Gene mutation

Introduction :

A change in the sequence of bases in DNA or RNA is called a mutation. In fact, most people have dozens or even hundreds of mutations in their DNA. Mutations are essential for evolution to occur. They are the ultimate source of all new genetic material—new alleles in a species. Although most mutations have no effect on the organisms in which they occur, some mutations are beneficial. Even harmful mutations rarely cause drastic changes in organisms.

Causes of Mutation

Mutations have many possible causes. Some mutations seem to happen spontaneously without any outside influence. They occur when mistakes are made during DNA replication or transcription. Other mutations are caused by environmental factors. Anything in the environment that can cause a mutation is known as a mutagen.

Examples of Mutagens. Types of mutagens include radiation, chemicals, and infectious agents

Types of Mutations :

There are a variety of types of mutations. Two major categories of mutations are germline mutations and somatic mutations. • Germline mutations :

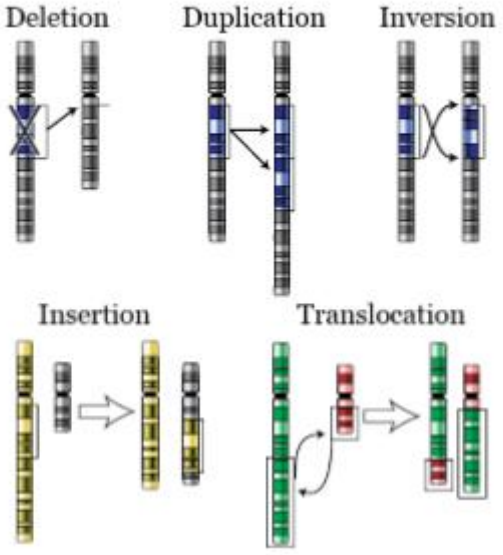
occur in gametes. These mutations are especially significant because they can be transmitted to offspring and every cell in the offspring will have the mutation.

• Somatic mutations:

occur in other cells of the body. These mutations may have little effect on the organism because they are confined to just one cell and its daughter cells. Somatic mutations cannot be passed on to offspring.

Mutations also differ in the way that the genetic material is changed. Mutations may change the structure of a chromosome or just change a single nucleotide.

Chromosomal Alterations Chromosomal alterations are mutations that change chromosome structure. They occur when a section of a chromosome breaks off and rejoins incorrectly or does not rejoin at all. Possible ways these mutations can occur are illustrated in Figure 1



Chromosomal alterations are very serious. They often result in the death of the organism in which they occur. If the organism survives, it may be affected in multiple ways. An example of a human chromosomal alteration is the mutation that causes Down Syndrome. It is a duplication mutation that leads to developmental delays and other abnormalities.

Point Mutations A point mutation is a change in a single nucleotide in DNA. This type of mutation is usually less serious than a chromosomal alteration. An example of a point mutation is a mutation that changes the codon UUU to the codon UCU. Point mutations can be silent, missense, or nonsense mutations, as shown in Table 1. The effects of point mutations depend on how they change the genetic code

Table 7.5: Point Mutations and Their Effects

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

Frameshift Mutations A frameshift mutation is a deletion or insertion of one or more nucleotides that changes the reading frame of the base sequence. Deletions remove nucleotides, and insertions add nucleotides. Consider the following sequence of bases in RNA: AUG-AAU-ACG-GCU = start-asparagine-threoninealanine Now, assume an insertion occurs in this sequence. Let's say an A nucleotide is inserted after the start codon AUG: AUG-AAA-UAC-GGC-U = start-lysine-tyrosine-glycine

Even though the rest of the sequence is unchanged, this insertion changes the reading frame and thus all of the codons that follow it. As this example shows, a frameshift mutation can dramatically change how the codons in mRNA are read. This can have a drastic effect on the protein product.

Spontaneous Mutations There are five common types of spontaneous mutations. These are described in the Table 2

Table 7.6: Spontaneous Mutations Described

Mutation	Description
Tautomerism	a base is changed by the repositioning of a hydrogen atom
Depurination	loss of a purine base (A or G)
Deamination	spontaneous deamination of 5-methycytosine
Transition	a purine to purine (A to G, G to A), or a pyrimidine to pyrimidine (C to T, T to C) change
Transversion	a purine becomes a pyrimidine, or vice versa

Effects of Mutations :

The majority of mutations have neither negative nor positive effects on the organism in which they occur. These mutations are called neutral mutations. Examples include silent point mutations. They are neutral because they do not change the amino acids in the proteins they encode. Many other mutations have no effect on the organism because they are repaired before protein synthesis occurs. Cells have multiple repair mechanisms to fix mutations in DNA.