Base pair Substitution :-

A base pair substitution mutation involves a change in the DNA such that one base pair is replaced by another.

There are two general types of base-pair substitution mutations. There are

i) Transition mutation
ii) Transversion mutation

i) Transition mutation :-

Transition mutation is a mutation from one purine-pyrimidine base pair to the other purine-pyrimidine base pair. The four types of transition mutations are AT to GC, GC to AT, TA to CG, CG to TA.

ii) Transversion mutation :-

Transversion mutation is a mutation from a purine-pyrimidine base pair to a pyrimidine purine base pair. The eight types of transversion mutations are AT to TA, TA to AT, GC to CG, CG to GC, AT to CG, CG to AT, GC to TA & CG to AT.

There are other types of mutations given below.

a) Missense mutation
b) Nonsense mutation
c) Neutral mutation
d) Silent mutation
e) Frameshift mutation
f) Reverse Mutation
g) Suppressor Mutations

Spontaneous and Induced Mutation :-

Mutagenesis, The creation of mutations, can occur spontaneously or can be induced. Spontaneous mutations are naturally occuring mutations. Induced mutations occur when an organism is exposed either deliberately or accedentally to a physical or chemical agent, known as mutagen, That interact with DNA to cause a mutation. Induced mutations typically occur at a much higher frequency than spontaneous mutations.

Spontaneous Mutations :-

All types of point mutations can occur spontaneously. Spontaneous mutation can occur during DNA replication. In human, spontaneous mutation rate for individual genes varies between 10^{-4} and 4×10^{-6} per gene per generation.

Induced mutation :-

Mutations can be induced by exposing organisms to physical mutagens, such as radiations or chemical mutagens. Induced mutations play an important role in the study of mutations. Since the rate of spontaneous mutations is slow, geneticists use mutagens to increase the frequency of mutations so that a significant number of organisms have mutations in the gene being studied.

* Disorders Caused by Mutation ⇒

The disorders are caused by mutations in genes encoding enzymes required for the catabolism of tyrosine. Tyrosinosis and tyrosinemia result from the lack of enzymes tyrosine transaminase and p-hydroxyphenyl pyruvic acid oxidase, respectively. Both enzymes are required to degrade tyrosine to CO_2 and H_2O.

Albinism, the absence of pigmentation in the skin, hair, and eyes, results from mutational block in the conversion of tyrosine to the dark pigment melanin. One type of albinism is caused by the absence of tyrosinase, the enzyme that catalyses the first step in the synthesis of melanin from tyrosine.