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Genetic mutations: causes and consequences

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Abstract

Gene mutation is an alteration in the sequence of nucleotides in DNA, which may affect a single nucleotide pair or a large gene segment of a chromosome. Mutations can be neutral, beneficial, or harmful (deleterious). A harmful mutation decreases the overall fitness of an organism, by impairing a protein's capacity to carry out its physiological function. Identification of these undesirable genes is crucial as they are responsible for affecting an individual's vitality and can pass on from one generation to the next generation.

Introduction

Any sudden change occurring in hereditary material is called a 'mutation'. Particularly, a gene mutation is defined as an alteration in the sequence of nucleotides in DNA. This change can affect a single nucleotide pair or a large gene segment of a chromosome. DNA-based single-gene mutations occur at the rate of one percent of live birth, while chromosome-based mutations occur at 0.6 percent of live birth. The mutation rate per generation is $2.5\times10–^{8}$ per base in the genome, while in mtDNA, it is 2.7×10^{-5} per base. Mutation in germ cells mostly occurs de novo during copying DNA, generally at the rate of 10^{-5} to 10^{-8} mutations per cell division. The mutation rate in sperm is higher than in eggs. An average of 2.9 new mutations occurs per year of the father's life. Each child on average inherits 76 new mutations from their parents.

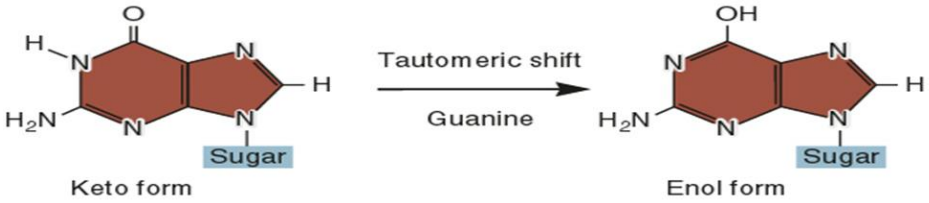
History

In the year 1900, Hugo de Vries first used the term mutation to describe the sudden heritable phenotypic changes in evening primrose *Oenothera lamarckiana*. Later, in 1904, T.H. Morgan reported white-eyed drosophila in the population of red-eyed flies. In 1905, Lucien Cuénot first discovered lethal alleles while studying the inheritance of coat color in mice, and H.J. Muller was the first to use X-rays to induce mutations in fruit flies in 1928.

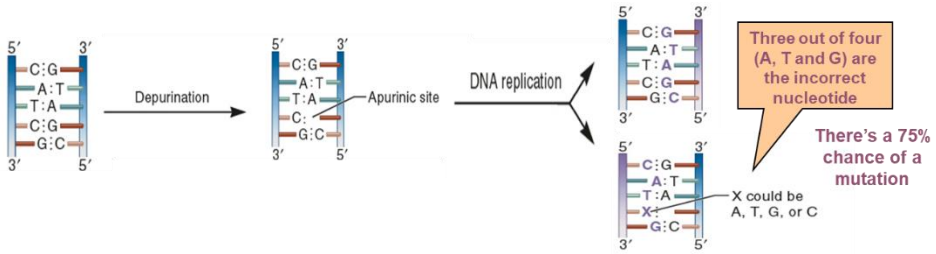
Molecular mechanism of mutation

1. **Spontaneous mutation** – These are the mutations that arise naturally and not through the action of the mutagenic agent. Spontaneous mutations may arise through errors in DNA replication/cell growth/cell division or spontaneous alteration of a nucleotide within an existing DNA molecule.

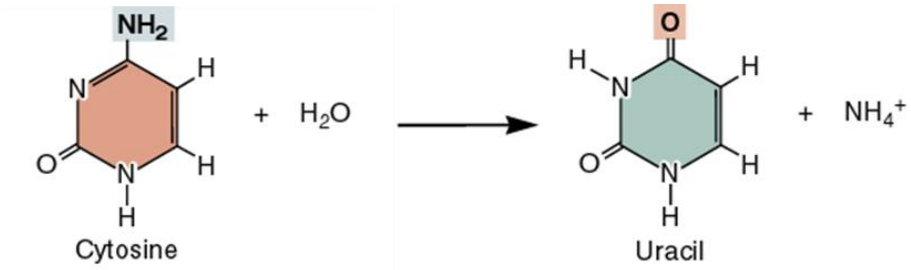
- **Tautomerism-** A base is changed by the repositioning of a hydrogen atom.



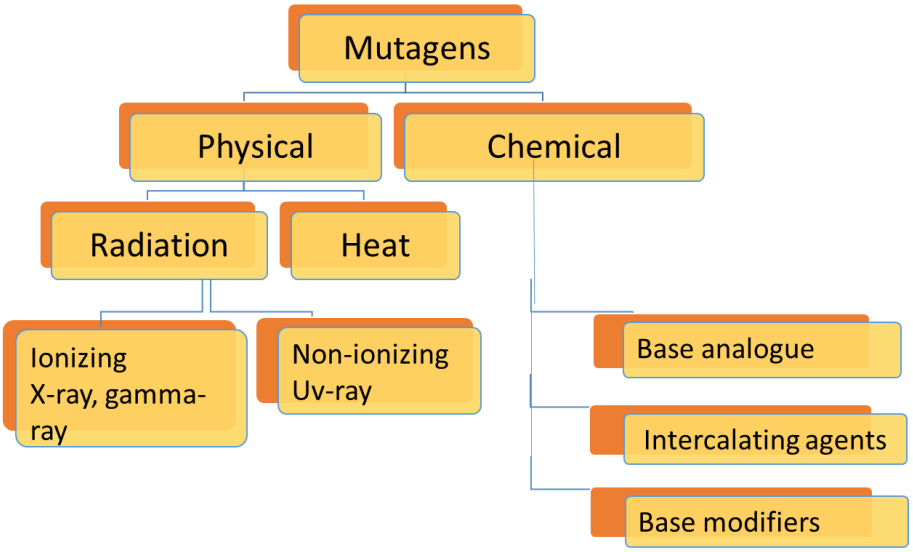
- **Depurination**- It involves the removal of a purine (guanine or adenine) from the DNA.



- **Deamination**- It involves the removal of an amino group from the cytosine base.



2. **Induced mutation –** During its lifetime, an organism may be exposed to variety of agents capable of causing damage to its genetic material.



Effects of gene mutation

- **Beneficial mutations -** Mutations that enhance survival and reproductive success are known as beneficial mutations. They are essential for evolution and they turn off harmful genes.

  Examples - antibiotic resistance bacteria superbugs such as MRSA.

- **Neutral mutations –** Neutral mutations are neither harmful nor beneficial to the organism

  Ex. Eye color, birthmarks

- **Harmful mutations** - These mutations are seldom lethal to life and are responsible for genetic disorders and dysfunctional proteins.

  Ex. Cystic fibrosis, sickle cell anemia, Albinism

The conditional lethal mutants may be able to live under certain permissive conditions. The most common type of conditional mutant is the temperature-sensitive mutant, which is able to live at the temperature of 35°C but not at the restrictive temperature (39°C). Mutations do not occur randomly throughout the genome. Some regions are more prone to mutate than others (hotspots of mutation). One such hotspot is the dinucleotide 5'-CG-3' (also called CPG) in which cytosine is frequently methylated in many animal genomes, changing it to 5'-TG'-3'

Diagnosis of genetic mutations

- Family history
- Clinical observations
- Hematological/Biochemical analysis
- Determining enzyme/protein activity
- DNA based testing

- Chromosomal analysis

Conclusion

Undetected genetic mutations get propagated from generation to generation, which increases the occurrence of undesirable genes in the population, thus negatively affecting the vitality of the individual.

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