MUTATION

Mutations: Any change in the structure of a gene and they consider the fundamental sources of evolution, there two types of mutation:

1- Chromosomal mutation : it is change in structure or number of Chromosomes . mentioned previously

2- Genetic (point) mutation: it is a change in a single nucleotide or a few number of nucleotides in DNA of a gene . This type of mutation is usually less serious than a chromosomal alteration

point mutation according to the change in DNA :

1- Base pair substitutions : a single base is altered **CCC → CTC**

2- Insertions: an extra base is incorporated **CCC → CCGC**

3- Deletions: a single base is deleted C**CC → CC**

Point Mutations and Their Effects on the level of protein

Type	Description	Example	Effect
Silent	mutated codon codes for the same amino acid	CAA(glutamine)→CAG(glutamine)	none
Missense	mutated codon codes for a different amino acid	CAA (glutamine) →CCA (proline)	variable
Nonsense	mutated codon is a premature stop codon	CAA (glutamine) → UAA (stop)	usually serious

Direction of the mutations :

1. Forward mutations : are changes away from the wild type
2. Reverse mutations : (reversions) are changes from the mutant back to the wild type .

Mutation according to the cell type:

1. Somatic mutations :

a) mutations that are in the somatic tissues of the body

b) mutations are not transmitted to progeny

2.Germinal mutations

a) mutations that are in the germ tissues of the body

b) mutations may be transmitted to progeny

Phenotypic effects of mutation :

1. Morphological mutations are mutations that affect the visible properties of an organism .
2. Lethal mutations are mutations that affect the viability of the organism
3. Conditional mutations are mutations in which the mutant allele causes the mutant phenotype only in certain environments
4. Resistant mutation make the organism resistant to antibiotics
5. Biochemical mutations are mutations that affect the metabolic pathway .

According to the Origin of mutation :

1- Spontaneous mutation : happens without any outside influence its frequency is very low 10^{-7} in generation , occurred because Errors in DNA replication or Errors in DNA repair .

2- Induced mutation : Induced by exposure to a variety of mutagens which increase mutation frequency . Mutagens may be biological like viruses , chemical mutagens like cigarette smoke and preservatives , physical mutagens like UV and X-rays

Beneficial Mutations

Some mutations have a positive effect on the organism in which they occur, They lead to new versions of proteins that help organisms adapt to changes in their environment , like Mutations in many bacteria that allow them to survive in the presence of antibiotic .

Harmful Mutations

The change in a gene's DNA which result in a protein that does not function normally or may not function at all. Such mutations are likely to be harmful they may cause genetic disorders like hemophilia there is a mutation in the Factor VIII gene(Factor VIII is a protein for blood clotting) resulting in uncontrolled bleeding . or cause cancer disease in which cells grow out of control and form abnormal masses of cells, It is caused by mutations in genes that regulate the cell cycle.

DNA repair :

Many mutations have no effect on the organism because they are repaired before protein synthesis occurs . Cells have multiple mechanisms to repair mutations in DNA .The importance of effective DNA repairs is highlighted by the approximately 130 human genes participating in DNA repair system.