genetic conditions.

A genetic mutation occurs when there is a change in the sequence of nucleotides in a gene. DNA is made up of four types of nucleotide bases: Adenine (A), Thymine (T), Cytosine (C) and Guanine (G). These bases are arranged in specific sequences to form genes, which provide the instructions for building proteins that perform essential functions in the body. When a mutation occurs, it can alter the way a gene is expressed or the protein it encodes, which may disrupt normal biological processes.

Genetic mutations can occur for several reasons. They may arise from copying errors during cell division, exposure to environmental factors like radiation or chemicals or can even be passed down from one generation to the next. Mutations that are inherited from a parent are called germline mutations, while those that occur in somatic cells (cells that are not sperm or egg cells) during a person's lifetime are called somatic mutations.

Types of genetic mutations

Point mutations: A point mutation occurs when a single nucleotide is altered. This can cause a shift in the amino acid sequence of a protein, potentially affecting its functionality. For example, a point mutation can cause sickle cell anemia, where the substitution of adenine for thymine leads to the production of an abnormal hemoglobin protein.

Insertions and deletions: These mutations occur when one or more nucleotides are either inserted or deleted from the gene sequence. Insertions and deletions can cause frameshift

mutations, which shift the reading frame of the genetic code. This can result in a completely altered protein, often leading to severe consequences. For instance, cystic fibrosis is often caused by a deletion of three nucleotides in the *CFTR* gene, leading to the production of a defective protein.

Duplication: In this mutation, a segment of the DNA is duplicated, leading to multiple copies of a particular gene. This can disrupt normal gene function and contribute to disorders such as Charcot-Marie-Tooth disease.

Chromosomal mutations: These mutations involve large-scale changes in chromosome structure. These can include translocations (where part of one chromosome breaks off and attaches to another), inversions (where a segment of a chromosome is reversed) or duplications.

Impact of genetic mutations on human health

Genetic disorders: Some mutations are linked to hereditary genetic disorders. These disorders can range from mild to severe and they often follow specific inheritance patterns, such as autosomal dominant, autosomal recessive or X-linked inheritance. Examples of genetic disorders caused by mutations include.

Cystic fibrosis: A genetic disorder caused by a mutation in the *CFTR* gene, leading to problems with mucus production in the lungs and other organs.

Huntington's disease: A neurodegenerative disorder caused by a mutation in the *HTT* gene, leading to progressive brain damage and loss of motor and cognitive function.

Duchenne muscular dystrophy: An X-linked recessive disorder caused by mutations in the dystrophin gene, leading to muscle weakness and degeneration.

Cancer: Somatic mutations, which occur in non-reproductive cells, are a major contributor to cancer. Mutations in key genes that regulate cell division, such as the *TP53* gene (which encodes a tumor-suppressing protein), can lead to the development of

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cancer. Cancer-causing mutations can result from exposure to environmental factors like tobacco smoke, ultraviolet radiation or certain chemicals.

Evolutionary significance: Mutations also play a critical role in evolution. While most mutations are neutral or harmful, a small fraction may provide a survival advantage, contributing to the natural selection process. For example, the mutation that causes lactose tolerance in some human populations allowed individuals to digest milk into adulthood, which may have been beneficial in pastoral societies.

CONCLUSION

Genetic mutations are an integral part of life, influencing human health, development and evolution. They can lead to

genetic disorders, contribute to diseases like cancer or even provide beneficial traits that are passed down through generations. The mechanisms behind genetic mutations and their effects on the body is essential for advancing medical study and developing treatments for genetic diseases. While mutations can cause significant challenges, they are also the driving force behind biological diversity and evolution. As our knowledge of genetics continues to grow, we gain importance of how these mutations shape our lives and health, opening new possibilities for diagnosis, treatment and prevention.