INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCHERS

www.wordlyknowledge.uz Volume: 3, Issue: 2, 2024

e ISSN : 3030-332X IMPACT FACTOR ( Research bib ) – 7,293

# MUTATIONS: DEFINITION, CAUSES, TYPES, BENEFITS AND DISADVANTAGES OF MUTATIONS

**Ortikova Mohira Izbosarovna**
**Absamatova Madina**
Jizzakh State Pedagogical Unversity

**Abstract:**Mutations play a crucial role in driving evolution and were previously thought to occur randomly, gradually, and uniformly throughout genomes, independent of environmental factors. However, recent discoveries of molecular mechanisms of mutation in bacteria have challenged this perspective. These mechanisms, which have been observed across various organisms, indicate that mutagenesis is highly regulated. It is up-regulated during times of stress and activated when cells or organisms are poorly adapted to their environments, potentially speeding up the process of adaptation. Mutation is defined as a modification in the amount and structure of DNA within a cell or organism, resulting in a change in the genotype. Importantly, this altered genotype can be inherited by future generations of cells through processes such as mitosis or meiosis.

**Keywords:**Mutation, DNA, cell, evolution, genotype, environment, human.

**Introduction:**In biology, a mutation is an alteration in the nucleic acid sequence of the genome of an organism, virus, or extrachromosomal DNA. Mutations are changes in the genetic information of an organism that can occur as a result of errors in the DNA replication process or under the influence of external factors. Viral genomes contain either DNA or RNA. Mutations result from errors during DNA or viral replication, mitosis, meiosis, or other types of damage to DNA (such as pyrimidine dimers caused by exposure to ultraviolet radiation), which then may undergo error-prone repair (especially microhomology-mediated end joining), causing an error during other forms of repair, or cause an error during replication (translesion synthesis). Mutations may also result from the insertion or deletion of segments of DNA due to mobile genetic elements.

Mutations can occur for various reasons. Some of them are natural processes that occur in the body, while others can be caused by external factors. Here are some of the main causes of mutations:

Random errors in DNA replication

During DNA replication, when a cell divides, a random error may occur, which leads to a change in the sequence of nucleotides in the gene. This may be due to an error in DNA polymerase, the enzyme responsible for copying DNA. Such errors may be rare, but they can still occur and lead to mutations.

Exposure to mutagens

Mutagens are substances or factors that can damage DNA and cause mutations. They can be chemicals such as radioactive substances, toxic chemical compounds or carcinogens, as well as physical factors such as ultraviolet radiation or X-rays. Exposure to mutagens can lead to changes in the nucleotide sequence or DNA structure, which in turn can cause mutations.

Genetic factors

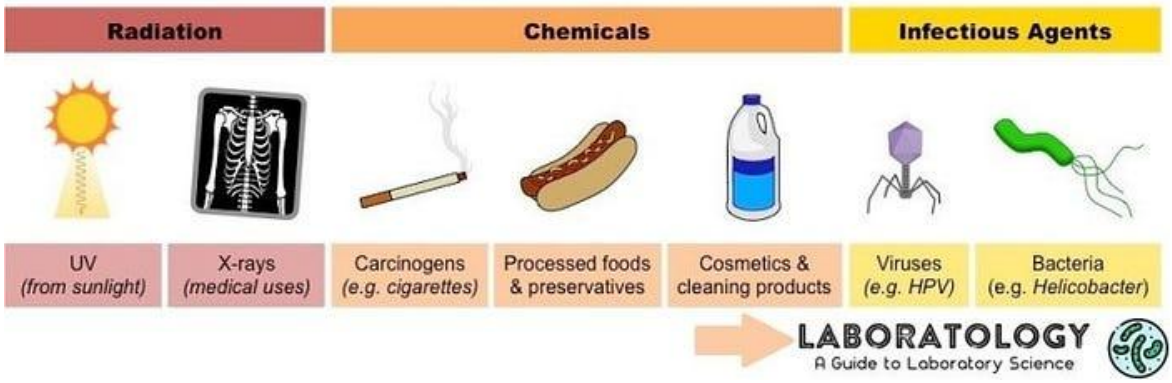
Some mutations can be inherited from parents. This may be due to the presence of genetic variations or mutations in gametes (sperm or eggs) that are passed on to offspring. For example, mutations in genes responsible for DNA repair or control of cell division can be inherited and lead to an increased risk of developing genetic diseases.

www.wordlyknowledge.uz Volume: 3, Issue: 2, 2024

e ISSN : 3030-332X IMPACT FACTOR ( Research bib ) – 7,293

External factors

Some mutations may be caused by external factors such as environmental pollution, exposure to chemicals or drugs, poor nutrition, or exposure to infections. These factors can damage DNA and cause mutations in the body's genome.




Examples of Gene Mutations:

The earliest documented instance of gene mutations dates back to 1791 when Seth Wright noticed a lamb with abnormally short legs in his flock of sheep. This particular sheep, known as an Ancon sheep, was unable to jump over low stone fences and cause damage to nearby crops. Wright saw potential value in having an entire flock of these short-legged sheep for this reason.

Over subsequent generations, this characteristic was passed down, and a line of sheep with short legs was developed. This trait resulted from a recessive mutation, and the short-legged individuals were homozygous recessive. This gene mutation was discovered at a time when the field of genetics was still in its infancy.

The scientific study of mutations began in 1910 when T.H. Morgan conducted research on fruit flies, specifically Drosophila melanogaster. He observed male flies with white eyes among the normal red-eyed females. It was later discovered that the gene responsible for this trait was located on a sex chromosome.

Another example of a gene mutation is the human blood disease sickle cell anemia. This condition is caused by abnormal hemoglobin S, which is unable to effectively carry oxygen. It has been observed that the abnormal hemoglobin differs from the normal hemoglobin by only two amino acids in its polypeptide chains (hemoglobin 2 alpha and 2 beta chains). Specifically, it contains valine instead of glutamic acid at the sixth position. This minor alteration, involving two nucleotides in the DNA, leads to a substitution in the amino acid sequence and a significant change in the synthesis of normal hemoglobin. Various other human diseases, such as thalassemia, phenylketonuria, and alcaptonuria, are caused by simple base substitutions in the nucleotides that disrupt the production of normal proteins. Although gene mutations may result in minor changes in base pairing, their impact on organisms carrying such mutant genes is often significant.

Generally, mutations are considered harmful or deleterious and may not produce visible effects. Less than 20% of mutations are lethal. When mutant genes are present in a homozygous condition, they can cause the death of the organism. Mutant genes are typically recessive to normal genes and only manifest their effects in the homozygous state, which can go undetected for a period of time. This implies that the mutation rate is actually much higher than the frequency of visible or detectable mutations.

Consequences of Mutations:

www.wordlyknowledge.uz Volume: 3, Issue: 2, 2024

e ISSN : 3030-332X IMPACT FACTOR ( Research bib ) – 7,293

Mutations can have various effects on the body depending on which genes and sections of DNA are affected. Here are some of the main consequences of mutations:

Alteration of Protein Structure: Mutations in genes can lead to changes in the amino acid sequence of a protein, which can affect its structure and function. This can disrupt normal bodily functions and contribute to the development of various diseases.

Changes in Gene Regulation: Mutations can also impact gene regulation, determining which genes are activated or suppressed under specific conditions. This can result in improper gene expression, leading to abnormal development and functioning of the body.

Emergence of New Traits: Some mutations can give rise to new properties or functions in the body. For example, a mutation in the gene responsible for skin color can lead to the appearance of a new pigment and a change in skin color.

Influence on Survival Rate: Mutations can affect an organism's survival under specific conditions. Certain mutations may enhance survival by providing protection against infections or enabling adaptation to new environments. Conversely, other mutations may reduce survival by causing genetic diseases or disrupting normal organ function.

Influence on Evolution: Mutations serve as the foundation for the evolution of organisms by creating genetic diversity that can be subject to natural selection. Some mutations may confer advantages to an organism, enhancing its survival and reproductive success. On the other hand, disadvantageous mutations can lead to decreased survival. Therefore, mutations play a crucial role in the evolution of organisms and the formation of new species.

| **Type of Substitution Mutation** | **Description** |
|---|---|
| **Missense Mutation** | A substitution that changes the amino acid encoded by the codon, resulting in a potentially altered protein. |
| **Silent Mutation** | A substitution that does not change the amino acid encoded by the codon, resulting in no change in the protein. |
| **Nonsense Mutation** | A substitution that introduces a premature stop codon, leading to the production of a truncated or nonfunctional protein. |
| **Transition Mutation** | A substitution that involves the exchange of a purine for another purine or a pyrimidine for another pyrimidine. |

www.wordlyknowledge.uz Volume: 3, Issue: 2, 2024

e ISSN : 3030-332X IMPACT FACTOR ( Research bib ) – 7,293

| | |
|---|---|
| **Transversion Mutation** | A substitution that involves the exchange of a purine for a pyrimidine or vice versa. |

In the field of genetics, it is crucial to have a comprehensive understanding of the different types of mutations. Genetic mutations can manifest in various forms, including substitutions, insertions, deletions, and large-scale mutations. These mutations have a significant impact on the development of inherited diseases and contribute to genetic diversity within populations.

Mutations can occur due to multiple factors, such as errors during DNA replication and recombination, chemical damage to DNA, and exposure to radiation. These factors can result in alterations to the DNA sequence, potentially affecting the function of genes and proteins.

Fortunately, cells have evolved intricate mechanisms to repair DNA and minimize the accumulation of mutations, thereby maintaining the integrity of the genome. Mismatch repair, nucleotide excision repair, direct repair, and recombination repair are some of the mechanisms employed by cells to rectify damaged DNA. These processes ensure that errors in DNA are corrected, preserving the accuracy of genetic information.

Through the study of different types of mutations, their underlying causes, and the mechanisms involved in DNA repair, scientists can gain valuable insights into genetic disorders and advance our understanding of biology. This knowledge is crucial for the development of novel therapies and treatments for inherited diseases, as well as for improving our ability to diagnose and prevent genetic conditions.

**References:**

**1.https://www.nature.com/scitable/topicpage/genetic-mutation-1127/ Genetic Mutation**

2. https://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1007995

3. https://metrics-lab.github.io/2020/05/24/mutations-and-polymorphisms.html Genetics Part III: Mutations & Polymorphisms

4. https://www.geeksforgeeks.org/mutation/ Mutation