**Alterations in the genetic material**

Although DNA replication is virtually error-free, mistakes do occasionally occur. An error in replication will result in a daughter molecule that contains a **mismatch,** a position at which a base pair will not form as the nucleotides opposite each other in the double helix are not complementary. When the mismatched daughter molecule is itself replicated the two granddaughter molecules will not be identical: one will have the correct nucleotide sequence, but the second will contain a **mutation.** A mutation is simply an alteration in the nucleotide sequence of a DNA molecule.

Mutations are also caused by chemical and physical **mutagens.** Several chemicals react with DNA molecules and change the structure of one or more nucleotides within the double helix. Heat, ultraviolet radiation and other physical mutagens have similar effects. Changing the structure of a nucleotide may affect its base pairing properties, resulting in a mutation when the DNA molecule is replicated.

As we shall see, mutations may be harmless, or they may have a serious, even lethal, effect on the organism. To minimize the number of mutations that become established, all organisms have **DNA repair** mechanisms, with which they attempt to correct mismatches and structural alterations in their DNA molecules. Despite these measures a few mutations slip through.

A mutation is a small-scale change in a DNA molecule. Larger-scale alterations occur by the process called **recombination.** Recombination can have any one of several effects, including the exchange of segments of polynucleotides between different DNA molecules, and the **transposition** of a piece of DNA from one position to another in a molecule. Far from being harmful, recombination plays several important roles in the life of a cell.

**Mutations**

As already stated, a mutation is a change in the nucleotide sequence of a DNA molecule. A mutation may occur within a gene or it may occur in the intergenic regions. If a mutation occurs in an intergenic region it will probably be **silent** and have no discernible effect on the cell. However, if amutation occurs in a gene it may alter the gene product and generate an observable change in the organism: this is referred to as a change in **phenotype.** The phenotype of an organism is its set of observable characteristics; in contrast the **genotype** is its genetic constitution. An organism displaying the usual phenotype for that species is called a **wild-type;** an organism whose phenotype has been altered by mutation is called a **mutant.**

**Different types of mutation**

Mutations can be looked at from three different angles. First, we can consider just the DNA sequence itself and list the different types of nucleotide sequence alterations that can occur. Second, there is the question of what effect the different types of mutation will have if they occur in a gene. And finally, there is the point of view of the organism and the different kinds of phenotypic change that can arise as a result of mutation.

**Mutations at the level of the DNA sequence.** The various types of alteration in DNA sequence that can occur as a result of mutation are as follows:

A **point mutation** is the replacement of one nucleotide by another. A point mutation is classified as a **transition** if it is a purine to purine (A - G) or a pyrimidine to pyrimidine (T -C) change, or a **transversion** if the alteration is purine to pyrimidine or vice versa (A or G - T or C).

**An insertion** or **deletion** is the addition or removal of anything, from one base pair to quite extensive pieces of DNA.

An **inversion** is the excision of a portion of the double helix followed by its reinsertion at the same position but in the reverse orientation.

**A: Find a suitable synonym in the text for the words below.**

immaculate (1st prg)

copied

exactly the same

*a* change

results (2nd prg)

to influence

mortal, deadly (3rd prg)

to lessen

permanent

limited in extent (4th prg)

parts

not at all

distinct (5th prg)

to produce, to bring about/on

composition

aspect, viewpoint (6th prg)

to emerge

due to

listed as (7th prg)

large, long

extraction, removal

**B: Complete the following sentences with a suitable word or phrase.**

1. Transposition is the of a genetic from one to

another in a DNA molecule.

1. An alteration in a DNA…………………….. that does not the expression or functioning of any gene or gene product is called a mutation.
2. A mutation that a single nucleotide alteration in a DNA molecule is called a point mutation.
3. A base pair is the hydrogen- structure formed between two

nucleotides.

1. Recombination is the………………………….. of segments of polynucleotides between two DNA
2. A gene, cell or organism which the typical phenotype or for the species and is therefore as a standard, is called a wildtype.

**C: Mark True or False next to each statement.**

1. Mistakes hardly ever occur during DNA replication, but when they do, they are always lethal.
2. A mutation is a change in the nucleotide sequence of a DNA molecule.
3. An inversion is the removal of a base pair off the double helix.
4. genotype is the word that refers to the genetic constitution of an organism.
5. Only phenotypic changes arise as a result of mutations.
6. A wildtype is an organism whose phenotype has been altered due to the effect of one or more mutagens.
7. The factors that may cause alterations in the DNA sequence are called mutagens and are purely chemical agents.
8. The occurrence of a mutation is irreversible.
9. All organisms have DNA repair mechanisms.
10. If a mutation occurs in an intergenic region it has no observable effects on the cell and is called a point mutation.